Autism Spectrum Disorders: A Case Study in Causation and Explanation in Psychiatric Conditions

by

Tracy Finn

A thesis
presented to the University of Waterloo
in fulfillment of the
thesis requirement for the degree of
Doctor of Philosophy
in
Philosophy

Waterloo, Ontario, Canada, 2013

I hereby declare that I am the sole author of this thesis. This is a true copy of the thesis, including any required final revisions, as accepted by my examiners.

I understand that my thesis may be made electronically available to the public.

Abstract

This thesis discusses epistemological and ethical issues in classification and diagnosis of psychiatric conditions, and briefly discusses realism about psychiatric conditions. I use autism spectrum disorders (ASDs) as a case study to examine whether the explanatory and predictive power of classification and diagnosis could be improved if psychiatry adopts a cause-based framework in place of a symptom-based framework. However, there is significant debate regarding the sort of explanatory pattern that will adequately represent the complex causation involved in psychiatric conditions. I develop a preliminary list of criteria for adequate explanatory patterns in psychiatry, and use these criteria to analyze explanations of ASDs. I show that explanatory patterns unable to meet these criteria limit the validity and reliability of diagnosis. However, I argue that an integrated pattern that includes biological, cognitive and social levels of explanation may meet the criteria. Thus, diagnosis of ASDs could improve if psychiatry adopted a cause-based framework informed by an intergrated explanation pattern. More accurate diagnosis of ASDs may allow earlier access to Intensive Behaviourial Intervention/Applied Behavioural Analysis treatment programs, which may increase the effectiveness of this treatment and reduce the amount of resources individuals with ASDs require from governments over their lifespans. Explaining these conditions using an intergrated pattern of explanation can further challenge myths regarding the causes of ASDs, and may provide support for Canadian lawsuits petitioning for expanded public funding of IBI/ABA programs.

Acknowledgements

Thank you to Paul Thagard for the continued support, guidance and encouragement. Thank you also to Ian Gold, Daniela K. ONeil, Chris Eliasmith, and Mathieu Doucet for their excellent comments, questions and suggestions on the final version of this thesis. Thank you to my family, who have supported me through this process and have given invaluable encouragement and advice throughout the process of completion. Thank you to W. Jim and Shelly J, Eric H, Angela C, Anna B, Corey M and Mike M for friendship, advice and encouragement. Thank you to S. and B., my constant and loving companions.

Dedication

To Kevin Kakonge, my rock and my shore, for your constant support and love. To Kerry Russell, for always, always being there. To Maria, Bernie, Terri, Dusty, Paul and Sachiyo, for being a second family. To Paul Moakler, for giving something irreplaceable. To S. S., my love, for everything.

Table of Contents

Chapter 1

Philosophical Issues in Psychiatric Diagnosis and Classification

1.1 Introduction

The way mental disorders are explained has a significant influence over the way these conditions are classified, diagnosed and treated. Examining the relationship between patterns of explanation for psychiatric disorders and how these conditions are diagnosed and treated identifies several philosophical issues in psychiatric practice. First, there is significant disagreement over the nature of causal explanations in psychiatry, how best to generate such explanations, and how these explanations should influence the classification and diagnosis of psychiatric disorders. Second, examining the relationship between patterns of explanation for psychiatric conditions, how these conditions are currently classified, and how they are diagnosed reveals some of the difficulties in developing and justifying realist claims about mental disorders. Third, the way in which psychiatric conditions are explained greatly influences how they are treated, how resources are allocated for such treatment, and how these conditions are understood by society at large. In this thesis, I focus primarily on questions regarding the epistemological and ethical issues in psychiatric diagnosis, although I touch on the metaphysical issues as well.

One of the central debates in the philosophy of psychiatry is whether the classification and diagnosis of psychiatric disorders should be based on causal theories, rather than on descriptions of cognitive and behavioural symptoms co-occurring over time. Currently, psychiatric disorders are classified based on differences in characteristic symptoms, and diagnosis of a particular disorder is based on the presence or absence of these characteristic

symptoms. However, many clinicians and theorists argue that diagnosis and classification of psychiatric disorders could have more explanatory and predictive power if based on causal theories from disciplines such as cognitive neuroscience, genetics and epigenetics.

In this thesis, I use autism spectrum disorder as an extended case study to help examine whether the problems in the diagnosis and classification of psychiatric disorders could be improved if psychiatry were to adopt a cause-based diagnostic and classification framework. I argue that basing the classification and diagnosis of autism spectrum disorder on causal theories may improve the explanatory and predictive power of the diagnosis of this condition. However, there are many different explanatory patterns that can be used to explain the causes of psychiatric conditions such as autism spectrum disorder. In chapters three, four, five and six, I examine different explanatory patterns from past and present conceptual frameworks in psychiatry and clinical psychology. I show that some explanatory patterns, such as those based on psychoanalytic or behaviourist conceptual frameworks, limit the explanatory and predictive power of diagnostic categories for conditions such as autism spectrum disorder, which reduces the effectiveness of treatment. Further, while some patterns, such as those based on cognitive, cognitive neurological, genetic and epigenetic causal theories, may be more powerful, I argue that none of these patterns will be robust enough to inform a cause-based diagnostic and classification system on their own. Theorists such as Kendler (2008) and Murphy (2006) state that psychiatric conditions, including ASD, will likely be explained using a multi-level approach incorporating all the fields discussed in this thesis if psychiatry does adopt a cause-based framework. I also argue that the diagnosis and treatment of ASD could be improved if informed by multi-level explanatory patterns identifying causes and how these causes interact to produce the characteristic impairments and symptoms. We can combine the more powerful explanation schemas discussed in this thesis into an integrated, multi-level mechanistic explanation of the causes and symptoms of autism spectrum disorder. When combined in an explanation pattern incorporating the biological, cognitive and social levels of explanation, the more powerful explanation schemas, i.e., cognitive, cognitive neurological, genetic and epigenetic, may help to better identify cases of ASD in the clinical population.

To begin, I discuss the arguments made by critics of the current system such as Poland et al, (1994) and Murphy (2006) who state that the diagnostic and classification framework contained in the *Diagnostic and Statistical Manual* is plagued by incoherence, poor explanatory and predictive power, and false assumptions regarding the nature of psychiatric disorders. To support these arguments, I identify problems with the diagnosis and treatment of autism spectrum disorder to illustrate the shortcomings in the current symptom-based diagnostic and classification framework. Like many of the theorists discussed in this thesis, I argue that the current problems with the diagnosis and classification of mental

disorders could be addressed if diagnostic categories were based on causal theories.

However, there are considerable disagreements among these theorists regarding the scope and levels that explanations of psychiatric conditions should include, and how to develop adequate explanations of these conditions based on causal theories. For instance, Kandel (1998, 1999) argues that psychiatric disorders can be explained in terms of molecular biology, and that we can reduce higher level explanations of these conditions to the levels of neurotransmitters and genes. In contrast, theorists such as Murphy (2006) argue that the conceptual and methodological problems in diagnosis and classification could be improved if diagnostic categories were based on causal theories from cognitive neuroscience. Further, Murphy (2006), Kendler (2008) and Mitchell (2008) highlight the limits of nomological explanations in psychiatry, and discuss the possibility of generating mechanistic explanations of these conditions. Based on an analysis of the various obstacles to generating causal explanations of psychiatric disorders, and the various arguments put forward by theorists such as those just discussed, I generate a set of criteria that adequate explanation patterns for psychiatric conditions should include. These criteria are not meant to be exhaustive or definitive, but rather a preliminary tool for evaluating the ability of different explanatory patterns to adequately represent the complexity of causation and progression of conditions such as autism spectrum disorder.

Chapters three, four and five explore past and current conceptual frameworks in psychiatry, and how different explanatory patterns for ASD have been developed within those frameworks. Through an analysis of these past and current patterns of explanations for these conditions, I argue that a diagnosis of ASD in an instantiation of the overall conceptual framework and general explanatory pattern for mental disorders that psychiatry adopts in a given time period. For this analysis, I use Thagard (1999)'s version of *explanation schemas*. Explanation schemas highlight the causal factors each explanation identifies in the development of psychiatric disorders like ASD. I argue that each of these explanation patterns do not meet all the criteria for adequate explanation patterns for psychiatric conditions developed in this thesis, and thus may be unable inform more reliable and valid diagnostic categories. However, I argue that an integrated explanation pattern for the autism spectrum may be the most powerful, and may improve diagnosis and treatment. Chapter six develops a sketch of an integrated explanation schema for ASD, incorporating the most powerful schemas discussed throughout the thesis, and there I argue that such a schema could be an adequate pattern of explanation for psychiatric conditions. Finally, I argue in chapter seven that cause-based diagnosis may help to further reduce the stigma associated with ASD and other psychiatric disorders and address the ethical issues involved in the allocation of resources to treat those with autism spectrum disorder.

1.2 Philosophical Implications of Psychiatric Classification and Diagnosis in the Wake of DSM-V

Since the beginnings of psychiatric practice, there has been significant disagreement regarding how to best explain psychiatric disorders. With the exception of only a few conditions, such as Downs Syndrome, disorders currently listed in the DSM do not develop based on a fixed set of physiological or environmental causes. The complexity of psychiatric disorders present significant difficulties in adequately explaining the causes and progression of these conditions, thus making discrete, non-overlapping diagnostic categories difficult to generate. Further, poor explanations of psychiatric disorders make it difficult to non-arbitrarily classify and differentiate between these conditions, which in turn affects the accuracy and predictive power of diagnosis (Murphy, 2006; Poland et al, 1994). While psychiatry and clinical psychology have had a standardized diagnostic and classification system since 1952, there is an ongoing struggle to develop diagnostic categories that accurately represent and identify the nature of psychiatric disorders.

The current diagnostic and classification system used in psychiatry is contained in the Diagnostic and Statistical Manual, which contains over 300 conditions. There has been much debate and discussion surrounding the compilation and publication of the fifth edition of the Diagnostic and Statistical Manual, just released in May 2013. The creation of a new edition of the DSM has sparked intense debate between clinicians and theorists regarding how psychiatric conditions should be classified and diagnosed. Several theorists and clinicians have discussed the shortcomings in current psychiatric diagnosis, and have debated the benefits of changing the way psychiatric conditions, including ASD, are diagnosed and classified in the newest edition of the DSM, as well as future versions (Sirgiovanni, 2009).

Since the publication of third edition of the DSM, clinicians have relied on characteristic patterns of cognitive and behavioural symptoms to classify psychiatric disorders, and use criteria based on these characteristic symptoms to diagnose these conditions. However, diagnosing psychiatric disorders based on their symptoms alone generates significant conceptual and methodological problems, which I discuss in more detail in the next chapter. These problems have motivated many philosophers and clinicians to re-evaluate how psychiatric disorders are defined, classified, and diagnosed.

In the language of clinical psychiatry and psychology, diagnostic categories need to have *reliability* and *validity* in order to accurately identify psychiatric disorders in the clinical population. *Reliability* in this case refers to the consistency with which a diagnosis is given, and thus reliable criteria accurately identify a particular disorder across individuals, over time in the same individual, and across different clinical assessments. On the other

hand, *valid* diagnostic criteria identify individuals with a particular disorder, based on accurate accounts of the fundamental or distinguishing features of that disorder, such as progression, onset, and prognosis (Jablensky & Kendel, 2002; Kendel, 2002). If diagnostic criteria have poor validity, they may be weakly related or potentially unrelated to the underlying malfunctions that are causing the outward symptoms, thus misidentifying the characteristics of a particular disorder. If the characteristic features of a particular disorder are not well understood, it can be difficult to generate criteria that consistently identify that condition, thus resulting in low reliability.

Symptom-based diagnostic criteria are prone to both validity and reliability problems. Identifying psychiatric disorders based on their symptoms often does not reflect their causal structure and processes, and does not accurately differentiate between disorders. Given the emerging and rapidly expanding research from neuroscience, genetics and epigenetics, clinicians such as Kupfer, First & Regier (2002) and philosophers such as Murphy (2006) argue that it is important for diagnostic categories to cohere with empirical data from these disciplines in order to improve the reliability and validity of diagnostic categories in psychiatry. Many of these theorists have argued that basing diagnosis on cause-based, rather than symptom-based, criteria will eventually lead to a diagnostic and classification system that is more accurate and more predictive of the onset, progression, and symptomatology of psychiatric conditions. Thus, it is important to address whether a cause-based diagnostic and classification framework is tenable for psychiatric conditions, given that emerging data from neuroscience, genetics and epigenetics may motivate significant changes in the way these disorders are classified and diagnosed.

The debate regarding the tenability of a cause-based diagnostic framework compared to the current framework provides an opportunity to investigate central metaphysical, epistemological and ethical problems in psychiatric diagnosis. One of the most important metaphysical issues is the way in which emerging data from neuroscience, genetics and epigenetics should impact and inform the concept of what a psychiatric disorder is. For instance, if psychiatric conditions are biologically-based disorders, where breakdowns in biological mechanisms result in cognitive and behavioural symptoms, there is little justification for the view that psychiatric disorders are purely socially constructed. Further, there is little justification for the current legal, social and professional distinctions between disorders of the 'mind' and disorders of the body that have existed since psychiatry's beginnings.

Epistemic issues include further elucidating the relationship between patterns of explanations, diagnosis and treatment for psychiatric disorders, such as the autism spectrum. One of the main issues I investigate in this thesis is the way in which past and present explanatory patterns for ASD increase or limit the validity and reliability of diagnostic

categories and the effectiveness of treatment. I show that weak or inadequate explanatory patterns for psychiatric disorders, such as the autism spectrum, limit the reliability and validity of diagnostic categories, and limit the success of treatment approaches. Through an examination of the way in which different patterns of explanation for ASDs influence the explanatory and predictive power of diagnosis and the effectiveness of treatment, I evaluate what sort of explanatory patterns are the most tenable, given the complex, interactive and multi-directional causation in the development and progression of psychiatric conditions such as the autism spectrum. I argue that the diagnosis and treatment of ASD may indeed be improved if psychiatry were to adopt a cause-based diagnostic and classification framework informed by multi-level, interactive mechanistic explanations that incorporate the biological, cognitive and social levels of description.

In addition to the metaphysical and epistemic issues involved in evaluating the tenability of a cause-based diagnostic framework, there are several ethical issues that arise as well. Based on the investigation into what explanatory pattern may be the most powerful for psychiatric conditions like ASD, and my argument that causal theories can improve the diagnosis and treatments of these conditions, I make two ethical claims. First, the legal and social distinctions that continue to exist between so-called 'mental' and physical disorders are unjustified if psychiatric disorders such as ASD are best explained using an integrated explanation pattern. Psychiatric disorders should not be considered different in kind than disorders treated by physicians in other branches of medicine, since 'mental' disorders and many 'physical' disorders have the same causal structure if psychiatric disorders are the result of complex interactions between biological, cognitive and social causes. For instance, conditions such as type-II diabetes, obesity and lung cancer from smoking are the result of complex interactions between biological causes (such as genetic predispositions), cognitive causes (such as choosing to continue to smoke or consume high amounts of fatty and high-sugar foods), and social causes (such as living in an area or subculture where tobacco smoking and a high fat diet are common). Further, the treatment of psychiatric disorders is just as important as the treatment of these 'physical' disorders, since psychiatric disorders can be just as financially, emotionally, and physically devastating if left untreated. Second, I argue that a cause-based diagnostic and classification framework will further reduce the social stigma associated with the diagnosis of a psychiatric disorder. If psychiatric conditions such as ASD are best explained using an integrated explanation pattern, they should not be considered character flaws or socially deviant attitudes. Rather, psychiatric conditions are real disorders that are *the result of complex interactions between biological, cognitive and social causes*, just like medical conditions such as type-II diabetes, obesity and lung cancer caused by smoking tobacco.

1.3 Autism Spectrum Disorder: A Case Study

We can begin to address these questions by looking at particular disorders as a case study. I begin with a brief overview of the clinical history of the disorders now known as Autism Spectrum Disorder (ASD). There has been much speculation and commentary regarding possible changes to psychiatric diagnosis and classification for future editions of the DSM, and one of the areas of controversy are the disorders previously recognized as subtypes on the autism spectrum.

The previous edition of the DSM, *Diagnostic and Statistical Manual, Fourth Edition, Text Revision* (DSM-IV-TR) defined autism spectrum disorder as a set of related conditions under the category of *pervasive developmental disorders*. In the fourth edition of the DSM, *Autism spectrum disorders* is an umbrella term for any disorder that shares three characteristic socio-cognitive symptoms: 1) impairments in communication and interaction, 2) lack of symbolic play, and 3) stereotyped and rigid behaviours. The disorders that fall under this category are: *Autistic disorder*, *Rett's disorder*, *Childhood Disintegrative Disorder*, *Asperger's syndrome* and *Pervasive Developmental Disorder Not Otherwise Specified*. DSM-IV-TR recognizes these disorders as related, but the variants of ASD besides autistic disorder, such as Asperger's syndrome, were not diagnostic entities until the release of DSM-IV. There are many issues regarding the diagnosis, classification, and possible explanations for the disorders on the autism spectrum, which I argue are partially the result of larger conceptual and methodological problems with the current classification and diagnostic system in psychiatry.

Some of the motivation for refining the diagnostic criteria for ASD include the fact that it is difficult to clearly differentiate between subtypes on the autism spectrum based on symptoms alone, and it is not clear why or how each subtype is distinct from the others (Volkar, State & Klin 2009). For instance, there has been some controversy regarding the placement of Rett's disorder and childhood disintegrative disorder on the autistic spectrum. However, Volkmar, State & Klin (2009) note that genetic and neurological studies of the disorders on the autism spectrum have helped to further define and differentiate between the subtypes of these conditions, and may motivate changes to future classifications of autism spectrum disorder.

There have been investigations into the genetic origins of Rett's disorder, which may be a different pathogenesis than other subtypes of autism, such as the previously-recognized autistic disorder, Asperger's syndrome, and pervasive developmental disorder, not otherwise specified (PDD-NOS). Amir, et al. (1999) found that a mutation in the MECP2 gene was present in 80% of girls with Rett's disorder. In DSM-IV-TR, Rett's disorder was identified as a subtype of autism spectrum disorder, but unlike the other previously-recognized

subtypes on the spectrum, it only affects female children, and its onset is preceded by a period of normal development. Further, unlike other disorders that were identified in DSM-IV-TR as the autism spectrum, which have much more complex genetic features that are still not well understood, Rett's disorder is thought to be the result of this single mutation (also see Van Acker, Loncola, & Van Acker, 2005; Schanen, 2006; Thagard & Findlay 2010). Thus, Rett's disorder may have different developmental and biological characteristics from the other conditions previously identified as part of the autism spectrum. If so, such differences in causation, progression, onset, and gender prevalence distinguish Rett's disorder from other conditions, and may justify the re-classification of this condition.

Also, there is evidence that childhood disintegrative disorder (CDD) has a different etiology and follows a different developmental course than the other disorders previously identified in the autism spectrum. Volkmar et al. (2009) state that this condition is relatively rare (compared to the others on the spectrum), and is marked by a specific developmental pattern that differs from autistic disorder, Asperger's disorder, and PDD-NOS. Children with CDD have a period of normal development, followed be a severe regression of socio-cognitive and cognitive development. Volkmar et al. (2009) state the prognosis for children diagnosed with CDD is worse than children diagnosed with other previously-recognized subtypes of ASD, since most of these children are unable to acquire these lost socio-cognitive and cognitive skills, and make little or no progress in developing new skills after the period of regression. For these reasons, Volkmar et al (2009) state that "[a]s a diagnostic entity CDD is of interest for several reasons...In the first place the disorder is so distinctive that it would seem ripe for delineation of specific genetic or other mechanisms (p. 111)." If biological underpinnings of CDD are further elucidated, there may be more compelling evidence that CDD differs from the other autism subtypes in terms of causation, onset, progression and prognosis. Similar to the case of Rett's disorder, further genetic and neurological research may provide more justification for considering CDD a distinct disorder from ASDs, even though individuals with this condition exhibit autistic symptoms.

Finally, another of the proposed changes to diagnostic categories and classification for DSM-V was the removal of Asperger's syndrome. Baron-Cohen (2000) suggests that Asperger's disorder represents a *difference* in cognitive style, rather than cognitive *deficiencies* or *disabilities*. He (2009) argues that Asperger's syndrome should not be included in the autism spectrum, where the other disorders are characterized by much more severe socio-cognitive and intellectual difficulties. Rather, he states that this 'difference' in cognitive style may make individuals with Asperger's more suited to occupations that require considerable knowledge in narrow domains of information and attention to technical details (such as engineering). Baron-Cohen (2000) notes that the only 'deficit' that marks this

'disorder' is the lack of interest and/or ability to interact with others (individuals with Asperger's, by definition, have IQs in the typical range and do not exhibit language delays; see Baron-Cohen 2000 for a more detailed discussion). However, he argues that individuals with Asperger's should not be considered 'disabled' because of a lack of social interaction, since these individuals are often able to function in society, and can even attain high-level employment in fields such as engineering or the hard sciences. While there are neurological and cognitive differences between individuals with Asperger's and individuals with typical development, Baron-Cohen (2000) suggests these differences may not be severe or 'different' enough to warrant calling Asperger's syndrome a disorder.

DSM-V introduces several changes to diagnostic categories, including the conditions previously identified as the autism spectrum. The separate subtypes of autism spectrum disorder have been melded into a single diagnostic category: *Autism Spectrum Disorder.* The previously separate categories of disorders are now considered to be a single condition, with various levels of disfunction and severity of symptoms in communication and social interaction, and various levels of restricted and repetitive behaviours and interests (DSM-V). Thus, the new category *Autism Spectrum Disorder* encompasses the previous diagnostic categories of *autistic disorder, childhood disintegrative disorder, pervasive developmental disorder not-otherwise-specified,* and *Asperger's disorder.* Finally, the diagnostic category Social Communication Disorder was introduced, and was intended to help distinguish between children with difficulties in language and children with ASD.

While these changes will spark new debate regarding diagnostic accuracy and treatment efficacy, this thesis discusses the debates and proposed changes leading up to the release of DSM-V, and discusses the effectiveness of a symptom-based diagnostic and classification system, rather than address specific changes to the symptom-based diagnostic categories. The arguments presented here deal primarily with patterns of explanation for conditions such as Autism Spectrum Disorder, rather than the accuracy or efficacy of particular diagnostic categories. Thus, the arguments contained in this thesis apply to both the current edition of the DSM as well as the previous editions, since DSM-V still uses the same type of diagnostic approach: categories of disorder based on similarity and severity of symptoms, rather than categories based on etiology.

1.3.1 Clinical and Classification History of Autism Spectrum Disorder

The developmental conditions now known as autism spectrum disorder were first introduced into the clinical literature in the 1940s. However, the term 'autism spectrum disorder' is a

fairly modern one, first used in the fourth edition of the *Diagnostic and Statistical Manual*. Leo Kanner and Hans Asperger published papers describing as-yet unknown psychiatric disturbances in children in 1943 and 1944 respectively (Frith, 1991). Kanner published the results of a study of 11 children in his clinical practice, while Asperger published his findings in his doctoral thesis "'Autistic Psychopathology' in Childhood." Both papers contain descriptions of the disorders now known as autistic disorder and Asperger's syndrome, included symptomatology and a description of subjects' developmental and familial history. In Kanner's (1943) paper, titled "Autistic Disturbances of Affective Contact" he identified the children's inability to interact with others in their social environment as the hallmark of this disorder. Kanner described this defining pathological feature as an

> inability to relate to themselves in the ordinary way to people and situations from the beginning of life. Their parents referred to them as having always been 'self-sufficient' [or] 'acting as if people weren't there' There is from the start the sort of extreme autistic aloneness that, whenever possible, disregards, ignores, shuts out anything that comes to the child from the outside. (Kanner, 1943, p. 41; in Donnellan, 1985.)

He noted that these children experienced language delays, and often displayed echolalia, where they would repeat phrases and questions posed to them (Kanner, 1943). Also, the children in his study had difficulty with personal pronouns, often being unable to apply "I" and "you" appropriately. Kanner (1943) discussed in detail his patients' developmental history, symptomatology, family history, and level of cognitive and intellectual functioning. In his commentary on the 1943 paper, Bishop (1989) states that Kanner

> did not attempt to specify strictly defined diagnostic criteria, but presented detailed case histories of eight boys and three girls, noting the following characteristic features:
>
> 1. Inability to relate to people, including family, from the beginning of life.
> 2. Failure to develop speech or abnormal, largely non-communicative use of language in those that did speak. Pronoun reversal in those that could speak (eight cases), and echolalia, obsessive questioning and ritualistic use of language in several.
> 3. Abnormal response to environmental objects and events, such as food, loud noises and moving objects. Kanner viewed the child's behaviour as governed by an anxiously obsessive desires for the maintenance of sameness, which led to a limitation in the variety of spontaneous activity.

4. Good cognitive potential with excellent rote memory and normal performance on the non-verbal Sequin form board test.
5. Normal physical status. Several children were clumsy in gait but all had good fine motor control (p. 109; See Kanner 1943 as well for an extended discussion of these clinical features).

These features were observed in varying degrees across all eleven patients. However, Kanner (1943) himself noted, as does Bishop (1989), that there was heterogeneity in symptoms and the severity of the language impairments. However, Kanner believed that all eleven children were suffering from the same, as yet unreported, disorder, which he called *early infantile autism*.

Kanner adopted the label "autistic" to describe these children. He drew upon Eugene Bleuler's (1916) discussion of dementia praecox, a general category of psychosis eventually narrowed to refer to patients now diagnosed with schizophrenia (Frith, 1991; Nadesan, 2005). Bleuler used the term "autistic" to describe his patients' withdrawal from the world around them into their delusions. Nadesan (2005) notes Kanner recognized that his use of the term "autistic" differed from Bleuler's use, since it did not describe schizophrenic symptoms in the case of his subjects. Further, Kanner (1943) stated that the behaviour he was observing in the children was not the behaviour of a child suffering from schizophrenia. Schizophrenic patients have developed socio-cognitive capacities that deteriorate as the condition progresses. Autistic children, Kanner (1943) noted, show arrested social development and cognitive dysfunctions early in their lives, which he argued led to later social withdrawal and isolation (Tidmarsh & Volkmar, 2003). While the term 'autistic' described the behaviour of the children in his study, Kanner (1943) maintained that he had identified a childhood disorder separate from schizophrenia.

Bishop (1989) states many clinicians found Kanner's account of the eleven children to be similar to cases of children with psychiatric disturbances they had been unable to diagnose. However, the adoption of 'early infantile autism' into the classification system created as much confusion as it alleviated. For instance, many clinicians were unwilling to accept that early infantile autism was a separate disorder from schizophrenia. Also, Wing notes that Kanner himself stated clinicians would often use the diagnosis far too widely, making an assessment of early infantile autism on only one or two of the features listed above (Wing, 1976). On the other hand, some clinicians would refrain from making a diagnosis of early infantile autism unless the child displayed total social withdrawal and no awareness of other people, even though none of Kanner's original subjects were this severely impaired (Wing, 1976).

At the same time Kanner was writing about early infantile autism, Hans Asperger wrote his doctoral thesis called "'Autistic Psychopathology' in Childhood." He based his thesis on research with a group of boys under his care in the remedial education program at the University Pediatric Clinic in Vienna. Like Kanner, Asperger also used the term 'autistic' to characterize the behaviour and interaction patterns of the boys he studied. He too, adopted Bleuler's use of the term, which described the social withdrawal and isolation of individuals with schizophrenia. While Kanner and Asperger had never met, their findings regarding the psychopathological characteristics of the disorders they studied agree in many respects. Frith (1991) notes that both researchers recognized the importance of the three central symptoms of autism: difficulties with language, communication and interaction, and stereotypic behaviours. Further, both argued that the disorders they identified were to be distinguished from childhood schizophrenia.

The major difference between Kanner's and Asperger's subjects is that Asperger's children did not exhibit the same language difficulties and delays that Kanner's patients did. While Asperger's boys displayed the same deficits with communication and interaction, they were able to express themselves (with varying degrees of relevance and attention to a particular situation) using sentences (Asperger, 1944, in Frith, 1991). Asperger also discussed the problems with socialization and educational progress displayed by these children. When their Intelligence Quotients and level of intellectual development were tested, the children often went on tangents and communicated the knowledge they possessed in a haphazard, convoluted way. Their answers confounded the test scores, making it difficult to assess the severity of the intellectual and language problems. Asperger also commented on the children's difficulties in learning social norms and proper classroom behaviour. He noted that the children under his care did not respond to the authority of their teachers, often acted against the teachers' instructions, and did not interact well with other students.

The similarity between the accounts of the disorders studied by Kanner and Asperger has been documented by Wing (1991). The common discussions of the same and similar symptoms could indicate that these two researchers were encountering disorders with similar underlying pathologies. Asperger (1979) himself wrote about the "astonishing similarities within these two groups which accounted for the same choice of name." (p. 98, in Firth, 1991). Asperger (1979) argued that Kanner's autism is a much more severe form of 'autistic psychopathy.' On Wing's account, these disorders share the same triad of symptoms described earlier, but differ in the severity of these characteristic symptoms. Frith (1991) also discusses the relationship between autistic disorder, as characterized by Kanner, and Asperger's disorder. She (1991) argues that these disorders should belong to the same diagnostic group because they share similar symptoms.

Rutter (1978) discusses the diagnostic inconsistencies that were common during the years leading up to the DSM-III, noting that several terms were used interchangibly to refer to the disorder Kanner identified, such as 'early infantile autism', 'childhood schizophrenia', and 'childhood psychosis'. These terms were applied to children that had some or all of the clinical features outlined by Kanner, and none was used consistently in the clinical literature (Bishop, 1989). While the three characteristic symptoms of autism remained key diagnostic criteria used by psychiatrists throughout revisions to the *Diagnostic and Statistical Manual*, autism's place in the classification taxonomy has changed over these revisions. In the first edition of the DSM, Kanner's 'early infantile autism' did not have its own diagnostic category, but was referred to as "Schizophrenia, childhood type." (DSM-I). 'Early infantile autism' remained a subcategory of schizophrenia in DSM-II (DSM-II), and was not a diagnostic entity on its own until the publication of DSM-III (Tidmarsh & Volkmar, 2003). In the third edition of the DSM, many changes were made to the classification of autism and its diagnostic criteria. First, the term *early infantile autism* was changed to *autistic disorder*, and was now identified as a distinct developmental disorder, unrelated to childhood schizophrenia. In DSM-III, *autistic disorder* was included under "Pervasive Developmental Disorders," a new diagnostic category added to the third edition (Tidmarsh & Volkmar 2003). Conditions listed under pervasive developmental disorders were categorized as such because the social and cognitive dysfunctions affected all spheres of a child's life (Tidmarsh & Volkmar 2003).

While the disorder Kanner identified has been included in the DSM since the publication of its first edition, Asperger's syndrome was not well known in the English speaking world and was not included in the DSM until the fourth revision in 1994. In 1990, the World Health Organization published the ICD-10, where Asperger's syndrome is included as a sub-group of pervasive developmental disorders (Wing, 1991). Francis, First & Pincus (1995) state that

> the diagnosis of Asperger's Disorder has been introduced into DSM-IV to identify a potentially interesting subgroup of patients who have the marked impairments in social interaction and the restricted and repetitive and stereotyped behaviours characteristic of Autistic Disorder but who do not have delays in language development (p. 389).

In DSM-IV, Pervasive Developmental Disorders (PDD) is a category of psychiatric disorders of childhood and adolescence, meaning that the onset of these conditions occur in infancy or early childhood. Francis et al (1995) state that all disorders falling under the category of PDD are characterized by the early onset of severe impairments in social interaction, communication, and the presence of inflexible or restricted behaviours. Although

there are many characteristics included in the diagnostic criteria, Francis et al. (1995) state that the three characteristic symptoms (social interaction, communication, and stereotyped or restricted behaviours) must be observed in the child before 3 years of age in order to make a diagnosis of autism. There is good evidence from the clinical literature that the disorders falling under the autistic spectrum share the same symptomatology, and may indeed be caused by the same underlying neurological impairment(s). Both Kanner (1943) and Asperger (1944) noted the disorders they had identified shared similar symptoms, and may be different manifestations of the same underlying psychopathology. Later theorists and autism researchers such as Wing and Frith echo this view. Clinicians like Firth and Wing have argued that the different types of autism, or different manifestations of autism seen in the clinical literature, are actually sub-types of the same disorder, something that has been adopted in the fourth edition of the DSM, and its text revision (DSM-IV; DSM-IV-TR).

1.3.2 Autism Spectrum Disorder and Cause-Based Diagnosis

There are several features of autism spectrum disorder that make them good case studies for evaluating the tenability and implications of a cause-based diagnostic and classification framework. The inclusion of autism in the DSM since the first edition provides an opportunity to discuss the relationship between the way mental disorders are explained, classified and diagnosed. While the issues discussed by Murphy (2006) and others apply to other disorders in the DSM as well, ASD present some interesting insights into how to causal explanations may improve the diagnosis and treatment of mental disorders.

First, throughout revisions to the DSM, the changes in the definition and classification of ASD have been influenced by the way these disorders have been explained, and the overall conceptual framework in which diagnosis and treatment occurs. Several explanations of the origins of ASD and their symptoms have been suggested since Kanner's and Asperger's original papers. Each of these explanations was generated using the conceptual framework that dominated psychiatric practice at a given time, such as psychoanalysis and behaviourism. However, despite major conceptual changes to both theory and practice in psychiatry, and drastically different explanations of the potential causes of ASD, the account of the three characteristic socio-cognitive symptoms of these disorders has remained largely unchanged since their introduction into the clinical literature by Kanner (1943) and Asperger (1994). The fact that the symptomatology of ASD has not been significantly altered across conceptual frameworks in psychiatry suggests these conditions have relatively stable underlying physical impairments, which have influenced the various explanations of these conditions since their inclusion in DSM-I. An evaluation of past and present explanatatory patterns for these disorders shows that weak or inadequate patterns

limit the validity and reliability of the diagnosis of ASD, and reduce the effectiveness of treatment. However, more powerful explanatory patterns identifying the possible causes of these disorders may improve the accuracy of diagnosis and the success of treatment approaches.

Second, ASD are pervasive, which means they are chronic disorders that first manifest in early development, but continue to affect the individual's socio-cognitive functioning throughout the lifespan. Many autism researchers discussed in this thesis argue that neurological impairments, possibly stemming from genetic abnormalities, cause cascading delays and impairments in cognitive and social development. Thus, ASD have a significant biological component, which indicates that these disorders are physical conditions and not merely social constructions. However, the expansive literature on ASD indicates that the manifestations of ASD's symptoms and their severity are influenced not only by the individual's level of biological and cognitive functioning, but also by conditions in the environment. Further, changes at the social and cognitive levels can cause changes in the neurological impairments characteristic of ASD, indicating that causation in these disorders is complex, multi-directional and the product of an on-going interaction between individual and environment. Thus, ASD present an opportunity to examine how understanding the interactions between biological and environmental causes can potentially generate more powerful patterns of explanation for the disorders on the autism spectrum to aid in the diagnosis and treatment of these conditions. Examining the interactions between biological, cognitive and social causes of disorders such as those on the autism spectrum indicate explanation patterns that are mutli-level, interactionist and mechanistic are the best ways to explain the onset and progression of mental disorders (also see Murphy, 2006, 2008; Kendler & Parnas, 2008; Kendler, 2008; Sirgiovanni, 2009).

Third, despite the stability in the core symptoms and consistency in the diagnostic criteria for ASD throughout revisions to the DSM, these disorders can be difficult to diagnose based on the current criteria, and are often misdiagnosed. Like many other disorders listed in the DSM, there is significant overlap in the symptoms of ASD and other mental disorders. The characteristic symptoms of ASD overlap with the symptoms listed for other disorders of childhood and adolescence, and with the symptoms of disorders on the schizophrenia spectrum (Tidmarsh & Volkmar, 2003). Further, ASD are highly co-morbid with other disorders of childhood and adulthood, which means they often occur along with other, sometimes unrelated disorders in many individuals. For instance, there is often a high co-morbidity rate of mental retardation in children with ASD, particularly autistic disorder. Also, Bishop & Norbury (2002) state that the language impairments seen in ASD are very similar to those seen in pragmatic language impairment (PLI). Further, Norbury & Bishop (2010) also document the similarities between language disorders and ASD, such

as pragmatic language impairment (PLI) and specific language impairment (SLI). These theorists discuss whether these language disorders should be thought of as atypical autism spectrum disorder, or whether ASD and language disorders represent different, and unrelated, kinds of disorders. Thus, classifying and diagnosing the conditions on the autism spectrum based on causal theories, rather than symptoms alone, may improve the accuracy and the diagnosis of these conditions and the effectiveness of treatment.

Fourth, several ethical issues involved in the treatment of ASD and psychiatric disorders in general are partially the result of the way these conditions are currently explained, classified, and diagnosed. The capacity for many individuals with ASD to integrate and function in mainstream society depends on accurate diagnosis, and focused, intensive treatment from an early age. Thus, a better understanding of the interactions between the biological, cognitive and social impairments involved in these disorders may help determine whether or not the treatment of these conditions should be considered medically necessary under the *Canada Health Act*, which would allow better access to behavioural intervention treatment programs and special education programs. Further, explaining the causes of ASD in terms of interactions between biological, cognitive and social causes may help further reduce the pervasiveness of false theories of the origins of these disorders, which often cause significant emotional, financial and social stress for individuals with ASD and their families. For instance, a better understanding of the causes of ASD may help to finally disprove the 'refrigerator mother' hypothesis of these conditions that still influences lay concepts of the disorder, and may also further disprove the vaccine hypothesis of the development of these conditions, thereby helping to reduce anti-immunization behaviours. Thus, an integrated explanatory pattern for psychiatric conditions may help debunk the myths and misconceptions about ASD and other mental disorders that continue to permeate popular culture.

1.4 Patterns of Explanation, Diagnosis and Treatment of Psychiatric Conditions

Several theorists and clinicians have discussed the role of causal theories should play in the next edition of the DSM, and how a better understanding of the causes of psychiatric disorders may begin to address the conceptual and methodological problems in psychiatry. Theorists such as Murphy (2006) and clinicians such as Kupfer et al. (2002) argue that cause-based diagnostic categories will have improved reliability. First, cause-based criteria for each category will be less likely to identify symptoms unrelated to the underlying malfunctions that characterize a particular disorder. Second, such criteria are less likely

to identify symptoms indicative of unrelated underlying malfunctions that characterize different disorders. Further, they argue that cause-based diagnostic categories will improve validity, since cause-based criteria will identify clusters of symptoms that are more likely to be robust indicators of underlying malfunctions, which makes the diagnostic category and criteria more predictive of variables like progression and prognosis. In a cause-based diagnostic and classification framework, treatment will be more focused on addressing these underlying malfunctions, rather than just ameliorating the symptoms they cause. However, it is important to discuss the relationship between patterns of explanation, diagnosis and treatment of disorders such as ASD, since the conditions currently identified as psychiatric disorders pose significant challenges to both classification and diagnosis.

First, psychiatric disorders are complex phenomena that span multiple levels of explanation, and are often the result of complex, multi-directional interactions between biological, cognitive, and social features (Kendler, 2008; Mitchell, 2008a, 2008b). There is significant heterogeneity and variability in the biological, cognitive and social features of individuals who develop a particular disorder, and determining the boundaries and distinctions between categories of psychiatric disorders is difficult based on this large variability. Further, the presentation of symptoms of psychiatric disorders and their severity can change based on the patient's environment, the interactions between the environment and neurological and cognitive functioning, and the effectiveness of treatment. Thus, determining the central, defining features of each disorder is difficult, since these features are the result of dysfunctions in complex interactive systems that are affected by the surrounding environment (Mitchell, 2008a; Woodward, 2008). Research into the genetic and epigenetic origins of psychiatric disorders indicate that these conditions develop through a complex, multi-directional interaction of genetic and environmental factors over an individual's lifetime (e.g. Nestler, 2009). Thus, what symptoms a particular patient presents during a clinical evaluation depends on that patient's early development, current environment, life experiences, and current patterns of thought and affect, all of which are influenced by both biology and the environment. Thus, theorists such as Kendler (2008) argue that complete explanations of psychiatric disorders would have to include many levels of description, since the social, cognitive and biological features of these conditions are not caused by a single factor, such as a genetic mutation or brain lesion.

Second, there is a large amount of variation among the types of disorders historically and currently listed as psychiatric disorders. For instance, some conditions, such as eating disorders and post traumatic stress disorder (PTSD), seem to have primarily environmental causes. However, other disorders such as autism spectrum disorder and schizophrenia seem to have primarily biological causes. The differences in both the symptoms and theorized causal structures in eating disorders, PTSD, ASD and schizophrenia illustrates the

heterogeneousness of conditions defined as 'psychiatric,' and highlight the difficulty in developing a concept and definition of these disorders that subsumes all conditions in the DSM. Causal complexity, heterogeneity in the categories of psychiatric disorders, and the lack of definitive causal theories for most of these disorders has made it difficult to determine what conditions should be considered 'psychiatric' and included in an empirically-informed, standardized psychiatric nosology.

Despite these difficulties, it is possible to generate a preliminary list of criteria for adequate explanation patterns for psychiatric conditions. I show that explanation patterns unable to adequately capture and represent the causal complexity of ASD reduce the reliability and validity of the diagnostic category *autism spectrum disorder*, which in turn limits the explanatory and predictive power of diagnosis and the effectiveness of treatment. To capture the dynamic and multi-level nature of the causes of psychiatric disorders, an explanation pattern must be able to identify causes on multiple levels, and explain how environmental factors can influence the biological and cognitive aspects of these conditions, not just how biology influences the cognitive and social features. While there are no 'psychopathological' laws determining the relationship between certain underlying impairments and observable symptoms, the complex causal interactions that lead to the development of conditions such as ASD in certain individuals can be represented as mechanisms.

Mechanistic explanations can link phenomena at different levels of description without having to subsume these phenomena under laws. A mechanism is an explanatory tool in the philosophy of science, where parts and the interactions between them cause a particular phenomenon (see Bechtel & Abramson, 2005; Machamer, Darden, & Craver, 2000). Mechanisms can bridge several explanatory levels by tracking the interactions between the parts of the mechanism if these parts are at different explanatory levels. Thus, a mechanistic representation of the characteristic neurological, cognitive and social impairments of ASD can capture the complex, multi-directional, and dynamic relations between the biological, cognitive and social aspects of this disorder. In this way, mechanistic explanations of the disorders on the autism spectrum may provide a better account of why certain neurological, genetic and epigenetic changes are associated with the characteristic cognitive and behavioural symptoms. While indirect or secondary causes may influence the nature of the symptoms a patient is presenting, to be useful in diagnosis, an explanation needs to only identify those causes that are necessary for a disorder to be present. If we can identify such primary causes, we can generate a multi-level mechanisms of the causal relations that result in a particular psychiatric condition.

Determining the best explanatory pattern for ASD can also provide insight into how best to explain psychiatric disorders in general. I argue in chapter six that these criteria indicate that mental disorders like ASD are best explained as conditions that arise

based on complex interactions between biological, cognitive and social causes. On this account, autism spectrum disorder is best explained as *breakdowns in neurological mechanisms caused by ongoing interactions between biological factors and the environment, which result in cognitive and behavioural symptoms.* Thus, an *integrated explanation pattern* may capture the complex causation of mental disorders by elucidating the relationships between environmental, cognitive and biological factors. If correct, an integrated explanatory pattern may help to identify and classify psychiatric disorders based on similarities in causal structure, rather than similarity of symptoms.

1.5 Realism About Psychiatric Disorders

This thesis concentrates mainly on issues of explanation and the nature of causation in psychiatric disorders such as ASD, but how these issues are addressed impacts two long-standing metaphysical debates. The first is the debate between realist and constructivist views of psychiatric conditions. The second is the related issue of how the relationship between 'mental' and 'medical' disorders should be understood. It is beyond the scope of this thesis to discuss realism about mental disorders in detail. However, I attempt to identify ways in which an integrated explanation pattern may help develop and support realist views of mental disorders, such as Wakefield's harmful dysfunction analysis (1992; 1997).

While a symptom-based diagnostic system ostensibly improves the reliablity of diagnostic categories, these categories often have weak validity, i.e. it is not clear that they refer to an underlying pathology, or what the nature of such pathology might be. However, merely identifying a cluster of behaviours or cognitive processes as problematic or harmful for the individual or others is not enough to identify that cluster of symptoms as a *disorder*. Symptom-based criteria can do little to determine whether mental disorders are real malfunctions in neurological and cognitive mechanisms, or whether they are simply clusters of behaviours that are problematic from the perspective of social functioning, such as criminal behaviour. As many theorists have argued (e.g. Wakefield, 1992; Murphy, 2006), problematic behaviours in a social context is not enough to determine whether a condition should be considered a *disorder*. Consider the example of criminal behaviour. Acts such as vandalism and violent crime are problematic social behaviours, but are not considered *pathological*, because there is (usually) no neurological or physical dysfunction causing these behaviours. Simply identifying clusters of behaviours as disorders can lead to purely socially constructed or politically motivated categories of psychiatric disorders, and raise serious questions as to whether these conditions are 'real'.

However, merely identifying biological dysfunctions or impairments is also not enough to warrant calling such impairments or dysfunctions *disorders*. For instance, Murphy (2006) discusses the example of the gourmand lesion. If individuals have this brain lesion, they experience more salient and complex tastes and smells than those without the lesion. However, the changes in sensation and perception individuals with this lesion experience do not cause harm to themselves or those around them. Thus, although the gourmand lesion is a neurological dysfunction, what determines whether it should be considered a *disorder* is the nature of the cognitive and behavioural changes caused by this lesion. Thus, an account of the cognitive processes and behaviours that result from the gourmand lesion determine whether the presence of it constitutes a disorder to be treated by psychiatrists. For those who have the gourmand lesion, the cognitive and behavioural changes do not harm the individual or others around them, and in fact having this lesion can enhance sensations such as taste.

Thus, in order to determine what conditions are *mental disorders*, we need to determine whether or not the neurological malfunctions result in cognition and behaviour that is harmful to the individual or others, and we cannot make these judgments on neurological or biological data alone. Thus, any viable realist account of psychiatric disorders will need to identify not just the causes and symptoms of these conditions, but why such conditions should be considered *disorders* in the first place. Realists about mental disorders like Wakefield (1992, 1997) and Murphy (2006) argue for a two-fold picture of mental disorders, where the mental disorders and their diagnosis have both descriptive and normative aspects. These theorists argue that the descriptive aspect of diagnosis comes from facts about the human body that lead to mental disorders, such as breakdowns in neurological mechanisms. However, whether these physical dysfunctions should be treated is based on the extent to which they cause harm in individuals' lives, which is a normative determination.

While I do not formally develop or commit to a particular realist account of mental disorders here, an influential version of this two-stage picture is Wakefield's Harmful Dysfunction analysis. On this account, mental disorders are conditions that "[lie] on the boundary between the given natural world and the constructed social world; a disorder exists when the failure of a person's internal mechanisms to perform their functions as designed by nature impinges harmfully on the person's well being as defined by social values and meanings (p. 373)" (Wakefield, 1992). Thus, both the social and biological aspects of a particular condition are vital to identifying that condition as a harmful dysfunction in need of treatment.

If an integrated explanation pattern of ASD is the most powerful, these conditions are real disorders that are best explained in terms of *breakdowns in neurological mechanisms, caused by ongoing interactions between biology and the enviroment that produce cognitive*

and behavioural symptoms, and can result in significant harm to the individuals who have these conditions. Thus, an integrated explanation pattern for psychiatric conditions may be able to fulfill both the descriptive and normative aspects of the concept *mental disorder* in the Harmful Dysfunction analysis. An integrated explanation pattern may be able to identify the breakdowns in biological and cognitive mechanisms that cause these conditions. However, such an explanation pattern for psychiatric disorders will also include the social and environmental aspects of these conditions, which have a significant influence on their onset and progression. Thus, an integrated explanatory pattern potentially provides a way to link biological impairments with cognitive and behavioural impairments that cause difficulties with daily functioning, gainful employment, etc. that affect many individuals with psychiatric disorders.

According to an integrated explanatory pattern of psychiatric conditions, the descriptive aspects of mental disorders are the interactions between genetic and enviromental factors that cause neurological impairments. The normative aspect of mental disorders, on this account, are judgments about the extent to which these biological, cognitive and social dysfunctions are harmful to the individual who has them, or to other people. These symptoms are harmful to the individual in terms of the ability to function in everyday life, and can be potentially harmful to society if the symptoms become violent or disruptive in nature. If an integrated explanation pattern is indeed the most powerful accounts of psychiatric disorders, we can argue that these conditions are real, biologically-based disorders and not merely social constructions. While social and political factors can shape what sorts of conditions are considered psychiatric disorders (consider the example of homosexuality, only removed from the DSM in its third edition), legitimate psychiatric disorders are those that have biological as well as cognitive and social features that pose significant harm to the patient or those around them.

The second is the related issue of how the relationship between 'mental' and 'medical' disorders should be understood. Murphy (2006) argues that poor explanations of the causes of psychiatric disorders make realist accounts of these conditions harder to develop and justify. He states that if we do not know what causes these conditions, it becomes more difficult to argue that the cluster of symptoms identified by clinicians should be considered a 'real' disorder, which type of disorder, or in fact a *disorder* at all. Further, poor explanations of the causes of the conditions currently listed in the DSM make it more difficult to determine why a particular condition should be considered a *mental disorder*, rather than a *medical disorder*, and why certain conditions should be included or excluded from either category.

Historically, conditions diagnosed and treated by psychiatrists were considered different in kind than conditions treated in other branches of medicine. Many aspects of this tradi-

tional distinction, such as proposed differences in causal structures of "mental" and medical disorders, have remained in the contemporary literature. For instance, even though most contemporary theorists hold that psychiatric disorders are biologically-based phenomena, many argue that they should be distinct from other medical disorders because of the larger role cognitive aspects such as beliefs, agency, and intention play in defining psychiatric conditions. For instance, Arpaly (2005) argues that psychiatric conditions, while real disorders, are "not just like diabetes" because of the nature of the mental and cognitive aspects of these conditions.

Arpaly (2005) states

> "I do not mean to argue that mental disorders are not like diabetes at all. Many mental disorders have all kinds of things in common with many non-mental diseases. Take bipolar disorder, for example. Drugs work for diabetes, and drugs work for bipolar disorder. Neither diabetes nor bipolar disorder can be wished away, trying to 'snap out' of bipolar disorder is as futile as as trying to 'snap out' of diabetes. One should not be ashamed of having diabetes, and similarly one should not be ashamed of having bipolar disorder. Both diabetes and bipolar disorder can be anything from mere chronic inconvenience to the cause of a tragic death, depending to a large extent on the patient's own commitment to treatment (p. 282)".

However, she argues that there is a marked difference between "mental disorders" and medical disorders like diabetes. While Arpaly asserts that both "mental disorders" and "medical disorders" are physical entities, she argues that there some that things that distinguish mental states from other biochemical states. She states that "[f]irst, mental states can be warranted or unwarranted (represent reality or miss-represent it)...whereas other non-mental biochemical states cannot be warranted or unwarranted in that way....Second, mental events can cause each other in ways that non-mental events cannot (p. 283)." However, I argue that such warranted/unwarranted and content-efficacious mental states play a causal role in several medical disorders as well, and as such are not the deciding criteria for whether a condition is "mental" or "medical."

Arpaly (2005) describes the difference between warranted and unwarranted mental states in terms of representations of reality. Arpaly (2005) argues that mental states can either represent reality, and are thus warranted, or do not represent reality correctly, and are thus unwarranted. However, we cannot call the insulin deficiency that characterizes diabetes as warranted or unwarranted, since such non-mental biochemical states do not represent or misrepresent reality in the way that mental states do. Non-mental bodily

states can be desirable or undesirable, but cannot be warranted or unwarranted. Further, she argues (2005) that while mental states can be warranted or unwarranted, they can also be desirable or undesirable. She notes that the difference between claims from psychiatrists that one has a mental disorder, and a lay person claiming that one has a mental disorder is the difference between warrant and desirability. She states "[o]ne big difference between the way psychiatrists use the term 'mental disorder' and the way laypersons use the term 'mental disorder' is that psychiatrists think of mental disorders as 'maladaptive' mental states, or states that cause 'impairment' or 'distress': in other words, they think of mental disorders primarily as *undesirable* mental states, the way that diabetes is an undesirable non-mental condition. To the layperson, being told that one has a mental disorder is first and foremost being told that one has *unwarranted* mental states (p. 284, emphasis in original)." Arpaly continues by stating that "[t]he main message a lay person gets from a psychiatric diagnosis is often not - or not only - 'you have a problem' but '*you are getting something wrong*'" (p. 285, emphasis in original).

The second difference between mental states and non-mental biochemical states Arpaly (2005) identifies is that mental states can cause each other in a way that other non-mental states cannot, a property sometimes called "content efficacy." Arpaly explains content efficacy as follows. She argues that "[m]ental states differ from other bodily states in that they have content: they are about something. One does not, for example, just have beliefs, one has beliefs about love, the location of one's keychain, and as Lewis Carol would put it, of cabbages and kings. Sometimes, though not always, mental states cause one another in a way in which their content plays a central role (p. 285)."

Arpaly is correct that the cognitive or "mental" aspects of psychiatric disorders often carry much of the explanatory weight, especially given the lack of robust causal data for many of the disorders listed in the DSM. These cognitive or mental aspects of psychiatric conditions are important, and arguably we cannot adequately explain or delineate between psychiatric disorders without an understanding of the cognitive impairments involved. For instance, content efficacious, unwarranted, and undesirable mental states can and do play a major causal role in the on-going complex causal interactions that result in the symptoms of several psychiatric conditions, including disorders that also have a significant biological component, such as depression and addiction (both disorders are discussed in more detail in chapter six).

As many theorists discussed in this thesis argue, the cognitive level is an important part of the causal structure of psychiatric conditions, and therefore integral to adequate explanations of psychiatric conditions. The cognitive aspects of psychiatric conditions can include maladaptive beliefs, attributions and judgments. As I argue in chapter six, the cognitive level, which includes phenomena like maladaptive beliefs can be included in *inte-*

grated explanation schema, along with the biological levels and social levels of explanation. In such an integrated schema, the cognitive level can be a significant part of the overall explanation of psychiatric conditions, and may be the primary explanatory level in conditions such as phobias, obsessive-compulsive disorder, body dismorphia and eating disorders.

However, as argued throughout this thesis, adequate explanation patterns for psychiatric conditions must also capture and elucidate the causal interactions between the cognitive, biological, and social levels. While cognitive aspects can be primary causes, "non-mental events" such as biochemical states of the brain and other organs can and do influence cognitive process and behaviour, and can be just as causally efficacious as beliefs and representations of the subject's reality. For instance, in the case of ASD, the impairment of the mirror neuron system, thought to be involved in the development of the disorders, causes specific cognitive impairments such as an underdeveloped theory of mind. Part of the theory of mind deficit seen in individuals with ASD involves the inability to identify and ascribe mental states to others. Even in disorders that have a large "cognitive" component such as major depression or addiction, the physiological level can and does effect the cognitive and social aspects of these conditions.

Further, the "mental" or "cognitive" factors of the type Arpaly (2005) identifies also seem to be present in many traditional medical disorders. For instance, content-efficacious, unwarranted mental states could be implicated as major causes at the cognitive level for conditions such as obesity, certain types of cancers, and type-II diabetes. Certain types of lung, throat and mouth cancers are caused by smoking tobacco, and conditions like obesity and type-II diabetes are caused by "lifestyle choices" such as consuming a high fat/high sugar diet, and not engaging in regular exercise. Despite the wealth of information available to the public about the dangers of smoking and the importance of regular exercise and a balanced diet, many individuals continue these unhealthy behaviours anyway, eventually leading to serious medical conditions. Decisions to continue to smoke or eat an unhealthy diet are cognitive states, or "cognitive causes," and often involve the false belief that one will not develop lung cancer despite the medical evidence to the contrary, or the belief that the pleasure of a nicotine rush or a high-sugar treat are worth the associated physical risks. One could argue that none of these beliefs are *warranted*, nor are they even *desirable*, since they can lead to unhealthy choices and actions. Further, when treating conditions such as obesity or type-II diabetes, patients often have to change how they view the risks versus benefits of continuing unhealthy habits, and attempt to effectively use information about the harms of such behaviour in order to successfully carry out the treatment regime (e.g. sticking to a healthy diet and engaging in regular exercise by reminding oneself of the risks of not doing so, and the benefits of these new habits, etc.).Thus, it seems that there are both *content-efficacious*, and *unwarranted* mental states involved in medical conditions

such as type-II diabetes, obesity and lung cancer caused by smoking. Thus the presence or absence of certain cognitive features on their own may not reliably distinguish psychiatric conditions as a discrete subset of medical disorders. Further, even the presence of content-efficacious, (un)warranted mental states does not seem to cleanly demarcate medical and "mental" disorders.

If the preceding analysis is correct, then psychiatric disorders do indeed have the same causal structure as disorders such as type-II diabetes, lung cancer from smoking and obesity. Thus, if psychiatric conditions like ASD are best explained as the result of complex interactions between biological, cognitive and social causes, they not different in kind from medical disorders, and are as 'real' as other medical disorders. Each of the medical conditions just discussed is caused by breakdowns in biologically-based mechanisms, and can be equally harmful and debilitating to the patients diagnosed with these conditions. Although psychiatric conditions often have environmental causes, many have biological causes as well, such as ASD, schizophrenia, and mental retardation. Likewise, conditions such as heart disease, cancer, type-II diabetes and obesity have both environmental and biological causes. Further, in all the conditions just named, the onset and progression of the symptoms are caused by the bi-directional interactions between these causes at multiple levels, and may be best explained mechanistically, since the deductive-nomological model of explanation is limited in the life sciences (see Bechtel 2007 for a more detailed discussion).

If mental disorders are the result of complex interactions between causes at the biological, cognitive and social levels of description, they should be diagnosed and explained in a similar way to medical disorders. If an integrated explanation pattern is the most powerful for explaining the causes and progression of ASDs, these conditions have the same causal structure as medical disorders such as heart disease, cancer, type-II diabetes and obesity. If correct, an integrated pattern of explanation for conditions like those on the autism spectrum may provide more justification than ever before for beginning to collapse the distinction between psychiatric disorders and other medical disorders. If we can further reduce the differences in the way psychiatric and other medical disorders are explained, future editions of the DSM may be able to incorporate the medical model to a greater extent, which theorists like Murphy (2006) argue is vital to increasing the validity and predictive power of diagnosis in psychiatry.

1.6 Ethical Implications of Cause-Based Classification and Diagnosis

The final chapter of this thesis briefly examines some of the ethical issues involved in a shift to a cause-based diagnostic framework. First, a better understanding of the causes of ASD may help to further reduce the false, but still popular, beliefs that these conditions are caused by poor parenting or the measles, mumps, rubella (MMR) vaccine. Explaining autism spectrum disorder using an integrated explanation pattern may help to further discredit these false casual theories by providing a powerful alternate causal theory to refrigerator mothers and MMR vaccination. Further discrediting these false causal theories in lay society may help reduce the emotional costs associated with feelings of responsibility or guilt by parents, and the financial costs of anti-vaccination behaviours. Finally, if psychiatric conditions are best explained using an integrated pattern, this pattern of explanation may help to further reduce the still pervasive view by lay society that mental and medical disorders are distinct kinds of conditions. Many people still believe that individuals with psychiatric conditions are blameworthy in some way, even though the physical causes of these conditions are beginning to be discovered. However, if psychiatric conditions are best explained as epigenetic disorders, they are biologically-based conditions that develop based on complex interactions with the environment, not character flaws or the product of weak wills and minds.

Second, more effective treatment, based on diagnostic categories and criteria that reflect underlying causes of psychiatric disorders like ASD can help to address several ethical issues in mental health care. First, a better understanding of the causes of ASD can help support legal claims for expanded funding for treatment of these conditions in provinces such as Ontario and British Columbia. If these conditions are best explained as complex conditions that are the result of complex interactions between biological, cognitive and social causes, the early environment plays a significant role in the development and progression of the characteristic symptoms. Thus, early access to intervention programs can help to structure the child's early experiences using behavioural techniques and tools to develop communication skills in order to mitigate and reduce the impact of environmental causes on the severity and progression of the symptoms of ASD. Second, if diagnosis becomes more accurate with a cause-based framework, children with ASD may be diagnosed earlier in their lives, thus saving costs incurred from multiple clinician and physician visits to gain a definitive diagnosis. Further, more accurate diagnosis will save total overall costs of care and treatment of indidivuals with ASD over the lifespan.

1.7 Issues and Arguments in the Following Chapters

This chapter introduced the philosophical issues in diagnosis and classification of mental disorders and how these relate to autism spectrum disorder. Chapter two provides more detail about the problems with symptom-based diagnosis of mental disorders such as ASD. In that chapter, I analyze the relationship between patterns of explanation, diagnosis and treatment in clinical psychiatry, and discuss what sort of explanatory patterns may be best for mental disorders. Finally, I develop a preliminary list of criteria for evaluating and adjudicating between explanation patterns in psychiatry, and discuss how explanation schemas can help to evaluate different accounts of autism spectrum disorder.

Chapter three discusses two early explanatory patterns for autism spectrum disorder from the psychoanalytic and behaviourist eras, and argues that these patterns of explanation do not meet the criteria for adequate explanation patterns for psychiatric conditions developed in chapter two. In chapter four, I discuss explanatory patterns for ASD developed in cognitive psychology and cognitive neuroscience. I argue that like the psychoanalytic and behaviourist explanation patterns, neither of these accounts are adequate explanation patterns for the conditions on the autism spectrum. Chapter five discusses genetic and epigenetic explanation patterns for ASD, and argues that while a genetic explanatory pattern does not meet the criteria outlined in chapter two, an epigenetic explanation pattern may be a potentially powerful one. However, in chapter six I show that an integrated schema meets all the criteria for adequate explanations in psychiatry, and thus may be the most powerful explanation pattern for representing the complex causation involved in the development of psychiatric conditions, such as those on the autism spectrum. Thus, I argue that the diagnosis and classification of psychiatric conditions like ASD may be improved if psychiatry adopts a cause-based framework informed by causal theories that identify the biological, cognitive and social features of psychiatric disorders.

Finally, chapter seven argues that a cause-based diagnostic and classification framework may help to address issues in resource allocation for the treatment of conditions on the autism spectrum. I discuss how biologically-based patterns of explanation for psychiatric disorders can further reduce the social stigma that continues to plague these conditions. Further, I argue that a better understanding of the causes of these disorders can help to support legal claims that the treatment of these conditions is medically necessary under the Canada Health Act, and that these conditions should not be considered different than medical disorders in the legal systems in the United States and Canada.

Chapter 2

Symptom-Based v. Cause-Based Classification of Psychiatric Conditions: Improving the Explanatory and Predictive Power of Psychiatric Diagnosis

2.1 Introduction

The last chapter discussed the classification and diagnostic history of the conditions now identified as the autism spectrum, where I outlined the features of these conditions that make them good case studies to investigate the relationship between patterns of explanation, diagnosis and classification. Like many theorists, such as Murphy (2006), Kupfer et al (2002) and Poland, Von Eckhardt, & Spaulding (1994), I argue that the lack of adequate causal explanations of psychiatric disorders such as ASD is the root of the conceptual and methodological problems that plague the diagnosis and treatment of these conditions. The analysis of the problems with both past and current diagnostic and classification frameworks I provide in this chapter supports the view that psychiatric diagnosis and classification should be based on causal theories rather than observable symptoms alone. However, there are several challenges involved in developing a viable cause-based diagnostic framework for psychiatry. For instance, there is significant disagreement among theorists who advocate a cause-based framework regarding the nature of explanations of psychiatric dis-

orders, such as the levels of description that should be included and emphasized, and how these explanations should influence and inform categories of disorder. Further, there is debate regarding how best to develop adequate explanations in psychiatry, given the complex and heterogenous causation involved in the conditions currently identified as psychiatric disorders. Through an analysis of some of these debates and disagreements, I identify a set of criteria that adequate explanation patterns in psychiatry must have in order to generate reliable and valid diagnostic categories. I argue that a cause-based diagnostic framework will lead to more accurate diagnosis and improved treatment of autism spectrum disorders, provided the explanatory patterns on which it is based meet the criteria outlined in this chapter.

2.2 Conceptual and Methodological Problems with Symptom-Based Classification and Diagnosis

By the 1970s, the early-caused based approach contained in the first two editions of the DSM had come under repeated criticism from the surrounding medical community, as had the overall psychoanalytic framework on which concepts of psychiatric disorders, their diagnosis and their treatment was based (Mayes & Horwtiz, 2005, Shorter, 1993). Mayes & Horwitz (2005) state that the publication of the third edition of the DSM brought with it a radical change in the classification of psychiatric disorders. DSM-III took a more atheoretical approach to diagnosis (Murphy, 2006; Poland et al, 1994), where diagnostic categories were not based on theories of the origins of these conditions, but on observable symptoms of the various conditions. The authors of DSM-III only included criteria that referred to the behavioural and cognitive symptoms that appear to characterize each disorder. This way, clinicians' diagnoses are based on a standardized set of criteria, something that was absent in the first two editions of the DSM.

However, Widiger & Clark (2000) state that the APA acknowledged there was a significant lack of systematic research on most of the disorders in the DSM. Thus, diagnostic criteria included in DSM-III were still heavily based on the clinical judgment of the task force and the committee responsible for diagnostic revision. Further, DSM-III continued to attract criticism on the grounds of poor validity and reliability of diagnostic categories. Widiger & Clark (2000) and Kupfer et al. (2002) argue that a number of problems with the criteria contained in DSM-III arose soon after its publication, since some of the categories contained inconsistent or even contradictory diagnostic criteria. In response to such criticism, DSM-III-R was released in 1987. Widiger & Clark (2000) state that the field

trials conducted by the task force assigned to make revisions to DSM-III were based on improving diagnostic validity, as well as continued improvements in diagnostic reliability.

In 1994, the *American Psychiatric Association* published DSM-IV. Widiger & Clark (2000) state that the committee appointed to construct the DSM-IV aspired to use more conservative thresholds for approvals of new diagnostic categories or criteria. Like the committee in charge of DSM-III, the task force assigned to create DSM-IV attempted to follow the research and clinical literature more thoroughly, and base their criteria on this literature. However, unlike previous editions of DSM, the DSM-IV committee compared diagnostic categories and criteria from other diagnostic systems, most notably the *International Classification of Diseases, 10th edition* (ICD-10, World Health Organization, 1990). Comparing DSM criteria to other psychiatric nosologies was done to standardize the diagnostic system more thoroughly, and to help ensure diagnostic categories incorporated the central features of each psychiatric disorder. In 2000, the APA published the text revision of DSM-IV, which contained some revisions to the categories and criteria contained in DSM-IV, but did not include massive classification or conceptual changes.

The categories included in DSM-IV-TR are what Poland et al (1994) call *lenient* categories, which allow for overlap between them. In this framework, diagnosis of a particular psychiatric disorder requires that an individual meets the central criteria for the disorder in question, i.e. the necessary conditions. However, such an individual may also exhibit symptoms that commonly occur along with the central criteria, symptoms which, by themselves, are not enough to warrant a particular diagnosis. Some psychiatric disorders do not have necessary conditions, but have many sufficient conditions, none of which by themselves are necessary to make a diagnosis of a particular psychopathology. One example of such a psychiatric disorder is substance abuse. For a clinician to give a diagnosis of substance abuse, a patient need only meet one of several sufficient criteria: 1) recurrent substance use resulting in a failure to fulfill major role obligations at work, school or home; 2) recurrent substance use in situations in which it is physically hazardous (e.g. operating a vehicle); 3) recurrent substance-related legal problems; 4) continued substance use despite having persistent or recurrent social or interpersonal problems caused or exacerbated by the effects of the substance. On the other hand, a patient diagnosed with *substance dependence* must exhibit, in addition to any or all of the criteria for substance abuse, 1) tolerance to the substance(s) used, and 2) physical withdrawal when the substance is not consumed.

Under the current diagnostic and classification framework contained in DSM-IV-TR, the way psychiatric conditions are explained and diagnosed differs significantly from other branches of medicine and even closely related fields such as neuroscience and cognitive neuroscience. First, disorders in other fields of medicine are classified terms of their causes,

course, *and* their symptoms, whereas disorders in psychiatry are only classified by their symptoms (Murphy, 2006). While the biological underpinnings of many psychiatric conditions are being discovered, diagnostic categories in contemporary psychiatry are based on sets of co-occurring symptoms over a given time period, rather than the presence of underlying malfunctions. Further, there is still a lack of adequate data about all but a few psychiatric conditions in terms of causes or disease progression. Most research is dedicated to the continued investigation of the signs and symptoms of the disorders contained in DSM-IV (Poland et al, 1994). Thus, DSM categories can become over-reified, where the concept of a particular psychiatric disorder, e.g. ASD, *just is* the set of symptoms listed in DSM-IV (Widiger & Clark, 2000).

Second, definitions of disorders in other branches of medicine include phenomena at a lower level of description that cause outward symptoms, the course of the disorder, and the prognosis based on the extent of the impairment in those lower-level phenomena. In contrast, the disorders contained in the DSM are defined simply as clusters of cognitive and behavioural disturbances co-occurring acutely over a given period of time. DSM criteria identify only cognitive and behavioural symptoms, and contain no mention of lower-level phenomena underlying or co-occurring with the symptoms observed. For instance, the diagnostic categories for the disorders on the autism spectrum only include criteria that identify the characteristic symptoms of these conditions (impairments in language, social interaction and rigid behaviours) and make no mention of possible underlying malfunctions identified by autism researchers, such as deficits in theory of mind or malfunctioning mirror neurons. For these reasons, critics of the current diagnostic system in psychiatry argue that a symptom-based approach cannot resolve, and in most cases exacerbates, the poor reliability and validity that has plagued the diagnosis of psychiatric disorders since the early days of psychiatry.

Third, the process of diagnosing a patient with a psychiatric disorder differs from other fields of medicine. Like other branches of medicine, psychiatrists try to explain their patients' symptoms by giving a diagnosis. However, in other branches of medicine, explanations of patients' symptoms are given in terms of phenomena at a lower level of description than the symptoms themselves (Murphy 2006). For instance, the symptoms of Alzheimer's disease, e.g. memory loss, personality changes, and dementia, may be explained by the presence of neuronal loss, senile plaques, and neurofibrillary tangles (Martin, 2002). A diagnosis of Alzheimer's disease involves identifying the characteristic symptoms of this condition and finding these senile plaques and neurofibrillary tangles in patients, which are phenomena at the neurophysiological level of explanation. In the case of a disorder like Alzheimer's disease the symptoms are described in cognitive and behavioural terms, but what may explain the presence of these symptoms and allows a physician to give a

diagnosis are physiological changes in the brain. In contrast, conditions such as 'autistic disorder' are identified only by the characteristic symptoms just mentioned, and a diagnosis of autistic disorder is made only on the basis of these symptoms, not on the presence of certain neurological or physiological changes.

There are several conceptual and methodological problems with the current symptom-based framework that reduce the reliability and validity of diagnostic categories, thus limiting the explanatory and predictive power of psychiatric diagnosis. First, the current symptom-based diagnostic and classification framework is supposed to be atheoretical, since diagnostic categories and the diagnostic process are not based on causal theories of the different conditions and how they arise (Poland et al, 1994; Widiger & Clark, 2000). However, Poland et al. (1994), Kupfer et al (2002) and Murphy (2006) argue that the current symptom-based diagnostic approach operates under the false notion that mental disorders are *syndromes with unity*. Syndromes with unity are conditions where the overt symptoms are robust indicators of underlying malfunctions, and the relationship between the appearance of certain symptoms and the presence of underlying malfunctions is a nomological one.

However, these theorists argue that the presence of co-occurring symptoms does not necessarily provide insight into the nature of the underlying biological pathology. Further, the relationship between the presentation of symptoms and the presence of certain underlying malfunctions is not a law-like one in the case of most psychiatric disorders, including ASD. The presentation of symptoms and their progression is affected by both the complex interaction of biological and cognitive factors, and the environmental context in which these symptoms appear (see Kendler, 2008, Poland et al, 1994; Kendler & Parnas, 2008). Conditions like ASD are not compact, discrete disease entities where one cluster of symptoms accurately identifies a particular disorder. Rather, psychiatric disorders are causally complex and casually heterogenous with significant overlap in overt symptoms (Kendler, 2008, Woodward, 2008; Shaffner, 2008: Mitchell, 2008a; Murphy, 2006; Poland et al, 1994). Classifying and diagnosing psychiatric disorders as if they were syndromes with unity results in heterogeneous diagnostic groups, which severely limit the validity and reliability of diagnostic categories.

One of the strengths of the DSM's approach to the classification of psychiatric disorders often identified is an increase in reliability in diagnosis. Reliability, i.e. the consistency of diagnosis across clinicians and over time with the same patient, is ostensibly improved with a symptom-based approach, since the criteria are based on standardized accounts of observable symptoms. However, reliability is limited in a symptom-based diagnostic framework. Recall that even unrelated diagnostic categories identify similar symptoms. Thus, even symptoms that are highly correlated do not necessarily determine whether

that particular cluster of symptoms indicates the presence of one psychiatric disorder, or whether the symptoms are associated with more than one disorder. Further, even highly correlated symptoms often do not indicate whether the patient is suffering from a genuine psychiatric disorder, or merely a problem of living (where an individual has interpersonal, financial or occupational difficulties but should not be diagnosed with a psychiatric condition). Thus, reliable diagnostic categories only really improves diagnosis if those categories also have validity, since false or dubious categories of disorder may be highly reliable. Validity is central to establishing whether the cluster of symptoms identified in diagnostic categories represent distinct psychiatric conditions, and whether the symptoms identified are indications of *disorders*, rather than the less severe problems of living.

However, because symptoms are poor indications of the underlying malfunctions that cause them, it is difficult to validate categories of disorder in a symptom-based approach. In psychiatry, good diagnostic validity occurs when the category and its criteria accurately reflect key variables such as progression, onset, and prognosis based on the severity of underlying malfunctions (Poland et al, 1994; Kendel, 2002). Since symptoms are often a poor indication of underlying malfunctions, and symptom-based diagnostic categories result in heterogenous diagnostic groups, it can be difficult to determine the typical progression, onset and prognosis of a disorder based on the symptoms identified in diagnostic criteria. Critics of the current diagnostic approach such as Poland et al (1994) and Murphy (2006) argue that most categories in the DSM suffer from poor validity, and significant increases in diagnostic validity are unlikely with symptom-based classification of psychiatric disorders (also see Kendel, 2002 and Jablensky & Kendel, 2002 for a discussion of the challenges regarding increasing diagnostic validity).

There are three main problems most often discussed by critics of the current diagnostic approach, which they argue are the result of the limited reliability and validity of symptom-based diagnostic categories. The first is *arbitrary boundaries between categories of disorder*, given that there is significant *overlap in the symptoms and criteria listed for distinct categories of disorders*. The problem of overlapping symptoms is a problem in many diagnostic categories, including ASD. If there is significant overlap between criteria for different disorders, it can be difficult to determine with which disorder a patient should be diagnosed. For instance, in autism spectrum disorders, there is significant overlap in the symptoms of these conditions and the symptoms of currently unrelated conditions that often occur along with ASD. Tidmarsh & Volkmar (2003) state that children with autism often exhibit symptoms from a wide variety of disorders, including depression, anxiety, oppositional behaviour, hyperactivity, poor attention, tics and compulsions. In these situations, it can be difficult for a clinician to determine whether a diagnosis of autism should

be given. The symptoms a child displays may indicate the presence of a co-morbid disorder as well as an ASD, or could indicate an autism spectrum disorder only, even though the symptoms are atypical.

The second problem often discussed is *over-inclusion* in diagnostic categories. Over-inclusion often occurs as a result of the problem of over-lapping criteria, since patients can and often do meet criteria for more than one disorder. For instance, ASD are highly co-morbid with mental retardation (particularly in the case of autistic disorder), and with language disorders that arise in childhood. Most children with ASD have difficulties with language, and even children with Asperger's syndrome can have some difficulty with language (Rogers & Williams, 2006; Frith, 1991). Further, Bishop (1989) notes that behavioural and language tests are not always able to determine whether a child is most likely suffering from an ASD, or from another disorder. One problem is that individuals with autistic disorder are often so language impaired that some of them do not develop speech at all, or have very limited verbal abilities. Accurately testing whether a child has mental retardation or autistic disorder, or both, is difficult to determine based on a battery of tests of verbal ability. Further, children with mental retardation exhibit similar behavioural symptoms to those of ASD, such as the social withdrawal, restlessness, and rigid behaviours.

However, over-inclusion in diagnostic categories can also occur if patients are not displaying all the symptoms of a particular disorder, but still seem to match some of the core diagnostic criteria. With current diagnostic practices, meeting the primary symptom-based diagnostic criteria is all that is required for a diagnosis of a psychiatric disorder (Poland et al, 1994). However, individuals who have problems of living, longstanding personality problems, interpersonal difficulties, or eccentricity often fit some of the criteria for psychiatric conditions, but should not be diagnosed with a full-blown disorder (Murphy, 2006; Wakefield & First, 2003, Volkmar & Klin, 2005) . For instance, many individuals share some of the characteristics as those diagnosed with Asperger's disorder, such as narrow interests, social awkwardness and lack of significant relationships with others. In such cases, it may be difficult in a symptom-based diagnostic framework to determine when social awkwardness and isolation end and Asperger's disorder begins, especially in individuals where these characteristics affect their ability to function in everyday life.

The third problem raised against current diagnostic methods in psychiatry concerns *arbitrary divisions between subtypes of diagnostic categories.* For instance, autism disorders are situated on a spectrum, and the disorders identified as the autism spectrum contain subtypes that are at the moment linked only by symptoms. Autistic disorder represents the most severe of the autistic disorders, and Asperger's syndrome represents the least amount of impairment and the highest level of social functioning. However, the other disorders

included in the category of ASD, Rett's disorder, Childhood Disintegrative Disorder and PDD-NOS, are not arranged from most to least severe, and have symptoms that are difficult to differentiate from autistic disorder and Asperger's disorder (Wing, 1991). Further, since there is high variability in the severity and presentation of the core symptoms of ASD in each subtype, and across patients in the same subtype, it can be difficult to determine the particular autism spectrum disorder the patient likely has (Wing, 1991). Thus, the distinctions between several of the disorders included in the autistic spectrum, which are often delineated based on severity and variability of symptoms, is not always clear.

Because of the heterogeneousness of the individuals within a particular diagnostic category, the treatment of psychiatric disorders has varying and often limited success. Patients who are diagnosed with a particular disorder are given similar treatments, i.e. a certain class of drugs, and/or cognitive-behavioural therapy focused on minimizing their symptoms. However, since the symptom presentation and underlying biological impairments can be drastically different between patients given a certain diagnosis, a certain drug or therapeutic technique may have limited success on many of these patients. For instance, current treatments for the conditions on the autism spectrum are targeted at ameliorating symptoms, but since the diagnostic category *autism spectrum disorders* includes individuals with a wide variety of genetic, neurological, cognitive features interacting with a wide variety of environments, such treatment will have highly variable levels of success. Thus, while techniques such as behavioural interventions are still the most effective method for managing the symptoms of ASD, they still have varying rates of success among patients, because each patient has different neurological and cognitive impairments, in different combinations, and at different levels of severity.

The conceptual and methodological problems discussed above limit the explanatory and predictive power of psychiatric diagnosis. Since symptoms are poor indicator of underlying malfunctions, individuals included under the same diagnostic category have a wide variety of biological, cognitive and social features. Individuals diagnosed with the same condition will display different patterns of the progression and severity of symptoms, and may have very different underlying biological impairments causing these symptoms. Thus, under the current framework, it is difficult to predict and explain the course, outcome, prognosis and progression of symptoms in patients with disorders such as ASD based on symptoms alone.

Given the problems with the current symptom-based framework, theorists such as those just above argue that the explanatory and predictive power of diagnosis can be improved if psychiatry adopts a cause-based framework. However, there are several obstacles to generating causal explanations and theories powerful and robust enough to motivate and inform a cause-based system. If we can begin to identify what adequate explanation patterns for psychiatric conditions such as ASD might be, we may be able to determine what

sort of causal theories can and should inform a cause-based diagnostic and classification framework in psychiatry. The rest of this chapter identifies the criteria that adequate explanation patterns in psychiatry need to meet by analyzing what makes generating causal explanations of psychiatric conditions difficult.

2.3 Causal Explanations, Diagnosis and Treatment of Psychiatric Conditions

The complex and interactive nature of psychiatric disorders such as ASD present significant difficulties in determining the causes and progression of these conditions, thus making discrete, non-overlapping diagnostic categories difficult to generate. If psychiatry and clinical psychology are going to base the diagnosis of psychiatric disorders on a cause-based, rather than symptom-based, framework, viable causal accounts must address the following problems. First, what sort of pattern of explanation will have the most power? What levels of description should such a pattern of explanation include? Second, how do we generate causal explanations of psychiatric disorders that capture their complexity, but remain general enough to be useful in diagnosis? What sort of indications of impairments should diagnostic criteria identify? Finally, how might adequate causal explanations address the problems of poor diagnostic reliability and validity?

Several clinicians and theorists have discussed the type of explanations most suitable to psychiatric disorders, and have argued that in order for explanations of the conditions contained in the DSM to be viable, they must have certain features. I analyze the obstacles to generating adequate causal explanations in psychiatry, and evaluate arguments by critics of the current diagnostic and classification framework such as Murphy (2006), Poland et al (1994) and others regarding how to address these obstacles. Through this analysis, I develop a preliminary list of criteria that adequate explanation patterns for psychiatric conditions should include, which may help to improve the explanatory and predictive power of psychiatric diagnosis and classification, thus potentially improving the effectiveness of treatment. I adopt Thagard's (1999) version of explanation schemas, which identify the causal features and structures of explanations, to identify and evaluate past and present patterns of explanation for the disorders on the autism spectrum. The next three chapters discuss the different patterns of explanation for ASD in the history of psychiatry and clinical psychology, and identify the strengths and weaknesses of each explanatory pattern. I argue that patterns of explanation that do not explain the causes of psychiatric disorders in terms of interactive, multi-directional mechanisms limit diagnostic reliability and validity, and thus only certain explanatory patterns for conditions like those on the autism spectrum

will be viable. Explanatory patterns and causal theories that do not meet these criteria are not viable candidates on which to base diagnostic categories in future editions of the DSM.

2.3.1 Insight from DSM-I and DSM-II: Problems with Early Cause-Based Classification and Diagnosis

We can begin to examine what sort of patterns of explanation for psychiatric disorders are the most powerful, and how cause-based frameworks can improve diagnosis and treatment by analyzing the shortcomings with the cause-based approach contained in the first two editions of the DSM. The first and second editions of the DSM were heavily influenced by psychoanalytic theory, the dominant conceptual framework in psychiatry at that time (Poland, et al, 1994). In this era, there was still little known about the biological causes of most psychiatric conditions, and psychoanalytic theories placed the origins of these conditions in psycho-sexual conflicts of early childhood. Mayes & Horwitz (2005) state "the focus of analytic explanations and treatments...was the total personality and life experiences of the person that provided the context for the interpretation of symptoms...DSM-I and DSM-II made little effort to provide elaborate classification schemes, because overt symptoms did not reveal disease entities but disguised underlying conflicts that could not be expressed directly (p. 249-250)." However, this early cause-based approach failed to increase the reliability and validity of psychiatric diagnosis, since the causal theories on which it was based were empirically unsupported and internally inconsistent.

There were several problems as a result of the psychoanalytic view of psychiatric disorders and their causes, only some of which will be discussed here. First, most categories of psychiatric disorders included in DSM-I and DSM-II contained psychoanalytic descriptions of the psychological and emotional characteristics individuals with these conditions can display, rather than standardized diagnostic criteria. Second, Willick (1990) and Shorter (1993) note that there was significant disagreement among analysts regarding the interpretation of psychoanalytic theory and how to differentiate between disorders based on such theories. For instance, many theorists in this era argued that most psychiatric conditions were caused by the same underlying psychological disturbance, and there only appeared to be different conditions based on the manifestation of symptoms, while others argued that differences in symptoms did indicate separate disorders (Mayes & Horwitz, 2005; Shorter, 1993). Third, many disorders now recognized as distinct conditions were classified as subtypes of the same psychiatric disorder, based on the psychoanalytic view that these conditions were the result of similar psycho-sexual causes. Fourth, the first and

second editions of the *Diagnostic and Statistical Manual* reflected the psychoanalytic view that childhood and adult disorders were continuous, and thus did not make a distinction between disorders that appear in childhood and those that appear in adulthood.

These issues often resulted in significant diagnostic problems, which can be clearly seen in the case of autism spectrum disorders. Willick (1990) notes that during the era of psychoanalysis and leading up the publication of DSM-III, there was still much confusion and debate among clinicians regarding whether autism spectrum disorders were a type of schizophrenia, or should be considered separate disorders. Kanner (1943) argued that the condition he identified was a separate disorder, and clinicians such as Mahler (e.g. 1952; 1958) and Bettelheim (1967) recognized ASD as separate conditions. However, Bishop (1989) states many clinicians in this era understood the symptoms of ASD to be indications of childhood schizophrenia, and did not recognize autism spectrum disorders as separate conditions. In this era, the disorders on the autism spectrum were considered to be 'psychotic disorders,' along with adult schizophrenia, based on the psychoanalytic view that ASD and schizophrenia were caused by stalled ego development early in life.

The only diagnostic category that included mention of autism symptoms in the first two editions of the DSM was "schizophrenic reaction, childhood type," which was classified under the larger diagnostic category of "Schizophrenic Reactions." The description presented in DSM-I of "Schizophrenic Reactions," states that the term "represents a group of psychotic reactions characterized by fundamental disturbances in reality relationships and concept formations, with affective, behavioural, and intellectual disturbances in varying degrees and mixtures (p. 26, DSM-I)." The disorders under this grouping "are marked by strong tendency to retreat from reality, by emotional disharmony, unpredictable disturbances in stream of thought, regressive behaviour, and in some, by a tendency to 'deterioration.' (p. 26, DSM-I)." 'Schizophrenic Reactions' included many adult-onset types of what is now recognized as schizophrenia, but only one childhood type. Thus, cases of what are today recognized as autism spectrum disorders would likely be included under the childhood type of 'schizophrenic reactions.'

However, the category "Schizophrenic Reaction, childhood type" did not contain a set of criteria to help clinicians identify cases of childhood schizophrenia or the conditions now known as autism spectrum disorders in the population. Instead, it contained the following description of the symptoms and nature of these conditions:

> Here will be classified those schizophrenic reactions occurring before puberty. The clinical picture may differ from schizophrenic reactions occurring in other age periods because of the immaturity and plasticity of the patient at the time of onset. Psychotic reactions in children, manifesting primarily autism, will

> be classified here. Special symptomatology may be added to the diagnosis as manifestations (DSM-I, p. 28).

Without criteria identifying characteristic, observable symptoms, it was left up to the clinician's interpretation of a child's symptoms as to whether a diagnosis of "childhood schizophrenia" was made, and whether the clinician understood the symptoms of autism as representing a distinct type of psychopathology, as some analysts such as Bettelheim and Mahler did.

DSM-II, published in 1968, also contained categories of disorder that were heavily influenced by psychoanalytic theories of the origins of these conditions, and also did not contain standardized diagnostic criteria. Similar to DSM-I, DSM-II contained a category called "Schizophrenia," which included disorders that were "manifested by characteristic disturbances in thinking, mood and behaviour (p. 33)," and by significant disturbances in the patient's interpretations and interaction with the surrounding environment. However, like its predecessor, DSM-II did not make a distinction between adult and childhood disorders, or between schizophrenia and autism spectrum disorders. Instead, the category of "Schizophrenia, childhood type" in this edition of the DSM contains the following description:

> This category is for cases in which schizophrenic symptoms appear before puberty. The condition may be manifested by autistic, atypical, and withdrawn behavior; failure to develop identity separate from the mother's; and general unevenness, gross immaturity and inadequacy in development... (p. 35).

In many ways, this category's description is more overtly psychoanalytic than its DSM-I predecessor. The DSM-II version actually mentions the role of the mother in the development of the child's ego, which reflects the overall psychoanalytic view that ego development in early childhood depends on the relationship between child and mother. Children exhibiting the characteristic symptoms of autism spectrum disorders could be identified using this description. However, without standardized diagnostic criteria identifying the key features of these disorders, whether or not a child displaying the characteristic symptoms of autism spectrum disorders would receive a diagnosis of childhood schizophrenia was heavily dependent on the clinician's interpretation of the symptoms and the relationships between a child and his or her parents. Further, like its predecessor, DSM-II did not recognize ASD as separate disorders, and thus identifying a child's symptoms as a case of autism still depended on the whether the clinician in question recognized these conditions as separate from schizophrenia.

Before the publication of DSM-III and the shift to a symptom-based diagnostic approach, the emphasis in psychiatric practice during the psychoanalytic era was placed on exploring the developmental histories and parental relationships to understand the origins of the symptoms. The emphasis on patient interviews and understanding the symptoms within the context of early childhood development and familial relations reflects psychoanalytic explanations of these conditions as manifestations of psycho-sexual conflicts (Mayes & Horwitz, 2005; Shorter, 1993). However, without standardized diagnostic criteria, diagnosis was heavily dependent on individual clinicians, which made the diagnosis of disorders such as those on the autism spectrum highly variable and inconsistent, thus resulting in poor reliability.

Further, without standardized criteria based on empirically informed causal theories, diagnosis in the era of psychoanalysis suffered from poor validity as well. The classification system and categories of disorders contained in DSM-I and DSM-II were based on a conceptual framework that had weak empirical foundations and inconsistent theories. Many clinicians and researchers argue that psychoanalytic theories of the causes of psychiatric disorders were not grounded in the larger body of scientific knowledge in medicine (e.g. Mayes & Horwitz, 2005; Shorter, 1993; Poland et al, 1994). Explaining most psychiatric disorders as manifestations of psycho-sexual conflicts does not reflect the underlying biological and cognitive impairments involved in psychiatric conditions like those on the autism spectrum, and does not accurately account for the social features of these conditions, since bad parenting is not a reliable indicator of the development of disorders such as ASD (see Rimland, 1964; Rutter, 1978; Paluszny (1979); Siefert, 1991) for a discussion of the tenuous link between parenting styles and the development of autism spectrum disorders). Thus, basing diagnostic categories on explanations that are empirically inadequate and internally inconsistent reduces the validity and reliability of diagnosis. As discussed in more detail in the next chapter, psychoanalytic approaches to treatment were largely ineffective, since the categories of diagnosis did not accurately reflect the nature of the underlying malfunctions causing the symptoms of conditions such as ASD. Thus, the categories of disorders contained in DSM-I and DSM-II were not based on empirically-supported causal theories, but rather on a body of literature and theory that was still isolated from the related disciplines such as neuroscience and medicine.

A cause-based diagnostic system must be based on explanatory patterns that accurately represent the underlying malfunctions that result in the observable symptoms of psychiatric conditions in order to properly differentiate between these conditions and successfully identify individuals who have them. A causal explanation should accurately represent and identify the underlying malfunction(s), and should provide effective targets for intervention. If a theory cannot be verified by research or by its effectiveness in developing a

treatment approach, it cannot inform valid and reliable diagnostic categories. Therefore, the first criterion an adequate pattern of explanation for psychiatric conditions must meet is *empirical support.* Further, a theory should be *consistent* in both its account of the underlying malfunctions and causal processes, and consistently applied in relevant clinical cases.

However, empirical support can include both the degree to which a causal theory accurately represents the underlying causal processes, and the ability to inform an effective course of treatment. Many causal theories and patterns of explanation in psychiatry and clinical psychology seem to include only one of these features, but not the other. For instance, the presence of the MECP2 mutation in female children explains the presence of the symptoms of Rett's disorder, but it is not clear how, or if, this genetic mutation can be a target for intervention or treatment. Further, behaviourist theories of psychiatric conditions have provided effective treatment approaches for a variety of disorders, including ASD. However, behaviourist theories were not concerned with discovering the underlying causal dysfunctions that cause the symptoms, but rather ameliorating or eliminating them. Ideally, an empirically supported causal theory or pattern of explanation will both accurately represent the underlying causal processes, and provide an effective target for intervention. However, given the highly speculative and 'sketchy' nature of most of the causal theories in psychiatry and clinical psychology, many theories have one aspect of empirical support but not the other. Even if a causal theory has either of these aspects of empirical support, it may still provide information that can help develop cause-based diagnostic categories, and can provide information on how to improve treatment. Thus, in order for diagnosis in psychiatry to effectively identify psychiatric disorders in the population and effectively treat these conditions, diagnostic categories and treatment approaches must be based on *empirically supported, internally consistent causal explanations.*

2.4 Criteria for Adequate Explanation Patterns in Psychiatry

Thus, we can identify the first two criteria for adequate explanation patterns for psychiatric conditions. First, such a pattern should have *empirical support*, since a theory that does not accurately represent the phenomena that it explains may lead to false or inaccurate conclusions. Because of the lack of empirical supported theories, treatments prescribed by analysts, usually psychotherapy to uncover and address psycho-sexual trauma, was largely ineffective. Second, explanation patterns for psychiatric conditions should be *consistent.* As the next chapter discusses, psychoanalytic theories of autism spectrum disorders differed

in both their definitions and descriptions of these disorders. Inconsistent accounts regarding the nature of ASD by psychoanalysts often resulted in the use of therapeutic approaches designed for conditions very different than those on the autism spectrum, including adult schizophrenia and adult anxiety disorders (Willick, 1990). Thus, adequate causal theories of psychiatric disorders like ASD should include a set of concepts that are standardized and adopted across all psychiatric evaluations.

We can also identify a third criterion for adequate explanation patterns for psychiatric conditions. Theorists such as Murphy (2006) argue that in order to increase the explanatory and predictive power of diagnostic categories, these categories need to have good validity. In order to improve diagnostic validity, diagnostic categories need to be more representative and predictive of variables such as onset, progression, and prognosis without treatment. However, the current classification system in the DSM contains many categories that are not significantly related to these variables, and thus the progression and prognosis of most psychiatric disorders is difficult to determine. There are different types of validity, but the type with which Murphy (2006) is most concerned is what he calls *predictive validity*. He states that explanations of psychiatric disorders that have 'predictive validity' are explanations that obtain when one moves from a representation of idealized, or typical, symptoms and course of a particular disorder to the real world, i.e. the explanation is unaffected by variations in patients and their particular environment. Thus, in the case of psychiatry and clinical psychology, explanations of particular disorders that have good predictive validity should be able to account for the symptoms a patient is currently presenting, and will continue to present in the future without treatment. Likewise, an explanation with *predictive validity* will inform what sort of treatment will be effective, given the nature of the particular symptoms from which a patient is suffering.

Murphy (2006) argues that predictive validity of diagnostic categories will be increased if they are based on causal explanations, since causal explanations are better able to identify variables like onset, progression and prognosis. However, generating viable causal explanations of conditions such as those on the autism spectrum is difficult for a number of reasons. In what follows, I discuss the most significant obstacles to generating *valid* and *reliable* diagnostic criteria. As the discussion progresses, I identify and evaluate suggestions by theorists such as Murphy (2006; 2008) regarding both the development of, and problems with, causal explanations in psychiatry, using ASD and other psychiatric disorders as examples. Although autism spectrum disorders are my main case study, the criteria I develop here can potentially be applied to other conditions in the DSM. So far, I have argued that an adequate explanation patterns for psychiatric conditions must be 1) *empirically supported*, 2) *consistent*, and 3) must have *predictive validity*. However, I argue that predictive validity depends on explanation patterns for these conditions meeting

certain criteria, which I identify below.

2.4.1 Causation on Multiple Levels: Representing Interactive Causation in Psychiatric Conditions

Theorists like Murphy (2006) and Boucher (2009) argue that causal explanations of psychiatric disorders should include *primary*, or *robust*, causes. However, one of the most significant obstacles to generating viable causal explanations of psychiatric disorders is the complexity of such conditions. The severity and exact presentation of the symptoms of a particular condition will vary depending on 1) the individual's physiology, 2) features of the individuals' social environment, and 3) how the individual's physiology and environment interact. These causal factors and the interactions between them often mean that patients can have virtually identical symptoms, but very different primary causes for those symptoms.

Consider two patients, both exhibiting the symptoms of major depressive disorder. Each patient is suffering from the characteristic symptoms of major depression: insomnia, lack of appetite, and anhedonia (the inability to take pleasure from things one previously enjoyed). Patient A is in his late teens, is an average student, and had a previously active social and family life before the onset of his symptoms. Patient B is in his late fifties, has a history of steady employment and stable relationships with friends and family, but had lost his spouse a year before seeking clinical assistance for his symptoms. Even though these patients have essentially identical symptoms, the causal factors are different in important ways. In the first case, the causal factors most relevant to the diagnosis may be the genetic and neurological changes that are causing impaired cognition and social isolation. In the second case, patient B may have the genetic disposition toward depression, but the recent traumatic events in his life are likely the most relevant and fundamental cause of his symptoms.

Even though these patients both display the characteristic symptoms of major depression, the exact nature of the etiology behind the onset of their symptoms is different, and the role certain causes play in their respective diagnoses is also different. Thus, adequate causal accounts of psychiatric disorders need to incorporate both social and biological causes, since both are important in the development of psychiatric disorders (Murphy, 2006; Mitchell, 2008a; Woodward, 2008; Boucher, 2009). Further, causal factors in psychiatric disorders are multi-directional and multi-level, which means that causes at different levels of description can influence phenomena and processes at both higher and lower levels. For instance, biological causes such as genetic mutations and brain lesions can affect not just

the cognitive aspects of a certain condition, but also the social aspects, such as a patient's behaviour and social interactions. However, social causes can also influence biological causes, since stressful life events can exacerbate the neurological and cognitive malfunctions that characterize a particular disorder. Thus, the malfunctions that characterize psychiatric conditions can influence phenomena at more than one level of description, and causes at different descriptive levels interact to produce the overt symptoms of psychiatric conditions. In the case of most psychiatric disorders, including ASD, there are multiple causes interacting at the social, cognitive and biological levels of description. However, causal explanations of psychiatric disorders need to be able to include certain causes and exclude others as central or fundamental in the development of a particular disorder in order to be useful in diagnosing that condition. Thus, such an explanation must only what Murphy calls *robust* causes, or in Boucher (2009)'s terms, causes that are *necessary* for the disorder to develop.

Murphy (2006) calls explanations that identify *primary* or *robust* causes *fundamental explanations*. On this account, a *fundamental* explanation of a given psychiatric disorder only identifies the causes implicated in most cases where the symptoms in question are present, and cannot be radically shifted depending on the patient's personal history. Murphy (2006) states fundamental explanations in psychiatry are those that:

1. cite maximally robust causal relations, including relations that cross levels of explanation

2. explain all the symptoms of a condition (p. 141).

A more fundamental explanation is one citing a factor that reliably produces an outcome despite different values for other relevant variables. A less fundamental explanation cites a factor that makes a difference to the outcome only under a very restricted range of circumstances (Murphy, 2006). However, what Murphy calls a 'fundamental level of explanation' or a 'robust cause' changes based on the disorder in question. As Murphy (2006) and others have noted, the types of causes involved in most psychiatric disorders are at different levels of description, such as the social level (which might include causes such as active duty in a warzone, natural disasters, or stressful life events that are associated with the onset of post-traumatic stress disorder or major depression), and the biological level (such as the genetic mutations that cause Huntington's chorea or Down's syndrome). For instance, consider the differences in causal theories for conditions such as anorexia nervosa versus causal theories for ASD. In the case of eating disorders like anorexia, the fundamental causes of these conditions may include social pressures to be thin, a competitive

home or professional environment (such as high parental expectations, pursuing a career in modeling or acting, etc.), or other environmental or social causes, along with certain biological and cognitive features. However, in the case of disorders like ASD, the fundamental causes are thought to include genetic, epigenetic and neurological malfunctions, along with characteristic cognitive and social features. Thus, some psychiatric disorders may have *primary social causes*, such as eating disorders, or *primary biological ones* such as ASD, or potentially *primary causes at both the social and biological level*, which may be the case for most disorders currently identified in the DSM (Kendler, 2008). Thus, patterns of explanation that identify the causes of psychiatric disorders should include those at the cognitive and social levels, and should not necessarily reduce these higher levels to the biological or genetic level of explanation.

Murphy's criteria for fundamental or robust explanations in psychiatry are not meant to be exhaustive. However, they do give us broad parameters with which to work. Adequate explanation patterns in psychiatry must be *parsimonious*, in that they must include only include the robust causes(s) of a particular disorder, and do not include secondary or indirect causes. Since causation of psychiatric disorders is complex, inter-level and multi-directional, we must be willing to include 'social' or 'environmental' causes in acceptable causal accounts in psychiatry. However, psychiatric conditions usually do not have one primary cause, or even a few 'primary' causes. As I argue in chapter five, even disorders that have a strong biological basis, such as ASD, do not (at least as yet) have simple biological explanations attached to them. Thus, while Murphy (2006) and Boucher (2009) are correct that we must identify *robust* or *primary* causes, finding them is no easy feat, which I discuss in more detail below.

2.4.2 Complex Etiologies, Complex Symptomatology: Identifying Primary Causes

Recent research indicates that most psychiatric disorders involve multiple genetic mutations and impairments in cognitive capabilities distributed across many areas of the brain. Further, the symptoms characteristic of many disorders seem to involve an interaction between multiple neurotransmitters and multiple neurological structures. Schaffner (2008) argues that the relation between genes and neuromolecular pathways is many-to-many, rather than one-to-one. For instance, patients diagnosed with major depressive disorder suffer from sleep disturbances, but these disturbances can be manifested as insomnia, or as hypersomnia, where a patient sleeps too much (Murphy, 2006; see also DSM-IV-TR). Also, many disorders listed in the DSM are associated with sleep disturbances, and other

diagnostic criteria for major depressive disorder are shared by other conditions, e.g. psychomotor agitation, changes in appetite, inability to concentrate, etc. Boucher (2009) argues that "for each set of defining behaviours, there are clearly numerous causal factors all contributing to that particular set of behaviours (p. 101)." Thus, expression of phenotypes in the etiology of psychiatric disorders cannot be predicted solely from knowledge of the genes involved. Further, even though individuals within a diagnostic category may have the same genetic or neurological deficits, the exact presentation of the symptoms associated with those deficits will not be the same across those individuals.

Another problem is raised by Woodward (2008). He discusses one of the common 'psychosocial' variables often implicated in the pathogenesis of various psychiatric disorders. 'Low socioeconomic status' is a predicting factor in many of the conditions listed in the DSM, including depression, anxiety, and schizophrenia. A 'macro-level' or coarse grained variable such as low SES does predict a higher probability of developing a psychiatric disorder, but SES does not tell us 1) which psychiatric disorder(s) (out of the set it predicts) are the most likely to develop, 2) why certain individuals in a socio-economic strata develop these psychiatric disorders and others do not, and 3) which direction the 'causal' relation goes.

For instance, as Woodward (2008) asserts, individuals with low SES may have more stressful lives, poorer health care, meager medical benefits, and less stable support systems. Thus, the stresses and difficulties that accompany low SES may contribute to the increased prevalence of psychiatric disorders in this population. However, as Woodward (2008) also states, individuals with pre-existing psychiatric disorders may be forced to take lower-paying and less-desirable jobs, since their conditions may make it difficult to function in jobs with high levels of responsibility or stress. Further, the quality of life for individuals with psychiatric disorders that go untreated tends to decline as the condition progresses and worsens, which can result in financial and social difficulties. Thus, just identifying predicting variables, no matter how 'robust' they seem at first, will still leave much diagnostic guesswork to the clinicians.

Likewise, several genes and neurological impairments are associated with more than one psychiatric disorder, and physiological factors or causes can end up contributing little to a differential diagnosis. Further, a genetic disposition to the neurological changes that are characteristic of a disorder like depression is not the only factor in the development of this condition in adolescence and adulthood. As the discussion above points out, conditions such as major depressive disorder is often also triggered by environmental causes, such as the death of a loved one, hard financial times, or socio-political upheaval.

Using the example of Major Depression, Mitchell (2008a) argues that psychiatric dis-

orders are malfunctions in *interactive complex systems.* She states that disorders like depression have complex etiology, where multiple genetic, neurological, cognitive, personality, and environmental factors contribute to their development. She argues that to understand the development of psychiatric disorders, we need to know not just how each component of the system works, but how the system as a whole adapts to its environment. Based on the complexity of the development of depression and most other psychiatric disorders, theorists such as Murphy (2006), Mitchell (2008a), and Schaffner (2008) argue that *multi-level* explanations of disorders are necessary for a viable causal diagnostic framework in psychiatry. Thus, adequate causal explanations of psychiatric disorders must include *multiple causes* at *different levels* of explanation. However, most psychiatric disorders do not have a stable and predictable progression or onset, because these causes interact to create the cognitive and behavioural disturbances in a particular patient. Thus, adequate causal explanations must also account for the *interactions* between causes.

2.4.3 Generating Mechanistic Explanations and The Lack of Psychopathological Laws

Psychiatric disorders have complex symptomatology generated by causes interacting at multiple levels of description. Thus, generating explanations of psychiatric disorders that will subsume all individuals with that particular disorder is difficult. This section discusses how best to represent these interactive and multi-level causal factors, since the complexity in the development of psychiatric disorders requires a particular type of explanatory strategy. With the exception of disorders like Huntington's chorea and some aphasias, which are caused by a single gene or brain lesion, the relation between symptoms and underlying malfunctions in psychiatric disorders is not a law-like one. Thus, attempting to generate explanations by appeal to 'psychopathological' laws, where certain symptoms always indicate a particular underlying impairment, has limited utility in psychiatry and clinical psychology. Further, many theorists, such as Bechtel (2007) argue that explanations based on laws have little applicability in any of the the natural sciences, including neuroscience and biology.

To generate multi-level explanations, theorists such as Murphy (2006), Mitchell (2008a; 2008b) and Thagard (2008) advocate developing *mechanistic explanations* of psychiatric disorders. Machamer, Darden & Craver (2000) describe mechanisms as "entities and activities organized such that they are productive of regular changes from start or set-up to finish or termination conditions (p.3)." Thus, mechanisms are representations of the parts of certain phenomena, and how those parts interact to generate the phenomena in

question. Mechanistic explanations allow us to account for the regularities seen in certain phenomena, and allows us to discuss causality, even though the phenomena in question are not subsumed under laws of nature.

In the case of mechanistic explanations of psychiatric disorders, the parts include genes, neurotransmitters, neurological structures and cognitive processes. Certain start-up conditions, or inputs, will influence the activity of the parts of the mechanism, and how these parts interact. In the case of psychiatric conditions, the inputs to the mechanism can include social and environmental factors, such as stressful events or aversive stimuli that can influence and exacerbate neurological and cognitive malfunctions. Malfunctions in parts at one level, such as chemical imbalances or breakdowns in neurological mechanisms, can influence the activities of other parts at both higher and lower levels (i.e. the genetic, cognitive, and social levels), and the interactions between these malfunctions produce the characteristic symptoms of a particular disorder. The interactions between the parts of the mechanism, i.e. how genetic, neurological, cognitive and social features influence each other and mutually cause the ongoing progression and presentation of cognitive and behavioural symptoms, explains how each of the aspects of a psychiatric disorder work together to produce the outward symptoms.

On this account, identifying the nature of the malfunctions at different levels, the interactions between these malfunctions, and how certain inputs produce regular changes in the mechanism's parts and activities explains how and why certain clusters of causes are related, and why certain symptoms are associated with particular malfunctions. For instance, a mechanistic explanation of autism spectrum disorders would identify parts such as certain genes involved in neurological and socio-cognitive development, neurological structures such as the mirror neuron system, and cognitive processes such as theory of mind. Breakdowns in these parts, such as genetic and epigenetic mutations, a malfunctioning mirror neuron system, and an underdeveloped theory of mind, interact to produce the characteristic socio-cognitive and behavioural symptoms of ASD: impairments in social interaction and communication, and stereotyped behaviours. Further, determining how genes, neurology, cognitive processes and social development interact to produce these symptoms can help elucidate how the characteristic symptoms and progression of conditions like ASD may manifest in a particular environment. In this way, mechanistic explanations allow us to elucidate the interactions between different variables that regularly produce psychiatric disorders, without having to subsume psychiatric disorders under psychopathological laws. Thus, another criterion for adequate explanation patterns for conditions like ASD is that such patterns are *mechanistic*, as well as *multi-level* and *interactionist*.

Throughout this section, I have argued that adequate explanation patterns in psychiatry are: 1) empirical supported, 2) consistent, and 3) have predictive validity. Predictive

validity for explanations of psychiatric disorders depends upon the explanation being *parsimonious*, which means it includes only *primary causes*. However, these primary causes are often at different levels of description, and thus an adequate explanation usually includes *multiple levels* and explain the *interaction* between primary causes. Finally, a viable causal explanation in psychiatry should be *mechanistic*, since there are no 'psychopathological' laws governing the relationship between the presence of certain genetic, neurological, cognitive and social features indicative of the different disorders listed in the DSM. Thus, an explanation pattern that includes the above features will be the most powerful for generating explanations that are *universal* enough to identify all individuals with a particular disorder, and *specific* enough to identify only those individuals with a particular disorder.

Identifying the differences in the causal structures of psychiatric disorders can help differentiate between these conditions, despite similarity in their symptoms. Given the causal complexity of most psychiatric conditions, multi-level, interactionist mechanistic causal explanations are the most viable way of adequately representing the differences in the causal structure of psychiatric disorders. If an explanation pattern meets the above criteria, explanations generated using that pattern may help to inform diagnostic criteria that are both reliable and valid. By elucidating the causal structures of psychiatric conditions, clinicians will be better able to differentiate and distinguish between disorders based on causal differences and the presence of outward symptoms, which improves diagnostic validity and reliability.

For example, consider the following case. A male child displays repetitive behaviours and language delays, and has a low IQ. These symptoms may indicate that this child has an autism spectrum disorder, such as autistic disorder. However, these symptoms could also indicate that this child has mental retardation. If diagnosis is based on symptoms alone, it can be difficult to determine if this particular child should be diagnosed with an autism spectrum disorder, mental retardation or both (recall mental retardation and ASD can often occur together). However, a multi-level, interactionist mechanistic explanation of conditions like ASD and mental retardation will be better able to distinguish between these conditions. For instance, children with ASD may have patterns of genetic and epigenetic mutations that are distinct from those associated with the development of some forms of mental retardation, which I discuss in more detail in chapter five. Further, children with ASD may have neurological impairments, such as a malfunctioning mirror neuron system, that are different than those seen in mental retardation. Finally, children with ASD have distinct cognitive impairments not seen in children with mental retardation, such as an underdeveloped theory of mind. Identifying these and other causal differences between these conditions reduces the amount of overlap in the diagnostic criteria for ASD and mental retardation, and further sharpens the boundaries of these diagnostic categories.

Thus, multi-level, interactionist mechanistic causal explanations are more likely to identify the underlying malfunctions that are causing the symptoms a patient is displaying, which will increase the validity of diagnosis. Diagnosis will also be more reliable in this case, it will be based on the presence or absence of certain causal features, rather than only on the presence of absence of symptoms. Even if the symptoms change or are atypical, the presence or absence of certain causal features will be what determines a particular diagnosis, which will reduce the problems of diagnostic overlap and over-inclusion in diagnostic categories.

2.5 Psychiatry and Explanation Schemas of Disease

Psychiatric theory and practice have undergone several conceptual changes, and the way in which psychiatric disorders are diagnosed and treated has been influenced by the dominant overall conceptual framework in a given era. Explanations of diseases and disorders, including psychiatric disorders, can be elucidated using explanation schemas (Thagard & Findlay, 2010; Thagard, 1999). Thagard (1999) states that explanation schemas have an explanatory target, which is the phenomenon to be explained, and a particular explanation pattern that includes identifying the causes of the phenomenon in question. On Thagard's account (1999), diseases are represented as causal structures, where

> Symptoms are the observable manifestations of a disease, which can develop over time in particular ways that constitute the expected course of the disease. The symptoms arise from the cause or causes...of the disease. Treatment of the disease should affect the symptom and course of the disease, often by affecting the causal factors that produce the symptoms. (p. 21).

On this account, diseases are conditions with certain causal factors that bring about the characteristic symptoms of a particular disease or disorder. Treatment of diseases reduces or eliminates the symptoms, and thus affects course and prognosis.

Representing diseases in terms of explanatory schemas also helps to explain why some beliefs change regarding the causes of particular diseases. Thagard (1999) gives the extended example of peptic ulcers, now understood to be caused by the bacterium *Helicobacter pylori*. Before Marshall & Warren (1984) published results finding *H. pylori* in almost all subjects with gastritis and peptic ulcers, ulcers were thought to be caused by stress. Bland foods and milk were thought to relieve the symptoms of ulcers (Thagard, 1999). Thus, an explanation of peptic ulcers, based on the theory that they are stress-induced, can be given as follows. A patient has symptoms such as nausea, vomiting, and a burning

sensation in the stomach because he or she is under stress, which causes acid to be secreted, causing the painful symptoms of peptic ulcers. This condition can be treated by ingesting bland foods and milk. We can also put this explanation of the development of peptic ulcers into a schema:

> *Explanatory Target:* Why does a patient exhibit the symptoms of nausea, vomiting, and a burning sensation in the stomach?
>
> *Explanatory Pattern:*
>
> The patient is experiencing a period of stress, which increases the secretion of stomach acid Increased stomach acid causes the symptoms of a peptic ulcer.

However, after Marshall & Watson published their results, another theory was accepted regarding the causes of peptic ulcers. Although initially the bacteria hypothesis of peptic ulcers was not universally accepted among physicians despite the discovery of *H. pylori*, eventually this hypothesis was adopted by the medical community. The explanation schema for the bacteria hypothesis of peptic ulcers is as follows:

> *Explanatory Target:* Why does a patient exhibit the symptoms of nausea, vomiting, and a burning sensation in the stomach?
>
> *Explanatory Pattern:*
>
> The patient's stomach is infected with the *H. pylori* bacterium The presence of *H. pylori* causes the symptoms of a peptic ulcer.

Thagard (1999) argues that hypotheses are accepted or rejected based on their coherence with evidence and other beliefs regarding the phenomenon in question. Thus, the bacteria hypothesis of peptic ulcers was not accepted by many physicians because the findings of Marshall & Watson (1984) did not cohere with current beliefs regarding peptic ulcers, and there was limited evidence to support the bacteria hypothesis. However, as more research was conducted, physicians accepted that *H. pylori* caused the majority of peptic ulcers because this hypothesis explained the evidence better than the stress hypothesis.

Thagard's (1999) analysis of explanatory schemas in medicine can be applied to the field of psychiatry as well. The history of psychiatry reveals several hypotheses regarding the etiology of the various psychiatric disorders, including ASD. By examining both past and present conceptual frameworks and the patterns explanations of autism spectrum

disorders created within these frameworks, we can begin to evaluate whether the various patterns of explanation for ASD developed since Kanner's and Asperger's original papers meet the criteria for adequate explanation patterns for psychiatric conditions discussed above. I adopt Thagard's version of the explanation schema, which I use in chapters three, four, five and six to elucidate explanations of autism spectrum disorders within different conceptual frameworks in psychiatry and clinical psychology. To identify such explanations, I develop schemas based on the way psychiatric disorders *as a set of conditions* are explained within each conceptual framework, and show that the pattern of explanation for ASD developed in a particular framework is an instantiation of that general pattern of explanation. In chapters three, four, five, and six, I show that we can adopt Thagard's method to represent autism spectrum disorders as causal structures, as discussed above with respect to peptic ulcers. In the case of autism spectrum disorders, the symptoms are the impairments in communication and interaction, and stereotyped behaviours. Treatments like behavioural interventions are designed to ameliorate these symptoms, and reduce the problematic behaviours and social difficulties seen in individuals with these conditions.

Unlike some diseases in other branches of medicine, the causes of ASD, as well as other psychiatric disorders listed in the DSM, are not well understood. The lack of causal information regarding psychiatric disorders makes generating an explanation schema like those above more difficult than for conditions like peptic ulcers. However, we can still analyze how these disorders were explained, classified and diagnosed, and discuss the strengths and weaknesses of causal theories of ASD in different conceptual frameworks that influenced such patterns of explanation, classification and diagnosis. Using explanation schemas to identify the different accounts of ASD allows us to evaluate how different patterns of explanation identifying the possible causes of these conditions affect their diagnosis and treatment. By evaluating past and present patterns of explanation for ASD, we can gain insight into what sort of explanation pattern may be the most powerful for elucidating the causes and progression of these conditions, and how explanations generated using these patterns may improve their diagnosis and treatment.

2.6 Conclusion

This chapter discussed the current diagnostic system in psychiatry, and the arguments made by several theorists who are critical of the current approach and hold that psychiatric diagnosis should be based on causal theories. Theorists such as Murphy (2006) state that explanations of the symptoms of psychiatric disorders must include an account of the

underlying biological impairments, and such explanations will not be nomological ones. As this chapter argues, causation in psychiatric disorders is complex, multi-level, and interactive, and thus classifications and diagnostic criteria in psychiatry must reflect the complex and dynamic nature of these conditions and their symptoms. Like these theorists, I argue that a cause-based framework is better than the current symptom-based framework from which to classify and diagnose psychiatric disorders. However, there are several challenges cause-based diagnosis must face. One of the most important obstacle to address is the complexity and heterogeneity of the biological, cognitive, social and developmental features of psychiatric disorders such as ASD. Based on an evaluation of the obstacles to generating causal explanations and discussions by the theorists above regarding how to begin to address these obstacles, I identified criteria that adequate explanation patterns for psychiatric conditions need to meet. Specifically, explanations of psychiatric disorders must be *empirically supported*, be *internal consistent*, and have what Murphy calls *predictive validity*. I argue that patterns of explanation will have increased predictive validity if these patterns are also *parsimonious*, *interactionist*, *multi-level* and *mechanistic*. Explanation patterns that meet all these criteria will be *universal* enough and *specific* enough to correctly identify individuals with the condition in question. Thus, patterns of explanation that meet these criteria may help to improve explanatory power of psychiatric diagnosis by increasing the validity and reliability of diagnostic categories.

Chapter 3

Explanation Patterns For Autism Spectrum Disorder: Psychoanalysis and Behaviourism

3.1 Introduction

The last chapter discussed the methodological and conceptual problems with the current symptom-based diagnostic system in psychiatry. However, the complex, interactive and multi-level nature of causation in the development of psychiatric conditions present significant obstacles to diagnosis and effective treatment. An analysis of the explanation patterns for the disorders on the autism spectrum developed in past and current psychiatric practice can provide insight into how psychiatric diagnosis and treatment can be improved if they are based on adequate causal explanations, and how the diagnosis and treatment of ASD will improve based on such causal explanations. By applying the criteria for adequate explanation patterns in psychiatry developed in the last chapter to different explanation patterns for ASD, we can gain further insight into what sort of explanation pattern might be the most powerful for explaining the complex causation involved in the development of psychiatric conditions. Further, we can analyze why certain patterns of explanation are not powerful enough to explain the development and progression of conditions such as those on the autism spectrum.

Since the publication of Kanner's (1943) paper, there have been a number of theories regarding the causes of autism spectrum disorders. Each explanation pattern has been

influenced by the conceptual and methodological frameworks that have dominated psychiatry and clinical psychology for the last 60 years. An analysis of these schemas reveals the central role that causation in psychiatric conditions and causal explanations of these conditions have always played in the diagnosis and treatment of these disorders. In some cases, casual explanations have played a key role in diagnosis, as in the era of psychoanalysis. However, causal data and theories have always played a role in the classification and diagnosis of psychiatric conditions. As Murphy (2006) and Poland et al (1994) note, while current DSM categories are supposed to be 'atheoretical' with respect to causes, causal data and theories has implicitly influenced the classification of disorders like those on the autism spectrum. Further, even the *absence* of causal data affects the way psychiatric disorders are diagnosed. Examining the relationship between diagnosis of ASD and explanations patterns identifying the possible causes of these conditions throughout the history of psychiatric practice reveals that explanatory patterns unable to accurately represent the complex and interactive causal structure of conditions like those on the autism spectrum limit the validity, reliability and explanatory power of psychiatric diagnosis.

This chapter examines psychoanalytic and behaviourist explanatory patterns of the disorders on the autism spectrum, and evaluates the effect these explanation patterns have had on the diagnosis and treatment of these conditions. I show that both the psychoanalytic and the behaviourist explanation schemas are poor candidates for causal information to include in diagnosis, precisely because of the problems that motivated the field of psychiatry to adopt theory-neutral, symptom-based criteria for the third edition of the *Diagnostic and Statistical Manual.*

3.2 Psychoanalytic Theories of Autism Spectrum Disorders: Refrigerator Mothers

Psychoanalytic theory dominated psychology and psychiatry from the 1940s to the 1970s (Shorter, 1993). Psychoanalysis had a major impact on theories of autism, and perpetuated one of the most well-known explanations of autism spectrum disorders. In this section, I review the psychoanalytic account of the origins of these disorders, also known as the 'refrigerator mother' hypothesis. Psychoanalysts such as Bettelheim (1967) and Mahler (1952; 1958) argued that the rigid behaviours, language deficits, and impairments in social interaction characteristic of ASD were psychological defense mechanisms to protect the infant's developing ego from an emotionally frigid care-giver. The therapeutic interventions recommended by psychoanalysts were designed to 'restart' normal ego development and allow autistic children to progress past the stage of development at which they were fixated.

In what follows, I discuss the development of this hypothesis, and how the 'refrigerator mother' account influenced the diagnosis and treatment of the disorders on the autism spectrum. I argue that psychoanalysis is inadequate as a viable causal account of psychiatric disorders. Psychoanalysis has been plagued with vague concepts, inconsistent clinical findings, and poor empirical support since its beginnings in psychiatry and psychology (Shorter, 1993). Further, the 'refrigerator mother' hypothesis of the origins of autism spectrum disorders has been condemned by many clinicians as both empirically false and damaging to the relationship between children with ASD and their parents. Finally, the lack of empirically verified causal information in the psychoanalytic explanation pattern for ASD made treatment ineffective and did nothing to help clear up diagnostic confusion with respect to these conditions. The lack of consistent and empirically supported causal accounts of psychiatric disorders, along with the lack of a valid and reliable diagnostic approach discussed in the last chapter, make psychoanalytic explanation patterns of autism spectrum disorders untenable.

3.2.1 A Psychoanalytic Explanation Pattern for Psychiatric Conditions

In the psychoanalytic era, there were some conditions understood by analysts to have a biological origin, such as the mental retardation and developmental delays associated with malnutrition and PKU (DSM-I). However, many psychiatric conditions, including the major affective, thought, personality and pervasive developmental disorders, were hypothesized to have "psychogenic" origins, given the lack of known biological causes for these conditions. For instance, the condition Kanner identified, and other cases of what are currently recognized as disorders on the autism spectrum, were some of the many conditions included under the general diagnostic grouping of "disorders of psychogenic origin or without clearly defined physical cause or structural change in the brain (p. DSM-I, p. 24)." The lack of known causes for these conditions prompted theorists in the psychoanalytic era to develop psychological theories of the origin of the conditions on the autism spectrum. Psychoanalytic theories placed the origins of most major psychiatric disorders in early childhood development, and held that these conditions were the result of unconscious psychological conflicts, caused by fixating at a particular stage of ego development. As mentioned in the last chapter, not all analysts recognized autism spectrum disorders as separate conditions from schizophrenia, and the boundaries between cases of childhood schizophrenia and ASD was not always clear. However, several prominent analysts, such as Mahler and Bettelheim, argued that autism was a separate condition, and developed a theory of the origins of this condition based on the overall psychoanalytic conceptual

framework.

According to psychoanalysis, the ego progresses through stages of psychosexual development, where psychological structures and socio-cognitive skills are developed through interaction with the outside environment (Mahler, 1952; 1958; Willick, 1990). Psychoanalysts often described this socio-cognitive development as a progression through stages of object-relations, i.e. understanding the relationship between the ego and the outside world, and argued that the relationship between the child and the mother was vital to progressing through these stages and for proper ego development. Prominent psychoanalyst Margaret Mahler (1952; 1958; Elkisch & Mahler, 1979) outlines the developmental stages of the ego. She states that infants begin in a state of 'normal autism,' where they are unaware of the difference between the outer environment and the inner world of the mind, and do not see the mother or primary care giver as an object distinct from themselves. After this phase of 'normal autism,' infants proceed to a phase of symbiosis, where the mother is seen as an object joined with the infants' developing psychological structures. At this point, infants are able to recognize that their mothers are part of an external reality, but believe that they and their mothers are fundamentally joined as two objects. The separation-individuation phase follows, where children understand their mothers as separate objects from themselves, and begin to differentiate themselves from objects in the external world. Finally, at the end of the separation-individuation phase, children are able to 'integrate' the objects in the world and themselves into a cohesive picture of reality.

In the case of psychiatric illnesses with no known biological cause, psychoanalysts looked to the relationship between the patient and the primary care-giver, particularly the mother. These clinicians hypothesized that it is a disruption in the development of object-relations that characterize the so-called 'psychotic' disorders such as schizophrenia and ASD (Malher, 1952, 1958; Bettelheim, 1967). Willick (1990) states that psychoanalytic theories relied on the developmental timeline above to ascertain the origins of the symptoms of disorders such as schizophrenia, bi-polar disorder, and autism spectrum disorders, all of which fell under the heading of 'disorders of psychogenic origin.' Analysts believed deficient and inadequate parenting on the part of the child's mother causes the child's ego to become fixated at a particular stage of psychological development (Willick 1990). However, since not every case of childhood psychiatric disturbances were accompanied by a neglectful parent, analysts such as Bettelheim, Mahler, and Klein argued that the child only needed to *interpret* the primary care-giver's attention as neglectful or unsatisfying (Willick, 1990). According to analysts like Bettelheim, the infant and young child are able to interpret the motives and unconscious thoughts of the primary care giver, and attribute to that care giver malicious and even murderous intentions (Bettelheim, 1967; Herbert, Sharp & Gaudiano, 2002; Seifert, 1990). Such attributions by children would cause them to fixate

at a particular stage of object relations and psychological development, thus determining which disorder they will manifest in childhood and adulthood.

Mahler (1952; 1958), like other analysts, held that the ego's alienation from, and distortion of, external reality was the pivotal disturbance in 'psychotic conditions' like schizophrenia and autism spectrum disorders. Many psychoanalysts believed that the various psychiatric conditions observed by clinicians were all manifestations of the same psychological disturbance, and the individual's particular unconscious conflicts determined the developmental stage at which the ego becomes fixated, and thus what symptoms were displayed (Shorter, 1993). According to psychoanalysts, conditions like ASD and schizophrenia occur at distinct developmental stages, where the child has a certain understanding of the relationship with the mother, i.e. as an undifferentiated, partially differentiated, or wholly differentiated object. For instance, childhood schizophrenia was thought to be the result of fixation at the beginning phases of the symbiosis stage of psychological development, where infants are unable to differentiate the outside world from the contents of their minds (Willick, 1990; Mahler, 1952; 1958; Elkisch & Mahler, 1979). On the other hand, autism spectrum disorders were thought to be the result of stalled ego development at the earliest stages, i.e. where even distinctions between self and other are not made (Mahler, 1958; Elkisch & Mahler, 1979). Finally, the symptoms of adult-onset psychiatric conditions, such as the deterioration of cognitive and basic life skills in schizophrenia after a period of typical psychological functioning, were interpreted as the patient's regression to an earlier stage of ego development (Willick, 1990). Depending on the stage at which the ego becomes fixated, the child will react to the real or imagined malicious intentions and inadequate care-giving by displaying the symptoms characteristic of an autism spectrum disorder, schizophrenia, etc. (Bettelheim, 1967; Willick, 1990). Thus, for theorists such as Mahler (1952; 1958), what differentiated schizophrenia and ASD was not the presence or absence of core symptoms like hallucinations or delusions, but rather the age of onset. On this account, adult-onset disorders, such as schizophrenia and bi-polar disorder, were thought to be continuous with so-called 'childhood psychoses' such as ASD and childhood schizophrenia, since the same disturbance lay at the root of all so-called 'psychotic disorders' - disrupted object-relations caused by psychological trauma, created by real or imagined inadequate parenting (Willick, 1990; Shorter, 1993).

We can frame the psychoanalytic account of psychiatric disorders as a pattern of explanation. To put this pattern into more formal terms, I adopt Thagard's (1999) version of the explanation schema, discussed in the last chapter. The psychoanalytic account of the development of psychiatric disorders is as follows:

Explanatory Target: Why do certain individuals develop psychiatric disorders?

Explanatory Pattern:

The ego progresses through stages of psychosexual development, where a child develops socio-cognitive skills, such as object relations.

All psychiatric disorders are the result of the same psychological disturbance, i.e. the representation of real or imagined psychological trauma.

The child's experience of real or imagined psychological trauma is caused by inadequate care-giving on the part of the child's mother or primary care-giver.

The child's experience of real or imagined psychological trauma caused by inadequate care-giving stalls the child's ego development.

Stalled ego development causes the child's ego to fixate at certain stages of psychosexual development.

Stalled ego development that occurs at certain stages of psychosexual development results in the type of psychiatric disorder an individual develops.

The symptoms of the patient's psychiatric disorder indicate the developmental stage where he or she is fixated.

Thus, psychoanalytic explanation pattern of psychiatric disorders of 'psychogenic' origin identified poor care-giving as the main cause of the symptoms characteristic of disorders such as ASD and schizophrenia. Since these conditions had no known biological features, the causes of these disorders were thought to result from unconscious psychological conflicts. Since the mother or primary care giver was considered vital for proper ego development through the various stages of object-relations, poor parenting on the part of the mother or care giver must be the reason for the individual's inability to progress through those developmental stages, which causes the symptoms of psychiatric conditions observed in clinical practice.

3.2.2 A Psychoanalytic Explanation Pattern for Autism Spectrum Disorders

The psychoanalytic tradition was the first conceptual framework to generate a theory regarding the origins of autism spectrum disorders. While psychoanalysis used a set of concepts not included in Kanner's and Asperger's original work, psychodynamic theories attempted to explain the same cognitive and behavioural disturbances these researchers identified as the characteristic symptoms of ASD.

While clinicians such as Mahler and Betteheim developed the 'refrigerator mother' explanation of ASD based on the concepts and theories prominent in the era of psychoanalysis, theorists such as Wing (1997) note remarks made in Kanner's (1943; 1949) original papers that hint at a possible psychogenic cause of autistic symptoms, thus influencing psychoanalytic theories regarding the etiology of ASD. In his early research, Kanner proposed a theory that inspired many other theorists to find a psychological cause for the behaviour demonstrated by children with autism. While Kanner suspected that autism may have a genetic component, he also theorized that the emotional 'frigidness' of the child's mother was a major cause of autistic behaviour. Mesibov, Adams & Klinger (1997) state that Kanner was unable to find any clear biological impairments in his original sample of eleven children, and this lack of evidence for a biological cause led him to examine the relationships between the parents and their autistic children. Kanner stated that the parent-child relationships were characterized by 'mechanistic' interactions, obsessiveness and lack of parental warmth (Kanner, 1949; Mesibov et al. 1997). Further, he theorized that children who had this type of interaction with their parents escaped the lack of affection by withdrawing and seeking comfort in solitary activities. Kanner was unsatisfied with, and skeptical about, his psychogenic theory, since many of his children's siblings did not have autism and many children with 'cold' parents are not autistic. However, this hypothesis motivated many clinicians within the psychoanalytic framework to establish a link between unemotional parents (particularly the mother) and autistic behaviour.

Theorists in the psychoanalytic era focused on the difficulty children with autism had with relating to people and objects in their environment. Mesibov et al. (1997) describe the rationale for this type of theorizing as follows: "[i]f children with autism could not relate to others, their repetitive action, language impairments, and other symptoms must...represent a withdrawal from an outside world that they found intolerable (p. 6)." As discussed above, the self-other relationship is a central component of psychoanalytic theories of early childhood development. Mahler's work on children with autism spectrum disorders is based on the psychoanalytic view that the symptoms of autism were a defense mechanism against the emotional unavailability of the mother, which resulted in her inability to assist the child in progressing through the stages of ego development. Mahler argued that in order to understand 'psychosis' in children, one must examine the stage of development at which the mother is not yet, or is about to be, differentiated from the infant's own ego. Since children with autistic symptoms often fail to differentiate themselves and others from inanimate objects, and fail to develop an emotional bond with their mothers, such children must be stuck at the very first rudimentary stages of ego development - the so-called 'autistic phase' (1952; 1958).

Following in this tradition, the most well-known psychoanalytic account of autism was

put forward by Bettelheim in his 1967 book *The Empty Fortress*. Bettelheim's account of the psychological origins of autism adapted and simplified psychoanalytic concepts such as self-other relations and ego development (Nadesan, 2005). He argued that autism was the result of maternal behaviour toward the infant during critical periods of development. Bettelheim rejected the possibility of biological or innate factors that could result in autistic behaviour, and focused solely on the mother's interaction with the child. Like the other psychoanalytic accounts of autism, Bettelheim linked autism with frustrated attempts at individuation and object relations. He argued that these failed attempts would lead to a complete lack of development if experienced in the first year of life, or to incomplete individuation if experienced in the second year of life (Bettelheim, 1967).

Like Mahler and other analysts working in the area of 'child psychoses,' Bettelheim attributed the causes of psychiatric disturbances in children to emotional unavailability and inadequate care on the part of the mother. However, as mentioned above, Bettelheim attributed more than just emotional frigidness to mothers of autistic children - he argued that these mothers held subconscious 'murderous' thoughts towards their children (Herbert, Sharp & Gaudiano, 2002), and wished that their children did not exist (Bettelheim, 1967). On this account, infants were able to ascertain the unconscious motives of their mothers, and retreat into an 'autistic state' in order to escape a reality in which they are cared for in an emotionless, mechanical way (Seifert, 1990; Herbert et al, 2002). Bettelheim (1967) gives his account of the origins of autistic symptoms and behaviour as follows:

> Infantile autism...stems from the original conviction that there is nothing at all one can do about a world that offers some satisfactions, though not those one desires, and only in frustrating ways. As more is expected of such a child, and as he tries to find some satisfactions on his own he meets even greater frustration: because he neither gains satisfaction nor can be do as his parents expect. So he withdraws to the autistic position. If this happens, the world which until then seemed only insensitive now appears utterly destructive, as it did from the start to the child who [fails to thrive in institutions] (1967, p. 46).

Thus, on Bettelheim's account, a child becomes autistic as a defense against inadequate care in the early years of development, that is, as a response to a 'refrigerator mother.' Based on the view that ASD had a psychological origin, psychoanalysts argued that psychotherapy was the way to treat these disorders. Analysts like Bettelheim recommended removing autistic children from their parents' care and placing them in the custody of nurturing care-givers. Many of the theorists of this time assumed that the autistic child would improve once they left the emotionally unavailable mother. Once removed from

the "frustrating" and "cold" maternally-dominated environment, an autistic child would undergo proper individuation and ego development, and proper ego development would ameliorate the symptoms of autism (Bettelheim, 1967).

The 'refrigerator mother' hypothesis of ASD and the diagnostic practices that stemmed from this hypothesis are instantiations of the overall psychoanalytic conceptual framework and the way psychoanalysis explained the origins and symptoms of psychiatric conditions. First, the psychoanalytic tradition held that most psychiatric disturbances could be understood as unconscious psychological conflicts, and thus even conditions now understood to have a strong biological basis, such as ASD, were explained in terms of unconscious psychodynamic causes. In the absence of known biological causes of the disorders on the autism spectrum, theorists like Mahler and Bettelheim developed a causal theory of autism spectrum disorders based on the psychoanalytic view that disorders without known biological causes were the result of real or imagined psychological trauma during early childhood development. Second, the identification of autism spectrum disorders in the clinical population and the diagnosis of these conditions relied on the psychoanalytic interpretation of the relationship between patient and primary care giver, and on the interpretation of the patient's symptoms based on classifications and categories developed by analysts.

We can summarize the refrigerator mother explanation of ASD, and develop an explanation schema like the one above for psychoanalytic theories of the origins of psychiatric conditions. The explanation schema in formal terms is as follows:

> *Explanatory Target*: Why do certain children exhibit stereotyped, rigid behaviours, and impairments in language and social interaction?
>
> *Explanatory pattern*:
>
> A child who has an emotionally unavailable mother will develop impairments in language, interaction, and stereotyped behaviours as a way of coping with the emotional neglect from the mother.
>
> A child will continue to exhibit the characteristic symptoms of autism as a way of protecting the ego from an emotionally frigid environment.
>
> An underdeveloped ego causes the child to be unable to differentiate himself or herself from objects in the world and other people, and the child will not progress through the stages of proper ego development, thus perpetuating the symptoms of autism.

Thus, psychoanalytic account of ASD holds that they are not biological impairments, but are psychological or emotional problems that are environmental in nature. To summarize, the explanation pattern for ASD in the psychoanalytic tradition is based on the

general psychoanalytic explanation pattern for the origins of psychiatric conditions discussed above: the symptoms of ASD, i.e. the impairments in communication, interaction, and language, are a coping mechanism to protect the child's ego from an emotionally unavailable mother. The symptoms of ASD will be ameliorated by removing the child from the mother's care and restarting 'normal' ego development, where the child learns to differentiate himself or herself from others, and moves effectively through the stages of ego development.

3.2.3 Psychoanalytic Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

Shorter (1993) notes that psychoanalytic theory and practice were isolated from traditional medicine and neuroscience, thus largely ignoring biological data that would better explain many of the disorders psychoanalysts attempted to treat. A conceptual framework, like psychoanalysis, that is isolated in both theory and practice provides little possibility of linking psychoanalytic theories with related research in other fields, some of which are vital to autism research, such as developmental psychology and cognitive neuroscience. Further, diagnosis of conditions like ASD based on psychoanalytic explanation patterns is plagued by poor reliability and validity, which limits the predictive and explanatory power of diagnosis in psychiatry. As I argued in the last chapter, diagnostic criteria that have weak reliability and validity fail to overcome the problems of over-inclusion and arbitrary diagnostic boundaries, which are the most significant issues with the current diagnostic and classification system in psychiatry. The psychoanalytic explanation pattern for ASD, while consistent with the overall psychoanalytic framework, is an inadequate explanation pattern for psychiatric conditions. The standard objections to both psychoanalytic theories in general, and psychoanalytic theories of ASD specifically, help to illustrate why a causal explanation like the 'refrigerator mother' hypothesis is untenable. Briefly discussing the flaws in psychoanalytic theory allows us to conclude that a psychoanalytic explanation pattern for ASD does not meet the criteria for adequate explanation patterns in psychiatry.

The psychoanalytic explanation pattern meets some of the criteria for adequate explanation patterns, but fails to meet others. The psychoanalytic explanation is multi-level in a way, in that it implicates the subconscious, the ego, and the environment. This pattern of explanation is also interactionist, since analysts like Mahler and Bettelheim argued that the symptoms of disorders like ASD were the result of interactions between the subconscious of the developing child and the environmental cause of a frigid mother. Further, the psychoanalytic explanation pattern can be represented in a mechanistic way, where

the parts are the child's developing ego, the emotional reactions of the mother, and the interactions between these parts explains the development of the characteristic symptoms of autism spectrum disorders.

However, the psychoanalytic theory had poor empirical support and was internally inconsistent, and thus was unable to correctly identify the primary causes that accurately predict the presentation and progression of autistic symptoms. The fundamental cause identified in the psychoanalytic explanation pattern of ASD was also implicated by analysts like Mahler and Bettelheim in the development of unrelated disorders such as schizophrenia. Cases of ASD being identified by clinicians depended more on the particular clinician's interpretation of the unsystematic and inconsistent psychoanalytic framework, rather than on explanatory and predictive power of the psychoanalytic explanation.

First, as with the overall psychoanalytic conceptual framework and explanation patterns for psychiatric conditions, the 'refrigerator mother' explanation of the disorders on the autism spectrum has poor *empirical support*. Clinicians such as Willick (1990), Leichtman (1990) and Kandel (1999) state that the concepts used in psychoanalytic theories have not been verified by current knowledge of early socio-cognitive development, and do not cohere well with established research in developmental psychology. Further, psychoanalytic accounts of psycho-social development have been heavily criticized for overestimating the capabilities of the developing child, such as Bettelheim's claim that children in the earliest stages of neonatal development are able to ascertain the unconscious, malicious intentions of nursing mothers (Siefert, 1990). Psychoanalytic theories of both normal and pathological socio-cognitive development hold that even very young infants have sophisticated conceptual knowledge and emotional complexity. In order for the development of psychiatric disorders like ASD to occur, children were supposedly able to interpret the actions of their parents in the very earliest stages of ego development. From what is currently known about infant and early childhood socio-cognitive development, such sophisticated conceptual and emotional development does not occur until much later in childhood.

Second, psychoanalysts working in the area of child psychiatry produced accounts that differed significantly in their descriptions of both typical and impaired psychological development (Willick, 1990). For instance, Mahler (1952; 1958) identifies a phase of 'normal autism,' where other clinicians, such as Klein do not recognize this phase of development (Willick, 1990) when placing the beginnings of psychotic illnesses in the developmental stages of the ego. Further, analysts were often inconsistent in the application of concepts like 'psychosis' and 'schizophrenia' to conditions unrelated to both of these categories of disorder, such as ASD. Many analysts failed to distinguish between child schizophrenia, adult-onset schizophrenia, and autism spectrum disorders, regarding these conditions as continuous (Willick, 1990). Thus, psychoanalytic theories of the origins of ASD had poor

internal *consistency*, which makes diagnosis of ASD within the psychoanalytic framework problematic.

Third, the psychoanalytic theories such as the refrigerator mother hypothesis failed to correctly identify the fundamental causes of ASD. The claim that the symptoms of autism were caused by an emotionally unavailable mother received only limited support from studies conducted with autistic children and their parents, and there is little evidence that poor or even abusive parenting tactics are responsible for conditions such as autism or schizophrenia. As Kanner himself noted, not all children with ASD have an emotionally frigid mother, and not all children with frigid parents have ASD. In the years following Kanner's original study, many other clinicians argued that the link between emotional frigidness on the part of parents and autistic symptoms was only weakly substantiated (e.g. Rimland, 1964; Rutter, 1978).

Empirically unsupported, inconsistent and poorly defined theories in this era made the diagnosis of psychiatric conditions highly unreliable. Diagnosis of conditions like those on the autism spectrum in this era was heavily dependent on the clinician's interpretation of the patient's symptoms and whether the clinician recognized ASD as distinct psychiatric disorders, and thus could vary drastically from clinician to clinician. In the case of the diagnosis of an autism spectrum disorder, the clinician would examine and interpret the interaction between the child and the parents, particularly the mother, and identify the characteristic symptoms in the children in question. Whether a child's symptoms were diagnosed as "autistic" depended more on the particular analyst's interpretation of the mother-child interaction, and the identification of 'unconscious' attitudes and feelings on the part of the mother and the child. Thus, characterizing the interaction between mother and child could be the subject of disagreement between clinicians, and if the mother is not seen as sufficiently frigid in the view of the clinician in question, a diagnosis of ASD may not be given. However, interpreting a mother's interaction with her child as emotionally frigid could lead a clinician to interpret ambiguous symptoms as indicative of ASD, when they could be better explained by another diagnosis. For example, a child who exhibits language delays and a limited repertoire of behaviours could be diagnosed as autistic if he or she had an emotionally cold mother, even though this child may be suffering from mental retardation, which is also characterized by limited behaviours and language impairments, rather than ASD.

As well as poor reliability, diagnosis within a psychoanalytic framework also suffered from poor validity, which often results when diagnostic categories do not accurately identify vital information such as the primary causes, onset and progression of the disorder. While the psychoanalytic explanation pattern for ASD is *parsimonious* and attempts to identify a *primary cause*, i.e. refrigerator mothers, simply attributing the symptoms of ASD to

uncaring mothers does not explain 1) the significant variation in the symptoms of ASD across patients and in the same patient over time, and 2) the interaction between the individual and the environment that affects the presentation of the characteristic symptoms of these disorders. Further, psychoanalytic theories of the origins of ASD do not identify a correct *primary cause*, based on empirical evidence since the era of Bettelheim's and Mahler's work that has definitively shown the refrigerator hypothesis to be false. Thus, many causes of ASD were likely misdiagnosed, since the psychoanalytic explanation pattern fails to reliably distinguish between cases of ASD, childhood schizophrenia, and other developmental disorders. Misidentifying the primary causes(s), even an interactionist, multi-level mechanistic explanation will likely misrepresent the nature of the disorder in question, and will be less likely to correctly identify the disorder in the population.

Poor validity also occurs when explanations of psychiatric conditions do not accurately differentiate between categories of disorder. In both DSM-I and DSM-II, which were based on psychoanalytic concepts and theories, ASD were classified as a subtype of schizophrenia, and was not distinguished from childhood schizophrenia. In the psychoanalytic era, autism spectrum disorders were included under the broader classification of 'psychotic disorders,' which included unrelated illnesses such as schizophrenia and bi-polar disorder. Classifying ASD and schizophrenia as types of the same disorder does not reflect the nature of the underlying pathology or characteristic symptoms of autism spectrum disorders, which have biological, cognitive and social components significantly different from those seen in schizophrenia. Thus, the psychoanalytic explanation pattern for ASD was too *universal*, in that it identified many other individuals who in all likelihood do not have an autism spectrum disorder, and also included ASD into a diagnostic group with unrelated disorders. Likewise, the psychoanalytic explanation pattern was not *specific* enough to identify only those individuals with an autism spectrum disorder.

Because of the problems in the psychoanalytic explanation pattern for psychiatric conditions, treatment of these conditions in the era of psychoanalysis had very limited success. As just discussed, the psychoanalytic explanation pattern for these conditions did not accurately represent the underlying malfunctions causing the symptoms of conditions like those on the autism spectrum, and thus psychoanalytic treatments were unable to repair or address these malfunctions. Further, since many analysts argued that most psychiatric conditions were the result of the same psychological disturbance, psychoanalysts treated almost all psychiatric disorders with the techniques used during psychotherapy, including free association and discussion of childhood relationships and events (Willick, 1990). However, psychoanalytically-based psychotherapy has limited success in most cases of ASD, since the impairments in language and social interaction make traditional techniques like free association impossible. Finally, the treatment prescribed by theorists like Bettelheim,

i.e. removal from the 'refrigerator mother,' failed to ameliorate the symptoms of ASD. The treatment of ASD that is prescribed based on psychoanalytic explanations for these disorders is ineffective, which also undermines the explanatory and predictive power of psychoanalytic explanations of autism spectrum disorders, such as the refrigerator mother explanation. Unlike children who are neglected or abused, autistic children show little improvement once they are removed from their homes (Paluszny, 1979).

While the psychoanalytic framework was the dominant conceptual framework in psychiatry and psychology, and the psychoanalytic explanation pattern meets some of the criteria, the psychoanalytic explanation pattern is not able to adequately explain and predict the onset and progression of psychiatric conditions such as ASD. The psychoanalytic explanation pattern could be represented in terms of *interacting, multi-level mechanisms*, and thus meets some of the criteria outlined in chapter two. While the psychoanalytic explanation pattern is parsimonious, this pattern misidentifies the *primary causes* of conditions like ASD, and is unable to reliably distinguish between ASD and disorders with similar symptoms. Further, the psychoanalytic explanation pattern lacks *empirical support* and *internal consistency*. Finally, this pattern of explanation is *too universal*, and *not specific enough* to accurately identify individuals with ASD. For these reasons, a psychoanalytic pattern of explanation is inadequate for explaining the complex causation in the development and progression of psychiatric conditions.

3.3 The Rise of Behaviourism and Behaviour Modification

Following the reign of psychoanalysis, behaviourist approaches dominated clinical psychology and psychiatry well into the 1970s (Riesman, 1991). From the beginning, behaviourists were critical of the psychoanalytic approach to diagnosing and treating psychiatric disorders, highlighting the limited effectiveness of psychoanalytic therapies for most of the major psychiatric conditions, such as schizophrenia, ASD and bi-polar disorder (Shorter, 1993; Eysenck, 1985). Although behaviourism and behaviour modification became popular in the later half of the 20th century, behaviourist psychology and behaviourist discussions of psychiatric disorders date back as early as the 1910s and 1920s. In a 1916 paper called "Behavior and the concept of a psychiatric disorder," behaviourist pioneer John B. Watson critiqued Freudian theories of the origins of psychiatric disorders, and discussed the role of habit and behavioural contingencies in the development of psychiatric conditions. Continuing in that tradition, modern theorists such as Eysenck (1985) argue that behavioural

therapy based on the principles of learning theory is more effective than psychotherapy in both the short and long term.

3.3.1 A Behaviourist Explanation Pattern for Psychiatric Conditions

In his 1916 paper, Watson highlights the distinction made by analysts such as those discussed above regarding disorders with known biological causes, and those without known biological causes. He states that physicians and clinicians in his time differentiated between conditions with biological causes and those that were "purely mental (p. 589)." As discussed above, many of the major psychiatric disorders were thought to be "purely mental" and attributed to unconscious psychological conflicts. However, Watson (1916) is skeptical of psychoanalytic concepts of psychiatric disorders, and states "...I began to attempt to formulate my own ideas as to the terminology I should use in describing a mental disease...I am strengthened in this attempt to give my concept of mental diseases by the difficulty I have had in understanding the terminology...of the psychoanalytic movement (p. 589)." While he argues that there is some genuinely valuable insight about psychiatric disorders contained in the psychoanalytic account, Watson maintains that there is a simpler and more scientific way to define these conditions.

Watson's (1916) theory of psychiatric disorders retains the core insight from psychoanalysis, which is that childhood experiences can shape the way we think and act as adults, often resulting in maladaptive habits and patterns of behaviour. He states "[t]he central truth that I think Freud has given us is that *youthful, outgrown, and possibly discarded habit and instinctive systems of reaction can and possibly always do influence the functioning of our adults systems of reactions, and influence to a certain extent even the possibility of our forming the new habit systems which we must reasonably be expected to form* (1916, emphasis in original, p. 590)." In many cases, "some of these [childhood habits] yield with difficulty and we often get badly twisted in attempting to put them away, as every psychiatric clinic can testify (p. 591)."

Further, Watson believed that many of the disagreements between clinicians regarding the meaning of psychoanalytic terms could at least be partially resolved by making reference to behaviour and using behaviourist terminology. He states

> [t]he implication is clear that in the psychoneuroses I should look for *habit disturbances* -maladjustments- and should attempt to describe my findings in terms of the inadequacy of responses, of wrong responses, and of the complete

> lack of responses to the objects and situations in the daily life of the patient. I should likewise attempt to trace out the original conditions leading to maladjustment and the causes leading to its continuation. To these statements most psychopathologists will subscribe, but most of them will insist that maladjustments can not be stated wholly in behavior terms (1916, emphasis in original, p. 591).

Thus, behaviourist accounts of psychiatric conditions differ greatly from psychoanalytic approaches, and attempted to move explanations of psychopathology away from unmeasurable, unseen psychodynamic causes.

Later behaviourists would continue to explain psychiatric conditions in terms of maladaptive stimulus-response pairings, and argued that the most effective treatment of these conditions is to alter those stimulus-response pairings. For instance, Eysenck (1985) argues that therapy based on the principles of Pavlovian extinction is the most effective treatment for conditions like obsessive-compulsive disorder. By changing the patient's environment, the stimuli to which the patient is exposed are altered, thus changing the nature of the responses. He states that behaviourists understand phenomena like anxiety and obsessions to be *the result* of the patient's inappropriate responses to stimuli, rather than the cause of the patient's reactions to particular stimuli. Psychoanalytic explanations of obsessive-compulsive disorder would likely implicate memories from the patient's childhood, unresolved feelings towards parental figures, and the feelings and beliefs the patient is repressing. However, Eysenck (1985) argues that psychoanalytic approaches were ineffective in reducing the intensity of the obsessions and compulsions, and the frequency with which they were experienced by the patient.

On the other hand, the behaviourist account of obsessive-compulsive disorder states that the fear and panic characteristic of this condition results from associating the object of the obsessions with feelings of anxiety or distress, and associating the compulsions that follow the obsessions with relief from anxiety. Over time, these associations get reinforced and strengthened by continued pairings, finally resulting in extreme anxiety anytime the subject of the patient's obsession is near. By focusing on how symptoms of psychiatric conditions are triggered, behaviourist clinicians were able to reduce the severity and frequency of these symptoms by changing features of the patient's environment. We can frame the behaviourist explanation of psychiatric conditions using an explanation schema, as we did for the psychoanalytic explanation of psychiatric conditions. The behaviourist explanation pattern is as follows:

> *Explanatory Target*: Why do certain people develop the symptoms of psychiatric disorders?

Explanatory Pattern:

A patient develops the characteristic symptoms of psychiatric disorders because of maladaptive stimulus-response pairings and conditioning.

Maladaptive behaviours, like the symptoms of psychiatric disorders can be extinguished by not reinforcing these behaviours, and reinforcing other, more desired behaviours.

The symptoms of psychiatric disorders can be ameliorated by altering the response patterns to particular external stimuli.

We can compare this explanation pattern for psychiatric conditions with the psychoanalytic explanation pattern discussed above. Like psychoanalytic theories, behaviourist theories of psychiatric disorders maintained that the symptoms of psychiatric conditions were a reaction to events in the external environment. However, unlike psychoanalytic theories, which held that psychiatric disorders were the result of maladaptive ego development, behaviourism understood most psychological and behavioural problems to be the result of external stimuli (Siefert, 1990). On the behaviourist account, it was not necessary to posit internal states that cause behaviour, since what causes behaviour are external stimuli. Thus, if a subject's social learning was altered, and the reactions to various stimuli changed, negative behaviour could be extinguished and new, positive behaviour could be put in its place. The connection between manipulating external stimuli and altering the presentation of the symptoms of psychiatric disorders like phobias, anxiety disorders and ASD is the central aspect of behavioural therapy to treat these conditions.

3.3.2 A Behaviourist Explanation Pattern for Autism Spectrum Disorders

A contrast can be seen in psychoanalytic and behaviourist explanations patterns for autism spectrum disorders. Behaviourist theories of autism spectrum disorders provide an interesting counterpoint to psychoanalytic theories, cognitive theories and neuroscientific theories of these conditions. Clinicians in the behaviourist tradition held that problematic behaviours like those seen in children with autism spectrum disorders were the result of conditioning, and that problematic behaviours could be reduced by altering that conditioning. Like the other theories discussed, behaviourist theories identified ASD by their characteristic symptoms. However, Paluszny (1979) notes that behaviourist theories of autism treatment approaches were not aimed, as psychoanalytic theories were, at discovering the reason autistic children exhibited the symptoms of autism. Instead, behaviourist

theories were aimed at teaching autistic children to overcome these symptoms to improve their social interactions and language. Thus, a behaviourist account is not a full explanatory account of autism spectrum disorders, but does contain a description of the three characteristic symptoms of ASD, a treatment for these conditions, and way to generate prognosis. We can generate an explanation schema for the behaviourist account of autism spectrum disorders, based on behaviourist theories of psychiatric conditions, and based on the research regarding the treatment of these conditions using behaviourist techniques.

In the 1960s, when psychoanalytic theories were still popular, Ferster (1961) made an important connection between learning theory and the symptoms of autism spectrum disorders. He argued that the behaviour of autistic children was maintained by external reinforcement, and could be controlled through behaviour modification. Ferster (1961; Ferster & DeMyer, 1961) stated that the way to alter a child's behavior was to eliminate the consequences the child receives for behaving in a particular way, and reinforcing different behaviours that are to replace the old ones. Behaviourists were unconcerned about the possible cognitive or biological features of ASD, and behaviour modification became the therapy of choice for children with autism. This therapy, like all behaviourist approaches, was based on learning theory, and focused on the child's reactions to external stimuli.

In behaviour modification, a therapist identifies the behavior patterns to be changed, engages in reinforcing the desired behaviour, and extinguishing undesired behaviours. In the case of autism, these patterns could be social withdrawal and self-injurious acts, such as banging the head. Paluszny (1979) outlines the steps in this form of therapy as follows. First, the therapist must define the symptoms to be modified, either to eliminate a particular behaviour or to produce one. Second, the therapist identifies the cues and stimuli that produce the behaviour of interest. Third, the goals and plan for the therapy are outlined. Finally, the therapist uses positive and negative reinforcement, in gradual steps, to guide the child's responses and produce the desired results. Behavioural interventions based on applied behaviour analysis have been shown to be the most effective treatments for disorders on the autism spectrum. Unlike the treatments prescribed by psychoanalysts, autistic children who receive 40-plus hours a week of applied behaviour analysis for a period of at least two years can show improvements in IQ scores, language, social interaction, and can be placed in a regular classroom when they reach school age (Lovaas, 1987; 1993).

Behaviourist theories emphasize the role of the environment in the generation, severity, and progression of the symptoms of ASD, and while rejecting biological or cognitive explanations of disorders like those on the autism spectrum. While not an explicit causal explanation, behaviourists argued that the symptoms of ASD were the result of learning to generate a particular response to external stimuli. Thus, the behaviourist explanation pattern for autism spectrum disorders holds that the characteristic symptoms of these

conditions arise as the result of maladaptive stimulus-response pairings, i.e. pairing problematic behaviours with particular external stimuli. These symptoms can be ameliorated by altering the conditioned responses to these stimuli. For example, a child who bangs his head against a wall when he becomes upset at sudden changes to his external environment can be conditioned to refrain from banging his head when these changes occur.

We can represent the behaviourist explanation of autism spectrum disorders as an explanation schema, which is as follows:

> *Explanatory Target*: Why do certain children exhibit rigid and stereotyped behaviours, and impairments in language and social interaction?
>
> *Explanatory Pattern*:
>
> A child exhibits the characteristic symptoms of autism spectrum disorders because of maladaptive stimulus-response pairings and conditioning.
>
> Maladaptive behaviours like banging of the head or limited patterns of social interactions can be extinguished by not reinforcing these behaviours, and reinforcing other, more desired behaviours.
>
> The characteristic symptoms of ASD, like limited patterns of social interaction and tantrums, can be ameliorated by altering the response patterns to particular external stimuli.

Recognizing the importance of the patient's environment provided a key insight into the development of applied behavior analysis, which remains the most effective treatment for ASD. However, while behaviourist approaches had more effective treatments, behaviourist explanation patterns for psychiatric conditions are too general and simplistic to effectively differentiate between categories of these disorders, and limit diagnostic reliability and validity.

3.3.3 Behaviourist Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

This explanation pattern for autism spectrum disorders, like the psychoanalytic explanation pattern just discussed, is consistent with the larger behaviourist conceptual framework, and is an instantiation of behaviourist theories of psychiatric disorders. Behaviourists held that all behaviour, including the disordered and erratic patterns of behaviour seen in psychiatric disorders, is the result of external stimuli. Theorists like Eysenck (1985) and

Ferster (1961) acknowledge the complexity of psychiatric disorders, but argue that the symptoms are largely determined by the external environment. The symptoms of ASD could be effectively treated using behaviourist techniques, and so these conditions must share the same general characteristics of psychiatric conditions, and are thus explained in the same way. Since behaviourist therapies were more effective than psychoanalytic therapies, behaviourists like Eysenck argue that understanding psychiatric disorders as the result of stimulus-response pairs is a better explanation than what was provided by psychoanalysts.

Despite further conceptual changes in both psychiatry and clinical psychology, behavioural interventions have remained one of the most popular treatments for autistic children, since this form of therapy is still the most successful in ameliorating the symptoms in children with autism (Mesibov et al. 1997; Paluszny, 1979). Effective treatment provides an important piece of information about ASD: the characteristic symptoms, while difficult to treat, are affected by changes in the environment. Thus, one of the factors that influences the severity and chronic nature of ASD's characteristic socio-cognitive symptoms is the environment with which a patient interacts. Like the psychoanalytic explanation pattern just discussed, the emphasis behaviourists placed on the external environment highlighted the social aspects involved in the development and maintenance of disorders like those on the autism spectrum.

Behaviourist theories give a more *consistent* account of the characteristic symptoms of ASD. Further, the treatments prescribed by behaviourists have *empirical support* in that they provide effective treatment approaches for ameliorating the severity of the symptoms of ASD. Behaviourst approaches are interested in treating and managing the symptoms, rather than elucidating the underlying causal mechanisms that potentially differentiate particular psychiatric disorders. However, the behaviourist explanation patterns for these conditions fare no better than the psychoanalytic explanation pattern in differentiating ASD as a distinct set of psychiatric conditions. Both the psychoanalytic explanation pattern for autism spectrum disorders and the behaviourist explanation pattern for these conditions reduce the potentially radically different causal history and pathology of different psychiatric disorders to one primary cause: psycho-sexual conflict and maladaptive stimulus-response pairings respectively. On the behaviourist view, ASD are much like other conditions listed in the DSM, in that they have the same 'cause' and can be treated in the same or similar manner. However, the behaviourist explanation pattern for autism spectrum disorders do not clearly differentiate these conditions from other psychiatric disorders. Social or environmental factors are identified in causal theories for many psychiatric disorders, and explaining the presence of symptoms as maladaptive stimulus-response pairings does not explain the significant variability in the type, onset, severity, and clustering of

symptoms across individuals diagnosed with psychiatric disorders.

Without more detailed causal information, diagnostic categories based on behaviourist explanations of psychiatric disorders have poor diagnostic validity. Diagnostic categories of psychiatric disorders based on behaviourist theories do not effectively distinguish between ASD and other disorders of childhood, or conditions dissimilar to ASD, such as depression or anxiety. The diagnostic category of *autism spectrum disorders* defined in behaviourist terms does not accurately identify cases of ASD because the symptoms of these conditions are also found in other disorders of childhood. Thus, the behaviourist explanation pattern for ASD, like the psychoanalytic explanation pattern, is too *universal*, since it does not reliably identify individuals with ASD versus another psychiatric condition. Because behaviourist accounts of ASD did not clearly differentiate these conditions from other illnesses, even if they could be treated the same way, the behaviourist explanation pattern is not *specific* enough to reliably identify only those individuals with ASD.

Thus, the behaviourist explanation pattern for psychiatric disorders ends up only providing an account of how to mitigate the symptoms of these conditions, rather than an account of the breakdowns of underlying mechanisms that cause the various behaviours that characterize the different psychiatric disorders. However, merely treating the symptoms of disorders like those on the autism spectrum may be ineffective in the long run, since behaviourist therapies such as ABA and other behaviour modification techniques were not developed to treat autism particularly, but as an explanation of psychiatric conditions in general. The problem with not differentiating between conditions that are potentially radically different is that the same treatment will have varying effectiveness, since the underlying pathology of each condition is different. Using the same treatment for most psychiatric disorders misses important features of the underlying impairments in each condition. Further, effective treatment of symptoms does not necessarily indicate the underlying malfunctions that cause them, nor does effective treatment of a cluster of symptoms necessarily indicate that these symptoms are caused by the same, similar or even related underlying malfunctions. While the symptoms of ASD can be reduced by applied behaviour analysis, a behaviourist explanation pattern as outlined above does not provide a way to differentiate the symptoms of ASD from similar symptoms characteristic of other disorders of childhood, such as mental retardation. Further, most psychiatric conditions, including conditions unrelated biologically to ASD such as depression, phobias, and post-traumatic stress-disorder, respond to the techniques of behaviour modification. Behaviourist treatments of autism spectrum disorders are effective, but not necessarily because behaviourist explanations are better at identifying these conditions in the clinical population, but because these treatments work on most psychiatric conditions.

Psychological behaviourism, like logical behaviourism, is a positivist view. Thus, the

behaviourist account of ASD cannot be mapped onto a *mechanistic* representation. The behaviourist view identifies the important role the environment can play in the progression and severity of the symptoms of ASD, a key insight that helped develop behavioural therapies, such as applied behaviour analysis (ABA) that are still used in the present day. However, a behaviourist explanatory patterns of conditions like those on the autism spectrum cannot fully account for changes in the progression and presentation of symptoms as a child ages, or why the presentation of symptoms changes even if the environment does not. First, this account does not explain how the more 'cognitive' aspects of these disorders, such as language impairments and the inability to relate to others as intentional objects, influence the outward behavioural symptoms. Second, the behaviourist account of ASD is unable to explain the neurological and genetic abnormalities found in individuals with these disorders, which also determine the presentation and severity of the characteristic symptoms of these conditions. While behaviourist theories were not intended to explain these features of ASD, the neurological, cognitive and genetic aspects of these disorders are vital pieces of their explanations. Thus, the behaviourist account of ASD does explain some of the *interactions* between symptoms and environment, the behaviourist explanatory pattern is too simplistic to fully capture the complexity of the conditions on the autism spectrum since it does not include causes from *multiple levels*. Diagnostic categories based on behaviourist explanation patterns for psychiatric disorders provide little predictive or explanatory power, and thus behaviourist explanation patterns are inadequate for explaining the development and progression of psychiatric conditions. By concentrating solely on the environmental causes of the symptoms of these conditions, a behaviourist explanation pattern for psychiatric disorders like ASD suffer from the same shortcomings as the psychoanalytic explanation pattern, although for very different reasons.

While the behaviourist schema has good *empirical support* and *internal consistency*, it does not meet all the criteria for adequate patterns of explanation for psychiatric conditions. The behaviourist pattern of explanation is not *mechanistic*, and does not include *causes at multiple levels*. Further, although the behaviourist pattern provides important insight into the environmental factors that influence the presentation and progression of the symptoms of particular psychiatric disorders, is unable to fully explain the *complex, multi-level interactions* between the biological, cognitive and social aspects of these conditions. Finally, even though the behaviourist pattern of explanation identifies a *primary cause*, the pattern is *too universal* and *not specific enough* to accurately demarcate the boundaries between psychiatric conditions and identify these conditions in patients.

3.4 Conclusion

This chapter examined two historical explanation patterns for the symptoms characteristic of autism spectrum disorders. As in the case of the psychoanalytic explanation pattern for ASD, the symptoms of autism were what generated theory construction and treatment procedures in the behaviourist framework, as they continue to do in present-day cognitive, cognitive neuroscientific, genetic and epigenetic theories. Each theoretical change influences what an explanatory account of ASD includes, such as the contrast between psychodynamic causes and environmental causes emphasized by psychoanalysts and behaviourist clinicians respectively. Both of the explanation schemas patterns discussed in this chapter have had significant impact in the history of the diagnosis and treatment of autism spectrum disorders. The psychoanalytic explanation pattern for the conditions on the autism spectrum is one of the most famous accounts of these disorders. The theory that these conditions are caused by poor parenting remains one of the most common and enduring myths about these disorders, and continues to impact lay concepts of these disorders to the present day. The emphasis placed on the external environment by behaviourist clinicians when explaining these conditions led to an approach to treatment that is still the most effective in ameliorating the symptoms of these conditions. However, neither of these explanation patterns are adequate for explaining the development and progression of psychiatric disorders like those on the autism spectrum, and limit the reliability and validity of diagnosis of these conditions.

The psychoanalytic explanation pattern suffers from poor empirical support and inconsistent theories among other weaknesses, and thus is unable to accurately identify cases of ASD in the clinical population and predict the progression of the characteristic symptoms. The behaviourist explanation pattern for these conditions produced an effective means of treatment, but without more detailed causal information, this pattern of explanation is too simplistic and general to generate reliable and valid diagnostic criteria. The behaviourists emphasized the role of the environment in the etiology of psychiatric disorders, an important insight still influencing currents treatment of psychiatric condition. However, the biological and cognitive aspects of psychiatric conditions, including ASD, are vital pieces of their explanations, and thus without these aspects an explanation pattern for psychiatric disorders is inadequate. While still treated using Applied Behavioural Analysis and the techniques of behaviour modification developed in the behaviourist era, explanations of the potential cognitive, neurological, genetic and epigenetic dysfunctions responsible for the symptoms of ASD have been developed in recent years, which the next two chapters discuss.

Chapter 4

Explanations Patterns for Autism Spectrum Disorder: Cognitive Psychology and Cognitive Neuroscience

4.1 Introduction

This chapter discusses two current explanatory patterns for autism spectrum disorders. The first explains the characteristic symptoms of ASD in terms of malfunctions in socio-cognitive processes, and the second is based on data from cognitive neuroscience. While cognitive explanations of ASD remain the most widely adopted theoretical views, these cognitive explanations have also been expanded and refined by theories and research from cognitive neuroscience and neuropsychology. I discuss cognitive views of autism and the cognitive explanation pattern, and then discuss the cognitive neurological explanatory pattern of the conditions on the autism spectrum. I evaluate these explanation patterns using the criteria for adequate explanation patterns in psychiatry discussed in chapter two. I argue that while these accounts seem to be promising candidates for explanation patterns from which to develop valid and reliable diagnostic categories, the cognitive and cognitive neurological patterns of explanation for autism spectrum disorders may not resolve the problems in psychiatric diagnosis discussed by theorists such as Poland et al (1994) and Murphy (2006).

The cognitive explanatory pattern for autism spectrum disorders I discuss below has

been developed by psychologists, rather than psychiatrists. Clinical psychology differs from psychiatry in several respects, including the theoretical and conceptual systems in each field, and the approach each field takes to the diagnosis and treatment of psychiatric disorders. In general, psychiatry employs a more biological approach when explaining and treating psychiatric disorders. The development of the first effective psychoactive drugs in the middle of the last century allowed psychiatrists to take a more pharmacologically-oriented approach to the treatment of psychiatric disorders, replacing psychoanalytic and behaviourist theories (Shorter, 1993). However, the cognitive revolution replaced behaviourism as the dominant conceptual framework in psychology during the 1960s, and has shaped both theory and practice after the era of behaviourism. Many aspects of behaviourism remained in cognitive psychology, such as the emphasis on empirical research and behavioural therapy as a way to treat the conditions listed in the DSM.

4.2 A Cognitive Explanation Pattern for Psychiatric Conditions

In the cognitive era in psychology, clinicians began to re-examine the psychological origins of the dysfunctional behaviours seen in psychiatric disorders. Cognitive psychologists argue that the mind is an information processor, where mental structures organize and interpret information learned and assimilated throughout our developmental history and social interactions. Cognitive theories are developed in psychiatry as well, such as Beck's (1967) cognitive approach to explaining and treating disorders such as depression and schizophrenia. Cognitive explanations of both typical socio-cognitive processes and psychiatric disorders often refer to 'schemas,' which include beliefs and patterns of reasoning that influence how an individual processes information, and interprets the external world. In a seminal work, Kelly (1955) argued that phenomena like anxiety, depression, and paranoia were the result of the patient's 'construction of reality' (Leahy, 1996). Beck's famous cognitive model of depression states that patients often attribute their failures and hardships to internal factors, such as lack of talent, ability, or character, rather than attributing such failures to factors beyond their control (see Beck, 1967, 1972; Beck, Rush, Shaw, & Emery, 1979). According to this theory, depressed patients distort information from the external world, emphasizing negative aspects of situations, and reinforcing negative self-images and beliefs.

Thus, cognitive explanations of psychiatric disorders usually focus on the way the patients interpret information, and what sort of attributions patients make regarding themselves and others. We can generate an explanation schema, like those discussed in the

last chapter, to elucidate a cognitive explanation pattern for psychiatric conditions in the following way:

> *Explanatory Target*: Why do certain people develop the symptoms of psychiatric disorders?
>
> *Explanatory Pattern*: Individuals exhibit the symptoms of psychiatric disorders because they have dysfunctions in cognitive processes, such as the generation and implementation of attributions and schemas.
>
> Attributions and schemas are the cognitive processes by which individuals process information from the external world and develop beliefs and affective judgments.
>
> Individuals with impairments cognitive processes such as the generation and implementation of attributions and schemas, develop beliefs and affective states that are maladaptive and interfere with social interaction and typical cognitive functioning.
>
> These maladaptive beliefs and affective states cause the characteristic symptoms of psychiatric disorders.

Thus, the cognitive explanation pattern for psychiatric conditions identify the cognitive processes that become impaired in these conditions, and explain the outward symptoms in terms of these cognitive deficits (Leahy, 1996). Even with the shift from behaviourism to cognitive psychology, behaviour modification remained a popular treatment regime, but in the cognitive conceptual framework, such therapy included an emphasis on interpreting and integrating information, not just extinguishing behaviours and replacing them with new ones (Mesibov et al. 1997). This type of therapy begins by identifying cognitive and affective distortions or impairments, and generating ways to change patterns of thought and behaviour to mitigate or reduce the symptoms caused by these impairments. Unlike traditional behaviourist interventions, cognitive behavioural therapy tailors the process to the individual's particular symptom presentation and progression, since these symptoms and their severity differ from patient to patient.

4.2.1 A Cognitive Explanation Pattern for Autism Spectrum Disorders

Cognitive theories of autism spectrum disorders also focus on information processing and impairments in cognitive processes. One major cognitive theory of ASD that has been put

forward is the theory of mind hypothesis. This hypothesis was first developed by Baron-Cohen and his colleagues in the 1980s. This hypothesis assumes that the way individuals with ASD process social information is different from the way typically developed individuals do. In particular, proponents of this view state that the cognitive deficits unique to autism spectrum disorders are the result of an undeveloped theory of mind, i.e. the ability to ascribe mental states to others, such as beliefs and desires, to explain their actions. Theory of mind enables us not only to explain the behaviour of others, but also to detect deception, humour, sarcasm and irony (Baron-Cohen, 2000). Baron-Cohen and other researchers argue that impairments in theory of mind occur early in childhood, and that theory of mind deficits are universal, i.e. an autistic individual can do none of the above things, or can do them in a very limited capacity.

Several tests have been used to investigate the theory of mind capacity in children with ASD. Many of these tests require that an autistic child ascribe beliefs, particularly false beliefs, to characters in vignettes presented to them (see Baron-Cohen et al, 2000 for a discussion of these tests and their results). Baron-Cohen (2000; also see Baron-Cohen, Leslie, & Frith, 1985) states that children with an autism spectrum disorder have difficulty with attributions of the intentions of others, and identifying whether a character in a vignette is being deceived. Such difficulty occurs even when these children possess the basic knowledge of the difference between themselves and the characters in the vignettes, and when they are familiar with social roles such as mother, father, and child. Baron-Cohen and others theorize that this 'mind-blindness' is what makes children and adults with ASD unable to relate to and engage with the world around them.

Baron-Cohen et al (1985) used an adaptation of the false-belief test to investigate whether the core impairments seen in autism spectrum disorders were the result of a theory of mind deficit. Baron-Cohen et al administered this test to children with ASD, children with Down's Syndrome and typically developed children. They (1985) found that 85% of the sample of typically developing children were able to answer the belief question correctly, and 86% of children with Down's Syndrome also passed this test. However, only 20% of children with ASD answered the belief question correctly.[1] Baron-Cohen (2000) states that the autistic children in this sample had a higher mental age and chronological age than either of the control groups (i.e., the typically developing children and the children with

[1]Baron-Cohen (2000) states that all children in this sample were tested on a number of cognitive dimensions, to rule out the possibility that failure on the belief question was due to these factors. The children were asked where the main character placed her marble at the beginning of the vignette, to ensure that the children who answered incorrectly did not do so because their memories where overloaded. These children were also asked where the marble actually is now that the other character had moved it, and asked to identify which of the dolls was identified as the main character, to rule out incorrect answers on the basis of inattention, or misidentifying the main character in the vignette.

Down's Syndrome), and argues that this strongly indicates that individuals with ASD have a poor understanding of beliefs. The results of Baron-Cohen's false-belief test have been replicated several times, and are now considered to lend strong support to the hypothesis that autistic individuals suffer from a deficit in theory of mind (see Baron-Cohen et al 2000 for a discussion of these replication studies).

With a cognitive view of ASD, psychologists are attempting to explain the symptoms of these conditions, instead of just treating them with therapeutic interventions as behaviourists did. As mentioned in the last chapter, applied behavioural analysis (ABA)-based interventions has remained the most successful treatment approach for conditions on the autism spectrum. However, cognitive behavioural approaches emphasize the scaffolding of critical socio-cognitive skills that are precursors to developing a typically functioning theory of mind such as imitation and pretense (Mesibov et al, 1997; Rogers & Williams, 2006). This type of therapy begins in early childhood, and involves identifying the stage at which a particular child with an ASD is in terms of development, and generating teaching and learning strategies to match the child's developmental level. Unlike traditional behaviourist interventions, ABA in the cognitive framework tailors the process to the individual child, since developmental levels and difficulties in learning can vary significantly from child to child. Since the theory of mind hypothesis contains an account of the socio-cognitive processes that may be malfunctioning in ASD, it thus provides more detailed information about how to address these malfunctions than behaviourist explanations alone.

We can re-frame the theory of mind hypothesis of autism spectrum disorders as an explanation schema as follows:

> *Explanatory Target*: Why do certain children exhibit rigid, stereotyped behaviours, and impairments in language and social interaction?
>
> *Explanatory Pattern*:
>
> Children who exhibit rigid behaviours and impairments in language and social interaction because they have an underdeveloped theory of mind.
>
> A theory of mind allows one to ascribe mental states to others, and predict behaviour based on the ascriptions of mental states.
>
> A child with autism spectrum disorders is unable to ascribe mental states to others, and is thus unable to understand the actions and interactions of other people.
>
> An underdeveloped theory of mind causes the characteristic symptoms of ASD by causing impairments in the ability to interact and to communicate effectively with other people, and limiting a child's repertoire of interactive behaviours.

Thus, according to the cognitive explanation pattern for the conditions on the autism spectrum, children and adults with these disorders have a malfunctioning theory of mind, which causes the socio-cognitive impairments characteristic of ASD. These symptoms can be treated using applied behavioural analysis, and this approach is tailored to the specific socio-cognitive and language deficits each individual presents. If the theory of mind hypothesis of ASD is correct, this account should reliably identify individuals with these disorders based on the presence of the characteristic symptoms and the underlying theory of mind deficit.

4.2.2 Cognitive Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

The theory of mind hypothesis of ASD put forward by Baron-Cohen and his colleagues provides some additional diagnostic information, since it provides an explanation of the nature of characteristic symptoms of ASD and why these characteristic symptoms appear in certain children. Also, the cognitive explanation pattern meets some of the criteria for adequate explanation patterns for psychiatric conditions developed in chapter two, and thus has some strengths as an account of the conditions on the autism spectrum. If Baron-Cohen and his colleagues are correct, an undeveloped theory of mind prevents individuals with autism spectrum disorders from developing socio-cognitive skills like the ability to ascribe mental states to others, and to relate to others' mental states.

Over two decades later however, several studies have challenged the theory of mind hypothesis and its ability to explain the deficits seem in children with ASDs. While the hypothesis still has some support, it may not adequately explain the symptoms of ASD. Further, Tager- Flushberg (2007) notes that more current research indicates that ASD involves delays and deficits in other aspects of socio-cognitive functioning and information processing that are not explained by the theory of mind hypothesis by itself. Thus, the theory of mind hypothesis does not have good *empirical support*. However, the theory of mind literature provides a *consistent* body of research that continues to provide insight into the disorders on the autism spectrum, and identifies a potential *primary cause* for the conditions on the autism spectrum - a malfunctioning theory of mind. Also, Baron-Cohen and others describe the theory of mind capacity and the malfunctions in this capacity *mechanistically*, where interactions between malfunctioning parts produce the characteristic socio-cognitive symptoms of ASD. Further, the theory of mind hypothesis is *parsimonious* in that it identifies a single factor that explains the symptoms of these disorders. If correct, this hypothesis of ASD could be *specific* enough to reliably identify individuals with ASD, and to exclude children with other developmental disorders, like Downs Syndrome.

However, while the theory of mind hypothesis potentially explains why individuals with ASD have trouble with social interaction, it does not explain how the environment influences the symptoms of these disorders, nor does it explain the genetic and neurological abnormalities seen individuals with ASD. The theory of mind deficit, while a potentially robust indicator of the presence of an ASD versus another disorder, is not a cause at a lower-level of description. Without an account of break-downs in lower-level mechanisms, identifying a case of ASD versus another disorder can still be difficult. The biological and social aspects of these disorders are vital to a complete explanation of these conditions, since the interaction between biology, cognition and the environment are responsible for the development of the conditions on the autism spectrum. Thus, the theory of mind hypothesis does not include *multiple levels*, and does not explain the*interaction* between the biological, cognitive and social aspects of ASD.

A test like Baron-Cohen's false belief test may help in distinguishing between a child who has language delays but has an intact theory of mind. However, children with lower functioning types of autism spectrum disorders and children with severe mental retardation not due to Down's Syndrome have similar symptoms, and both can experience language impairments so severe that they do not develop speech. If a child is non-verbal, the false belief test will not be able to determine whether a child has an autism spectrum disorder, an unrelated condition such as mental retardation, or has both mental retardation and an autism spectrum disorder. Thus, while the false belief test can reliably differentiate between Down's Syndrome and autism, it may not reliably differentiate between a case of autistic disorder and mental retardation of a different type and severity than seen in Down's Syndrome.

The theory of mind hypothesis faces similar diagnostic problems on the other end of the autism spectrum. Since the symptoms of Asperger's syndrome tend to be milder than those of other disorders on the spectrum, children who have Asperger's and are high-functioning may have partial theory of mind abilities (note that some children in Baron-Cohen's sample passed the false belief test, Baron-Cohen et al, 1985). In the case of Asperger's syndrome, the false belief test may have limited clinical utility, since the deficits in Asperger's disorder are markedly different than those seen in other disorders on the spectrum, in that children with Asperger's have mostly normal language development and have IQs in the normal range. If a child with Asperger's manages to pass the false belief test, which at least 20% of the autistic population in Baron-Cohen's studies do, a diagnosis of another developmental condition may be given, since autistic-like symptoms appear in many psychiatric disorders, such as childhood coordination disorder, schizophrenia, and schizoid personality disorder. Thus, the theory of mind hypothesis may not be *universal* enough to reliably identify only those individuals with ASD.

The theory of mind hypothesis is one of the most well-known theories of the causes of autism spectrum disorders. Although it meets some of the criteria for adequate patterns of explanation for psychiatric disorders, it is not powerful enough on its own. The cognitive explanation pattern for psychiatric disorders meets several of the criteria developed in chapter 2, and thus can important diagnostic information to help better identify cases of ASD versus another developmental disorder. While the cognitive pattern of explanation does not have strong *empirical support*, it is *internally consistent*, *mechanistic*, and *parsimonious* since it identifies a single primary cause. However, this explanation pattern doesn't include *causes at different levels of description*, and also does not explain the *interactions* between biological, cognitive and social factors in the development and progression of the symptoms of ASD. Thus, the theory of mind test does provide information that can assist in making a diagnosis of autism spectrum disorders in some cases. However, there are still many aspects of the conditions on the autism spectrum that the cognitive explanation patterns for ASD may not be able adequately explain. While cognitive theories like the theory of mind hypothesis may be *specific* enough to differentiate between cases of ASD and cases of Downs Syndrome, they may not be *universal enough* to reliably identify cases of ASD across the spectrum.

However, in recent years, the cognitive explanation pattern for ASD just discussed has been extended and refined based on data from cognitive neuroscience, such as the discovery of the mirror neuron system. Rather than replacing previous explanations of ASD, cognitive neurological theories such as the mirror neuron hypothesis provide an account of the possible neurological underpinnings of the socio-cognitive deficits seen in individuals with ASD, such as an underdeveloped or malfunctioning theory of mind. If correct, the cognitive neuroscience explanation pattern for ASD discussed below (which I take to include the theory of mind and the mirror neuron hypothesis together) provides an account of the breakdowns in lower-level mechanisms. Thus, a cognitive neurological pattern of explanation may 'fill in' important details regarding the nature of theory of mind deficits and the symptoms of autism spectrum disorders.

4.3 Neuroscience and Brain Dysfunction: A Cognitive Neurological Explanation Pattern for Psychiatric Conditions

Cognitive neuroscience investigates the neural correlates of cognitive processes, such as perception, memory, attention, language, and theory of mind. Cognitive neuroscientists use

methods such as neuroimaging, lesion studies and computational modeling to help identify the neurological structures and systems that are involved in these and other cognitive processes (Harris, 1995). Ritchie & Richards (2002) state that by the 1980s,

> "...theories of localization in cognitive functioning validated by neuroimaging had been largely accepted within neurology. In parallel, the development of efficient psychopharmacological treatment of many major psychiatric disorders led to renewed interest in the neurobiology of these disorders and the neuroanatomical correlates of their associated cognitive disturbances. Cognitive dysfunction was also recognized within psychiatry as a major cause of disability (p. 182-183)."

These theorists argue that theories in cognitive neuroscience and neuropsychology can help identify underlying neuropathology. A better understanding of the underlying neuropathology can provide an explanation for why certain types of cognitive dysfunction occur in disorders such as depression, schizophrenia, obsessive-compulsive disorder and autism spectrum disorders.

For instance, neuropsychological deficits are commonly seen in patients with major depressive disorder, and can include dysfunction in the hippocampus, mediotemporal lobe and frontostriatal circuits. These dysfunctions are associated with difficulties in cognitive processes such as attention, executive functioning and memory. Also, patients with schizophrenia have typical patterns of neurological dysfunction associated with characteristic symptoms such as poor integration and assimilation of information, delusions, and disorganized thinking. Ritchie & Richards (2002) state that individuals with schizophrenia often have significant difficulty with tasks such as the Wisconsin Card Sorting Test, which indicates frontal lobe dysfunction. Further, these authors state that "functional imaging and morphometric studies point...to more widespread neuropathology [in schizophrenia]," where the cognitive deficits characteristic of this disorder are thought to result from "dysfunction in cortico-subcortical connectivity with a neurotransmitter imbalance in the thalamic-prefrontal motor cortex and basal ganglia (p. 186)".

Thus, patterns of explanation for psychiatric disorders in cognitive neuroscience identify malfunctions in neurological systems and structures that cause cognitive and behavioural disturbances. We can construct an explanation schema for cognitive neurological explanations of psychiatric disorders, where the symptoms of these conditions are caused by malfunctions in lower-level neurological mechanisms. The cognitive neurological explanation pattern for psychiatric disorders can be developed as follows:

Explanatory Target: Why do certain individuals exhibit the symptoms of psychiatric disorders?

Explanatory Pattern:

Certain individuals exhibit the symptoms of psychiatric disorders because there are disruptions in cognitive processes.

Disruptions in cognitive processes are caused by neurological malfunctions.

Neurological malfunctions occur because of breakdowns in neurological systems and mechanisms.

Breakdowns in neurological systems and mechanisms affect socio-cognitive processes.

Breakdowns in neurological systems and mechanisms cause the cognitive and behavioural symptoms of psychiatric disorders.

On this account of psychiatric disorders, patients display the characteristic symptoms of the various conditions listed in the DSM because they are suffering from breakdowns in neurological systems and mechanisms that underly cognitive processes such as attention, language, memory and executive control. Such breakdowns disrupt typical cognitive functioning, and thus result in the cognitive and behavioural symptoms listed as the primary diagnostic criteria for psychiatric disorders. If psychiatric disorders are explained in terms of breakdowns of neurological mechanisms, treatment of these conditions involves identifying and addressing these breakdowns, which will help to control and ameliorate the cognitive and behavioural symptoms that result from such breakdowns.

4.3.1 A Cognitive Neuroscience Explanation Pattern for Autism Spectrum Disorders

In the case of autism research, neuroscientists are attempting to identify the brain dysfunction(s) that cause the socio-cognitive deficits characteristic of ASD: impairments in social interaction, communications and rigid behaviours. Recently, research from cognitive neuroscience has received much attention from autism researchers. A class of visuomotor neurons, called 'mirror neurons' were discovered in non-human primates, and evidence suggests that such a system exists in humans as well. 'Mirror neurons' are linked to socio-cognitive processes like language acquisition, imitation, and social contagion. Because of

the potential involvement of mirror neurons in these processes, malfunctions in this neuronal system have also been implicated in the theory of mind deficit seen individuals with autism discussed above.

'Mirror neurons' were discovered in the F5 region of the premotor cortex in monkeys, and in areas such as the superior temporal sulcus (Rizzolatti & Craighero, 2004). A similar system is thought to exist in these regions of the human brain as well. Mirror neurons are activated both when a monkey performs an action, and when it observes another performing directed actions. All mirror neurons code directed movement, but the number of actions that each region of these neurons code depends on where they are located in the brain. While their exact function is unknown, researchers argue that these neurons are central to both imitation learning and understanding of action. It is thought that mirror neurons mediate the understanding of actions in the following way:

> Each time an individual sees an action done by another individual, neurons that represent that action are activated in the observer's premotor cortex. This automatically induced, motor representation of the observed action corresponds to that which is spontaneously generated during active action and whose outcome is known to the acting individual. Thus, the mirror system transforms visual information into knowledge (Rizzolatti & Craighero, 2004, p. 172).

While the presence of mirror neurons in non-human primates has been confirmed, there is no direct evidence that these neurons exist in humans as well. However, numerous neurological studies have provided indirect evidence for a human mirror neuron system. For instance, neurophysiological studies demonstrate that the motor cortex becomes active, in the absence of motor activity by the subject, when the subject observes others performing actions (Williams et al. 2001). Also, transcranial magnetic stimulation (TMS) experiments indicate that the motor system in humans has mirror properties (Rizzolatti & Craighero, 2004).

Williams et al. (2001) state that mirror neurons may be involved in many aspects of social cognition, including reading the emotions of others from their body language, and empathizing with others. These aspects of social cognition could account for the phenomenon of emotional contagion, where we emulate the postures and moods of others. Further, these theorists (2001) suggest the part of the brain that contains mirror neurons "has evolved to subserve speech in humans, with language building on top of a 'prelinguistic grammar of actions' already existing in the primate brain (p. 290)." Thus, mirror neurons may act as a bridge between perceived and performed action and speech, which could be the foundations of language in primate species. Also, if mirror neurons encode auditory

representations as well as visual ones, they may play an important role in representing the relation between words and their speaker, such as understanding personal pronouns (Williams et al, 2001). Based on these potential functions of the mirror neuron system, theorists such as Williams et al. and Rizzolatti & Craighero believe that mirror neurons are connected to theory of mind, and that we are able to 'reconstruct' a person's action in our mirror neuron system. Gallese & Goldman (1998) suggest that the activation of mirror neurons generates an executive plan to perform an action like the one being observed, which allows the observer to 'put themselves in the subject's shoes.'

Williams et al. (2001) argue that dysfunction in the mirror neuron system could be involved in the development of the characteristic symptoms of autism spectrum disorders. If mirror neurons have the functions described above, then impairments in this system could cause significant difficulties in language development and socio-cognitive skills such as imitation and pretense, both of which are impaired in individuals with ASD. Also, if mirror neurons play a role in language development, a malfunctioning mirror system could explain why autistic children have significant language delays, and why they have trouble using and understanding personal pronouns (recall Kanner, 1943 and Firth, 1991 have documented this impairment in individuals with ASD).

Further, Rizzolatti & Fabbri-Destro (2010) state "Autism affects a variety of nervous structures, from the cerebral cortex to the cerebellum and brainstem[h]owever, in the context of a broader neurodevelopmental deficit, a set of ASD symptoms (impairment in communication, language, and the capacity to understand others) appears to match functions mediated by the mirror mechanism (p. 230-231)." These researchers cite several behavioural and brain imaging studies that indicate a malfunctioning mirror neuron system could be present in individuals with ASD.

In this way, the mirror neuron research could extend and refine the cognitive theory of ASD discussed in the previous section. Thus, neuroscientific accounts of autism do not represent a shift in theoretical views, but are rather an extension and expansion of the data from cognitive psychology about the nature of the symptoms of autism spectrum disorders. On this view, a malfunctioning or impaired mirror neuron system is thought to be involved in the development of deficits in theory of mind seen in individuals with ASD, and thus partially responsible for the characteristic symptoms: language delay, communication and interaction difficulties, and stereotyped behaviours. Williams et al. (2001) also discuss the link between mirror neurons and other autistic deficits, such as difficulties with emotional contagion and shared attention. Individuals with autism spectrum disorders often have difficulty relating to those around them as intentional objects. If the mirror system is malfunctioning in individuals with autism spectrum disorders, this could partially explain why they are unable to interact with those around them -they are unable to process and

interpret the perceptual stimuli that are normally fed into a typically developed mirror neuron system. Further, individuals with autism often do not respond to social cues regarding emotions, as Asperger and Kanner discussed in their reports of the behaviour of autistic children in the educational environment. Apserger (1944) noted that one can communicate with higher functioning autistic individuals and get them to note social norms by talking in an intellectual, affect-free way, but these individuals still do not understand the emotions of others.

We can construct an explanation schema based on the cognitive neuroscientific theory of ASD, and the possible connection between the mirror neuron system and an underdeveloped theory of mind. The cognitive neuroscience explanation pattern for ASD is as follows:

> *Explanatory Target*: Why do certain children exhibit rigid and stereotyped behaviours, and impairments in language and social interaction?
>
> *Explanatory Pattern*:
>
> Children with autism spectrum disorders exhibit rigid behaviours and impairments in language and social interaction because they have an underdeveloped theory of mind.
>
> A theory of mind allows one to ascribe mental states to others, and predict behaviour based on the ascriptions of mental states.
>
> A child with autism spectrum disorders is unable to ascribe mental states to others, and is thus unable to understand the actions and interactions of other people.
>
> An underdeveloped theory of mind may partially be caused by a malfunctioning mirror neuron system.
>
> An underdeveloped theory of mind causes the characteristic symptoms of ASD by causing impairments in the ability to interact with other people, to communicate effectively with other people, and limits the repertoire of interactive behaviours of a child with autism spectrum disorders.

The cognitive neurological explanation pattern potentially provides more causal information by hypothesizing that the socio-cognitive deficits seen in these disorders could be the result of neurological malfunctions, such as an impaired mirror neuron system. Thus, according to the cognitive neurological explanation pattern for ASD, the characteristic symptoms of ASD are caused by an undeveloped theory of mind, which could be caused in

part by a malfunctioning mirror neuron system. The mirror neuron hypothesis is consistent with several clinical features of both Kanner's and Asperger's children in their respective studies, such as the language and communication difficulties of individuals with autism. If this account is correct, it potentially provides more discriminating criteria for identifying cases of autism spectrum disorders, since the characteristic symptoms must be present, and these must be caused by specific neurological impairments that prevent the normal development of socio-cognitive capacities like theory of mind. If this explanation is correct, the symptoms of these disorders can still be treated by behavioural interventions, since this type of therapy is designed to facilitate the development of socio-cognitive processes like imitation and pretense. Understanding the neurological dysfunctions that cause the cognitive impairments characteristic of these disorders can help in developing more focused intervention designed to better address, mitigate and compensate for these dysfunctions, which may make the learning and integration of skills like imitation and pretense more manageable.

The discovery of mirror neurons is recent, and their exact role in causing or aggravating the symptoms of autism spectrum disorders is not clear. The causal link between damaged or malfunctioning mirror neurons is highly speculative. There is good empirical support for the presence of mirror neurons in non-human primates. However, their function in the human brain and the role they play in social cognition in humans is not well understood. It is also unclear, at this point, whether the discovery of mirror neurons will give a full explanation of autism spectrum disorders, on which to base diagnostic categories that predict causes, symptoms, progression and response to treatment. Further, several studies do not support the mirror neuron hypothesis. For instance, Fan et al (2010) produced a study that indicates that mirror neuron function may be preserved in individuals with ASD, to varying degrees. Fan et al (2010) do note that more severe deficits language and social communication could be correlated with greater degrees of mirror neuron dysfunction, but they urge caution in drawing causal links between mirror neuron dysfunction and the development of ASD.

4.3.2 Cognitive Neuroscience Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

The cognitive neuroscience explanation pattern for ASD has several strengths, and meets some of the criteria for an adequate explanation patterns in psychiatry. While the 'mirror neuron hypothesis' of ASD has received attention in recent years, this hypothesis does not have strong *empirical support.* However, if more research further elucidates the role

of the mirror neuron system in the development of the disorders on the autism spectrum, then the cognitive neurological explanation pattern for these conditions may provide more causal information than schemas previously discussed. Further, the discovery of the mirror neuron system in non-human primates and the indirect evidence for such a system in humans lends more empirical support to the theory of mind hypothesis. Thus, the mirror neuron hypothesis coheres well with one of the most popular cognitive theories of ASD, and provides a *consistent* account of the neurological mechanisms that might underlie the socio-cognitive processes that are impaired in these disorders.

Mirror neurons are at a lower-level of description than the cognitive processes they facilitate. Thus, if correct, cognitive neurological theories such as the mirror neuron hypothesis could begin to provide an account of the underlying neurological mechanisms in socio-cognitive skills like theory of mind, and extend the theory of mind hypothesis by providing a way to link language and social deficits with impairments in neurological systems. Thus, we may be able to fill in some of the biological aspects of a *multi-level mechanistic* representation of the disorders on the autism spectrum. Also, investigating malfunctions in the mirror neuron system can generate insight into how breakdowns in neurological systems affect the severity and presentation of the socio-cognitive deficits characteristic of ASD, thus providing some information regarding how the parts of the mechanism *interact.* Finally, if correct, the mirror neuron system hypothesis identifies a potential *primary* cause that may partially explain the core symptoms of ASD.

According to the cognitive-neurological explanation pattern, the individuals that should be included in the diagnostic category *autism spectrum disorders* have the characteristic cognitive and behavioural symptoms and certain neurological impairments that cause these symptoms. On this account, the presence or absence of certain neurological impairments and the symptoms determines whether an individual has an autism spectrum disorder. It contains a causal account that, if correct, identifies not just individuals with autistic symptoms, but individuals with particular physical conditions that underlie these symptoms. Thus, understanding ASD as cognitive neurological disorders could help to generate diagnostic categories with more *validity* and *reliability*, since it is not the presence or absence of a cluster of symptoms that identifies a patient with an autism spectrum disorder, but rather these symptoms *and* the presence of certain neurological impairments.

However, there are some problems with the cognitive-neuroscience explanation pattern for ASD. The mirror neuron hypothesis, even in conjunction with the theory of mind hypothesis, does not meet all the criteria for adequate patterns of explanation for psychiatric conditions. Williams et al. (2001) state that mirror neuron dysfunction could be the result of genetic abnormalities, of external conditions that inhibit the development of this neural system, or an interaction between organic and environmental causes. While we gain

an understanding of the neurological deficits that could underly underlying the cognitive symptoms, information about how these neurological deficits are affected by genetic and social factors can be important for the diagnosis and effective treatment of autism spectrum disorders. A neurologically-based explanation pattern for psychiatric conditions such as those on the autism spectrum may not be able to account for the significant variability in the symptoms of these conditions in the same patient over time. As mentioned previously, the symptoms of ASD can shift and change as the child ages, as the child interacts with the social environment, and if the child receives treatment. The cognitive neurological explanation pattern for ASD may not be able to account for why the symptoms of these disorders change in these ways throughout the lifespan, given that the impairments in the mirror neuron system happen early in life. In order to explain why the severity and presentation of the symptoms of these disorders might change, an adequate explanatory pattern would have to cite other biological factors that lead to changes in the mirror system, or to social influences such as stressful environments.

Also, the mirror neuron hypothesis may not account for the variability of symptoms seen in the different autism spectrum disorders. Patients diagnosed with different disorders on the spectrum often have significant differences in the presentation and severity of the characteristic socio-cognitive symptoms, such as the relatively typical IQ and language development in individuals with Aspergers versus the severe deficits in IQ and language characteristic of autistic disorder. While the mirror neuron hypothesis, when combined with the theory of mind hypothesis is a multi-level explanation, there are other levels, such as the social level that must be included to explain ASD. Further, mirror neurons are potentially involved in many aspects of language development and social interaction, and different types of dysfunctions in these processes are features of many psychiatric disorders. Thus, mirror neurons may also be involved in the development of other psychiatric conditions, both related and unrelated to ASD, if those disorders involve deficits in social interaction, language, and processing of information from the environment. The cognitive neurological explanation pattern for ASD provides some important insights into the biological and cognitive aspects of these disorders. However, if mirror neurons are also involved in the development of other psychiatric disorders, the presence of mirror neuron deficits may not be able to differentiate between a case of ASD and another disorder(s). Thus, the cognitive neurological explanation pattern for explaining the development and progression of the disorders on the autism spectrum may not be *specific* enough to reliably identify patients with these conditions. Further, the cognitive neurological explanation pattern for ASD may not be *universal* enough to identify cases of ASD given the variation and heterogeneity of the characteristic symptoms of these disorders.

This section analyzed the cognitive neurological explanation pattern for psychiatric

conditions such as those on the autism spectrum. The mirror neuron hypothesis has good *empirical support*, and provides a *consistent* account of the possible neurological dysfunctions that are involved in the development of the disorders on the autism spectrum. Further, if correct, this explanation pattern may help to generate a multi-level mechanistic account of ASD, and how some of the parts of this complex mechanism interact. Finally, if correct, the cognitive neurological explanation pattern may identify a possible *primary cause* for the development of ASD. However, the socio-cognitive symptoms characteristic of the disorders on the autism spectrum may be the result of more than one type of neurological dysfunction, and mirror neuron hypothesis may not be able to explain how and why these symptoms change over time, since these changes may also be the result of the interaction with environmental and social factors. Further, the pathogenesis of the malfunctions in mirror neuron functioning is not clear, and there may be genetic, environmental or genetic x environmental factors that contribute to mirror neuron function and dysfunction. Thus, while cognitive neurological patterns of explanation may provide more causal information that the previously-discussed schemas, such a pattern may not be *specific nor universal enough* to reliably and correctly identify cases of ASD.

4.4 Conclusion

This chapter examined two contemporary explanatory patterns for the conditions on the autism spectrum, and discussed the strengths and weaknesses of each pattern. The theory of mind hypothesis of ASD provides a potential causal theory regarding the disorders on the autism spectrum, and augmented with the mirror neuron hypothesis, provides a multi-level explanation. Identifying the neurological deficits that underlie the cognitive and behavioural symptoms of these ASD potentially provides more robust diagnostic criteria, however, the presence or absence of certain neurological deficits may not be specific enough criteria for classifying psychiatric disorders. In the next chapter, I discuss genetic and epigenetic patterns of explanations for ASD. While all the explanatory patterns discussed in this thesis will likely be helpful in explaining psychiatric conditions, I argue that an integrated explanation pattern that includes the genetic, epigenetic, neurological, cognitive, and social levels of description may be the most powerful for explaining the causes and development of conditions like those on the autism spectrum.

Chapter 5

Explanation Patterns of Autism Spectrum Disorder: Genes and Epigenetics

5.1 Introduction

So far, I have shown that the psychoanalytic, behaviourist, cognitive and cognitive neurological patterns for ASD are unable to meet all of the criteria for adequate patterns of explanation for psychiatric disorders, and are thus unable to capture the complex and multi-directional causation involved in the development of these conditions. Patterns of explanation that do not capture the causal complexity of psychiatric disorders have weak explanatory and predictive power, which leads to poor reliability and validity in psychiatric diagnosis. Thus, the patterns of explanations for psychiatric disorders discussed far so may not be able to inform the development of diagnostic categories that reduce the problems of poor reliability and validity, and arbitrary diagnostic boundaries.

This chapter explores genetic and epigenetic explanation patterns for autism spectrum disorders. Instead of focusing solely on the nature of cognitive and behavioural symptoms, genetic and epigenetic patterns of explanation provide more information about why such impairments are seen in autism spectrum disorders, and why these disorders affect certain children. While genetic and epigenetic explanations of these disorders are far from complete, such explanations can provide important pieces of the complex etiology of ASD. Genetic explanations of these conditions attempt to identify the causes of the cognitive and neurological impairments characteristic of ASD, and determine what distinguishes an

individual with an autism spectrum disorder versus another developmental disorder (or other psychiatric disorder) at the genetic level. On the other hand, epigenetic explanations of these conditions identify the mechanisms by which certain genes are expressed, which can help elucidate how the phenotypes for certain psychiatric conditions, including ASD, can develop. According to epigenetic theories of human development, phenotypes for both individuals with and without psychiatric conditions are expressed based on complex interactions between genes, and interactions between genetics and the perinatal, neonatal and early infant environments.

Genetic and epigenetic theories of psychiatric disorders such as those on the autism spectrum provide valuable causal insight into the genetic and genetic x environmental interactions that are involved in the development of these conditions. However, like the other schemas discussed in this thesis, neither the genetic nor the epigenetic patterns of explanation meet all of the criteria for adequate explanations of psychiatric conditions. Thus, while such patterns of explanation are powerful and explain some of the causal features of psychiatric disorders, these patterns cannot accurately and reliably identify cases of ASD in the clinical population by themselves.

5.2 A Genetic Explanation Pattern for Psychiatric Conditions

Genetic explanations identify phenomena at a lower level of description that can fill in important details about both the neurological and cognitive aspects of psychiatric disorders. Thus, genetic explanations of psychiatric disorders can help explain why certain types of neurological and cognitive impairments are seen in some disorders listed in the DSM, such as Down's Syndrome and Fragile X syndrome. In the case of Down's Syndrome, a third 21st chromosome is responsible for the lower IQ, language difficulties, and other socio-cognitive deficits characteristic of that disorder. Fragile X syndrome is the most common known cause of inherited mental retardation, and is caused by a break in the X chromosome at region X27q3 (Persico & Bourgeron, 2006). This condition affects approximately 1 in 4000 males and 1 in 8000 females (Narayanan & Warren, 2006; in Moldin & Rubenstein, 2006)). Individuals with Fragile X syndrome experience developmental and language delays, and have IQs in the lower range. Thus, the presence or absence of certain genetic defects, such as a third 21st chromosome, includes or excludes an individual in a diagnostic category, instead of a particular set of symptoms alone.

Genetic mutations or defects may also partially explain the neurological and cognitive

deficits in more casually complex conditions such as addiction, bi-polar disorder, major depression and anxiety. However, in these conditions, many genetic mutations in different genes interact to cause the neurological and cognitive impairments, and these conditions also have significant social causes that influence the presentation, onset, and severity of the symptoms. Even though genetics may not entirely explain the presence or absence of symptoms, genetic explanations can indicate which patterns of neurological dysfunctions may be more likely to occur, given the genetic defect(s) that are present. Understanding which neurological dysfunctions are more likely can help to predict what sort of cognitive deficits an individual may have, given the nature of the neurological impairments. Thus, genetic explanations can be combined with neurological and cognitive theories to potentially provide a more detailed and multi-level account of why certain cognitive and behavioural symptoms appear in certain individuals. We can formalize a genetic explanation pattern for psychiatric disorders as follows:

> *Explanatory Target* Why do certain individuals develop the symptoms of psychiatric disorders?
>
> *Explanatory Pattern*
>
> The symptoms of psychiatric disorders are partially caused by interacting genetic abnormalities and defects.
>
> Patterns of interacting genetic abnormalities and defects are associated with certain kinds of neurological malfunctions.
>
> Certain kinds of neurological malfunctions are associated with certain kinds of cognitive and behavioural impairments.
>
> Certain cognitive and behavioural impairments are identified as the characteristic symptoms of a particular psychiatric disorder.

Thus, understanding the genetic abnormalities involved in the conditions listed in the DSM can provide a partial account of why certain cognitive and behavioural symptoms appear in particular psychiatric disorders. According to a genetic pattern of explanation, what distinguishes one psychiatric disorder from another is a particular set(s) of genetic mutations or abnormalities and a particular cluster(s) of cognitive and behavioural symptoms. Thus, in one way, a genetic explanation pattern can make distinctions between the different psychiatric disorders sharper, since the presence or absence of certain genes can include or exclude an individual in a diagnostic category.

5.2.1 The Genetics of Autism Spectrum Disorders

In recent years, there has been much research dedicated to discovering the genes involved in the development of autism spectrum disorders. Genetic explanations of autism spectrum disorders, while still speculative, attempt to provide information about the causes of the neurological impairments associated with these conditions, which are identified by explanations of ASD like the mirror neuron hypothesis. In their review of the current research investigating the genes involved in ASD, Muhle, Trentacoste & Rapin (2004) argue there is strong evidence that disorders on the autism spectrum are heritable. These researchers note that the rate of recurrence in siblings of affected individuals is 2% to 8%, which is much higher than the prevalence rate of ASD in the general population. Twin studies conducted in Scandinavia (Steffenburg, Gillberg, Hellgren, Anderson, Gillberg, Jakobsson, et al 1989) and Britain (Bailey, Le Couteur, Gottesan, Bolton, Simonoff, Yudza, et al 1995) report that identical twins had a rate of concordance of greater than 60% for autistic disorder, with no concordance for fraternal twins (Muhle et al., 2004; Rutter, 2005; Bonora et al., 2006). Further, Muhle et al (2004) state "when the unaffected twin discrepant for autism was re-evaluated for broader autistic phenotypes, including communication skills and social disorders, the concordance among the [twins in the British study] rose...from 60% to 92% in [monozygotic] twins and from 0% to 10% for [dizygotic] twins (p. 475)."

The concordance rate for monozygotic (identical) twins is not 100%, and thus there are pairs of MZ twins studied where only one of the twins has an autism spectrum disorder. However, even in pairs of identical twins where only one twin has an autism spectrum disorder, there are several traits that the non-autistic twin may exhibit that are similar to the impairments seen in ASD. For instance, Folstein & Rutter (1977a, 1997b) found that the non-autistic twin in a pair of MZ twins usually had language disorders and social impairments, but did not meet the diagnostic criteria for ASD. Further, Bonora et al (2006) note that social impairments, difficulties in communication and rigid behaviours are more common in the relatives of children with autism that in relatives of non-autistic children. Thus, the data from family and twin studies indicates that the heritability estimate for autism is greater than 90%, making ASD some of the most heritable of the disorders listed in the DSM (Bonora et al. 2006).

The high rate of concordance between monozygotic twins lends strong support to the hypothesis that ASD have a significant genetic component in their etiologies. However, despite the high condordance rate, the exact genetic origins of ASD are unknown. Most cases of ASD are 'idiopathic', meaning that no obvious genetic abnormality is present. Investigations into the genetic basis of ASD indicates that idiopathic autism, i.e. autistic disorders without a clear genetic origin, does not follow a pattern of monogenic inheritance,

even though the heritability estimate is over 90%. Bonora et al. (2006) state

> the difference in pairwise concordance between MZ and DZ twins and the rapid decline in recurrence rate with decreasing genetic relatedness indicates a non-Mendelian, complex mode of inheritance. The falloff in monozygotic to dizygotic twin concordance rates is too steep to be explained by an additive hypothesis, regardless of the genes involved, and evidence for multiplicative genetic interaction (epistasis) is provided when the risk ratio decrease exponentially across different degrees of relationship... (p. 51)

Most cases of autism spectrum disorders seen in clinical practice are 'idiopathic,' with the exception of Rett's disorder, which is discussed in the following section.

Further, there are several developmental-neurological conditions caused by similar genetic abnormalities to the ones implicated in the development of ASD. The genes thought to be involved in the development of ASD are also implicated in developmental-neurological conditions such as Angelman's syndrome, Prader-Willi syndrome, Fragile-X syndrome and Turner's syndrome.

5.2.2 Pieces of the Genetic Puzzle: Related Neurodevelopmental Conditions and Complex Models of Expression

Research into related conditions like those mentioned above provides clues as to which genes and what sort of genetic abnormalities are responsible for the characteristic symptoms of autism spectrum disorders. Angelman's syndrome (AS) and Prader-Willi syndrome (PWS), for example, often involve autistic-like behaviours but are not considered part of the autism spectrum. Idiopathic autism spectrum disorders are thought to be caused by as many as 5-15 genes, and some candidate genes have been identified. Numerous studies have indicated that X-linked deficits and abnormalities on chromosomes 15 and 7 may be involved in the development of 'idiopathic' autism.

Investigations into the genetic origins of conditions like Angelman's and Prader-Willi syndromes indicate that abnormalities on chromosome 15 are involved in the development of ASD (see Bonora, et al, 2006; Muhle et al, 2004; Schanen, 2006). Bonora et al. (2006) state "[t]he most prevalent chromosome 15 abnormalities are supernumerary isodicentric chromosome 15 and maternally derived interstitial duplications of the 1511-q13 region...(p. 52)." Muhle et al (2004) state the cytogenetic abnormalities in the 15q11-q13 region of chromosome 15 point to several other candidate genes for future study. These authors

argue that the GABAa receptor gene cluster is strongly implicated in the development of autism spectrum disorders, since this gene cluster is involved in the inhibition of excitatory neural pathways and in neurodevelopment.

Abnormalities in the q22-q33 region of chromosome 7 have also been implicated in the pathogenesis of autism spectrum disorders (Muhle et al, 2004). Persico & Bourgeron (2006) state that a link between ASD and abnormalities on chromosome region 7q is one of the most replicated findings in ASD research. Muhle et al (2004) argue that "[t]he protein reelin (RELN), which localizes to a site of chromosomal translocation at 7q22, is a large secreted glycoprotein potentially involved in neural migration during development. It is of particular interest given that it binds to neuronal receptors and that the pathology of autism can include migration cell deficits (p. 477)." This region of chromosome 7 has been called an autism susceptibility locus, or AUTS1, by the International Molecular Genetic Study of Autism Consortium (Muhle et al, 2004). AUTS1 contains several genes that may be involved in the development of ASD, including FOXP2, RAY1/ST7, IMMP2L, GRM8, CADPS2, and WNT2, all of which are associated with speech and developmental delays (Muhle et al, 2004).

Finally, certain X-linked disorders and abnormalities are also associated with autistic symptoms. Some of these conditions, such as Fragile-X and Turner's syndrome, are co-morbid with ASD, indicating that some of the candidate genes are located on the X chromosome. Both Fragile-X and Turner syndrome patients exhibit mental retardation and autistic-like behaviours. Turner syndrome affects young girls, and is caused by monosomy of the X chromosome. Females with Turner Syndrome display an increased susceptibility to ASD as opposed to XX females (Persico & Bergeron, 2006, p. 350). This data is compatible with the existence of imprinted genes on the X chromosome. Muhle et al. (2004) state that two separate studies have identified links to the Xq13-q21 region of the X chromosome, which contains the neuroligin genes. These genes code for cell-adhesion molecules, called neuroligins, which are thought to be involved in synapse development. Also, Persico & Bourgeron (2006) state that Fragile X syndrome, briefly discussed above, is often associated with autistic features and approximately 2-3% of males with autism also have Fragile X syndrome. Further, these researchers state that 20-40% of Fragile X patients meet the diagnostic criteria for autism spectrum disorders.

Persico & Bourgeron (2006) state "[the] proteins that have been implicated in ASD[s] to date...are involved in neurodevelopment and many have roles in synaptic function (p. 350.)" While speculative, these authors state that the candidate genes identified so far in the development of ASD indicate three aspects of the pathogenesis of these conditions. The first, which is associated with 7q22-q33 and the protein REELIN, involves neural migration and its role in neurodevelopment. Persico & Bourgeron (2006) state "the evidence surrounding

the reelin pathway, in conjunction with neuropathological studies, underscores the role of altered neuronal migration in generating the aberrant neural networks that underlie altered information processing in autism (p. 355)." The second aspect of the development of ASD concerns the 15q11-13q region and the GABA receptor cluster in that region. 15q11-13q is likely involved in the development of ASD because "converging evidence from functional studies of MeCP2 and NLGN, and from chromosomal rearrangements involving the GABA receptor gene cluster, underscores the crucial role of unbalanced excitatory-inhibitory networks in abnormal CNS excitability and function in autism (p. 355)." The final aspect of the pathogenesis of ASD that genetic studies have indicated is "that abnormal synapse formation and dendrite spines could contribute to ASD (Persico & Bourgeron, 2006, p. 355)," which are associated with several of the candidate genes just discussed.

5.2.3 A Genetic Explanation Pattern for Autism Spectrum Disorders

Unlike conditions such as Huntington's chorea, where the cognitive and neurological impairments are caused by a single gene mutation, ASD have a complex genetic origin. Muhle, Trentacoste & Rapin (2004) state "autistic disorders are polygenic; that is, several synergistically acting genes in an affected individual's genome may be required to produce the full autistic phenotype (p. 475)." Since there are several genes likely involved in the development of these disorders, a multi-gene model is needed. Borona et al. (2006) discuss a multilocus epistatic model of ASD that has been proposed to explain the genetic etiology of these disorders. A multi-locus epistatic model of the etiology of ASD is different from a traditional model of genetic heritability, where mutations in certain genes cause the same or a similar phenotype, and the presence of each mutation is sufficient on its own to cause the disease (Borona et al, 2006). In an epistatic multilocus model, numerous genes interact with one another to produce the phenotype in question. Thus, no single genetic variant or defect is necessary or sufficient to cause ASD. Instead, multiple variants inherited through one or more genes cause the neurological and socio-cognitive impairments that characterize autism spectrum disorders. Further, "the multilocus-epistatic model of autism is congruent with the aggregation of features of the broader autistic phenotype among first-degree relatives of individuals with autism (Borona et al, 2006, p. 52)." This may indicate that those first degree relatives have only some of the predisposing genes for ASD, and autistic features in first degree relatives can also indicate variable expression of the set of genes involved in these disorders.

We can generate a genetic explanation pattern for the condition on the autism spectrum as follows:

Explanatory Target Why do certain children develop idiopathic autism, which are disorders characterized by impairments in communication, interaction and stereotyped behaviours?

Explanatory Pattern

Idiopathic autism is caused by the interaction of multiple genes and genetic abnormalities.

These genetic abnormalities are thought to include genes in the 15q11-13q region of chromosome 15, the q22-q33 region of chromosome 7, and Xq28 locus on the X chromosome.

These genes code for proteins that are involved in neural migration, the creation of synapses and other aspects of neurodevelopment.

These genes interact to cause particular socio-cognitive impairments in communication, interaction, and motor behaviour during early phases of development, which are identified as the characteristic symptoms of autism spectrum disorders.

On this account, the symptoms of ASD are explained by the presence of certain genetic abnormalities and defects, which are associated with certain types of neurological dysfunction, which in turn causes the cognitive and behavioural symptoms of these disorders. If ASD are genetic conditions, the presence or absence of certain genetic abnormalities will demarcate a case of ASD from another psychiatric disorder with similar symptoms. The presence of certain symptoms, even if they are similar to other conditions listed in the DSM, are explained by genetic abnormalities, and thus the similarity in symptomatology between ASD and other conditions will not confuse or blur diagnostic boundaries.

5.2.4 Genetic Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

Genetic explanations of ASD may provide additional causal information about the origins of the neurological deficits that are involved in these conditions, which are associated with the characteristic cognitive and behavioural symptoms. Further, genetic data may indicate why certain conditions may be co-morbid with ASD. If we can identify which abnormalities cause particular neurological and socio-cognitive deficits, e.g. by finding individuals with one developmental condition and not others, we have a better understanding of the boundaries of the category of autism spectrum disorders. Thus, even though genetic explanations

of ASD are speculative, a genetic explanation pattern may provide more information than previous schemas, and fills in more of the causal history of these disorders.

However, the interaction between variations and mutations in several genes makes it difficult to classify and define the disorders on the autism spectrum based solely on the genes involved in their development. Unlike genetic disorders that are caused by variations or malfunctions in a single gene or set of genes, the lack of a clear genetic origin makes ASD harder to classify. In the case of single-gene disorders, the presence or absence of that gene reliably tells us whether someone has or will have that disorder or not. In the case of disorders that are polygenic and epistatic, clinicians cannot be as accurate in determining whether a patient has a particular condition, and the progression and severity of the symptoms. ASD are complex genetic conditions, involving multiple genes interacting to produce a constellation of symptoms. Identifying *primary* causes for ASD at the genetic level can be difficult, since many of the candidate genes are also implicated in the development of other conditions. At present, the genetic data does not clearly identify the syndromes we call autism spectrum disorders. While candidate genes have been identified, the exact genetic origins of idiopathic autism have yet to be uncovered. No one gene, or set of genes, is a reliable indicator of the presence or absence of an autism spectrum disorder, with the exception of Rett's disorder, to be discussed in the next section. Thus, there is good *empirical support* for the genetic origins of ASD, but *consistent* findings of the exact genes involved and their interactions are yet to come.

Even though genetic studies like those reviewed above have identified a number of candidate genes thought to be involved in the development of autism spectrum disorders, none of the chromosomal abnormalities nor the candidate genes identified fully correlates with the symptoms of ASD. Even we are able to identify the patterns of interacting genetic abnormalities involved in the development of ASD, doing so may not fully explain the progression, onset, and symptomatology of these disorders. The strong concordance for monozygotic twins indicates that autism spectrum disorders are heritable conditions, but researchers caution that the autism phenotype is difficult to isolate, since the genes identified in related conditions like Angelman's and Prader-Willi syndromes are found in a small percentage of the autistic population (Veenstra-VanderWeele & Cook, 2004; Muhle et al, 2004). Thus, a genetic explanation pattern for autism spectrum disorders is not *specific* enough to identify a case of ASD versus another developmental disorder, like Angelman's or Prader-Willi syndromes.

As more of the genetic origins of the conditions on the autism spectrum are discovered, genetic explanations of ASD could become part of an *inter-level mechanistic* explanation that represents the social, cognitive and biological features of these disorders. However, causation in genetic explanations runs in one direction only: genetic abnormalities cause

neurological deficits, that in turn cause cognitive and behavioural symptoms. However, Borona et al. (2006) note that environmental factors may also influence the severity of the expression of the autistic phenotype. Further, genetic changes only occur during chromosomal and physical development, and mutations in genetic expression early in life do not explain the onset or changes of certain symptoms as the individual ages and interacts with the environment. Recall that adequate explanation patterns for psychiatric conditions must be able to not only represent causes on multiple levels, but how these causes *interact*. As I have argued throughout this thesis, the social and cognitive features of psychiatric disorders can affect the biological level. Further, changes in the biological level continue to cause changes at the cognitive and social levels throughout the individual's lifespan. Thus, the genetic explanation pattern for ASD is not *universal* enough to identify cases of ASD if the disorder is discovered later in life, when the presentation of symptoms has been influenced by environmental factors.

Thus, a genetic explanation pattern can provide important causal insight into the development of disorders like those on the autism spectrum, but it does not meet all the criteria for adequate explanation patterns for psychiatric conditions. This section argued that genetic explanations of ASD have good *empirical support*, however the findings are not *consistent* with respect to the candidate genes involved or the interactions between these candidate genes. Further, while genetic explanations can be *mechanistic*, and incorporated into a *multi-level explanation,* they are not able to explain the *interactions* between the biological, cognitive and social aspects of psychiatric conditions, nor do they explain how cognitive and social factors can influence the biological level. Therefore, the genetic explanation pattern of ASD is not *specific* enough nor *universal* enough to reliably and accurately identify individuals with these conditions in the clinical population.

5.3 Epigenetics and Psychiatric Conditions

The last section highlighted the limits of the genetic explanation pattern for explaining the causes and progression of psychiatric conditions, given that these disorders are not caused by mutations in single genes and do not follow a Mendelian pattern of gene expression. Recently, researchers have begun to investigate the *epigenetic* mechanisms involved in the development of psychiatric disorders. The field of epigenetics studies the changes in gene expression that occur during early development that are not directly coded for in DNA. Just as DNA sequences are conserved during mitosis, epigenetic states can be inherited by the daughter cells during DNA replication followed by somatic cell division. Epigenetic explanations of diseases identify changes to genes based on environmental influences, and

thus attempt to account for changes in neurological development and functioning at the molecular and genetic level throughout early socio-cognitive development. Several studies indicate that gene transcription errors and changes to regulatory regions of genes play a role in mediating alterations in gene expression associated with certain psychiatric disorders (Nestler, 2009). Epigenetic processes are thought to be at work in the development of conditions like ASD, since many of the neurological, cognitive and behavioural symptoms cannot be explained by genetic abnormalities alone.

5.3.1 Epigenetics: Broad and Narrow Definitions

There are several definitions given for what the field of epigenetics involves, some of which are quite broad and go well beyond the scope of molecular biology and genetics. Under some broad definitions, 'epigenetic changes' refers generally to changes in genetic material not coded in DNA, which may include a wide range of phenomena. On the other hand, some definitions are much narrower and exclude certain processes, mechanisms and environmental causes as being 'epigenetic.' The different definitions have an impact on what sort of causes will be included in epigenetic explanations of psychiatric conditions, including those on the autism spectrum. While some broad definitions of epigenetics allows for both environmental and genetic causes in explanations of psychiatric conditions like ASD, these definitions can be vague about what counts as an 'environmental' cause, and the link posited between genes and environment can often be too speculative. Thus, some definitions of epigenetics do not lend much explanatory power to a causal account of autism spectrum disorders or other psychiatric disorders, while others provide potentially powerful explanatory patterns.

Waddington (1942) was the first to coin the term 'epigenetics' and defined it as "all those events that lead to the unfolding of the genetic program for development (p. 635, in Goldberg, Allis, & Bernstein, 2007)." Unfortunately, this definition is too broad to lend much explanatory power, since events that lead to the unfolding of the phenotype can encompass an enormous range of phenomena, from DNA methylation to environmental pathogens to social and political forces that shape one's cultural environment. However, in a paper entitled "The inheritance of epigenetic defects," Holliday (1987) introduced a narrower definition of epigenetics that referred to changes in DNA methylation. The term *epimutation* was also introduced at this time, which refers to heritable changes in genes that are not caused by changes in DNA sequences.

Other definitions were introduced in the 1990s, two of which Holliday (2006) discusses. The first is broader than the second, and he argues that while both are incomplete, together they encompass most of the known epigenetic mechanisms. The definitions are as follows:

> 1) The study of the changes in gene expression which occur in organisms with differentiated cells, and the mitotic inheritance of given patterns of gene expression
>
> 2) Nuclear inheritance which is not based on changes in DNA sequence.

Holliday (2006) states the first definition can include DNA methylation, but also a number of other epigenetic mechanisms. The second definition includes mechanisms like imprinting, but would exclude cytoplasmic events which would be included as epigenetic under the first definition. A similar definition of epigenetics was proposed by Alan Wolffe, who defined epigenetics as heritable changes in gene expression that occur without a change in DNA sequence (Nakao, 2001). Nakao (2001) states that there are several processes included in Wolffe's definition, which include DNA methylation, histone-modifying enzymes, chromatin remodeling, and transcriptional factors. Further, chromosomal structures such as the centromere, kinetochore and telomere are included under the category of epigenetics even though they are not directly involved in gene function (Nakao, 2001). Under this broader definition, Nakao (2001) suggests that epigenetics can be understood as a system to selectively utilize genome information, through activating or silencing particular genes.

Adrian Bird (2007) proposes another definition that attempts to clarify what sort of alterations to the genome should be considered 'epigenetic', and how broad the definition should be. Bird defines epigenetic events as "the structural adaptation of chromosomal regions so as to register, signal or perpetuate altered activity states(p. 398)" Bird favours this definition over the many others in the literature because it

> is inclusive of chromosomal marks, because transient modifications associated with both DNA repair or cell-cycle phases and stable changes maintain across multiple cell generations qualify. It focuses on chromosomes and genes, implicitly excluding potential three-dimensional architectural templating of membrane systems and prions, except when these impinge on chromosome function. Also included is the exciting possibility that epigenetic processes are buffers of genetic variation, pending an epigenetic (or mutational) change of state that leads an identical combination of genes to produce a different developmental outcome (p. 398).

Nestler (2009) also defines epigenetic mechanisms in this broad sense, which he says includes cellular processes that integrate environmental stimuli, which influence gene expression through regulation of chromatin. Thus, the epigenetic explanation schema generated in this section will be based on a broader definition of epigenetics.

Central to epigenetic explanations is the metastable epiallele. Dolinoy et al. (2007) define metastable epialleles as "alleles that are variably expressed in genetically identical individuals due to epigenetic modifications that were established in early development. The term 'metastable' refers to the labile nature of the epigenetic state of these alleles, while 'epiallele' defines their potential to maintain epigenetic marks transgenerationally (p. 32)." Further, these researchers note that "gestational exposure to nutritional agents and other environmental factors has been demonstrated to alter epigenetic marks at metastable epialleles (p. 32)." Thus, metastable epialleles refer to regions of the genome affected by epigenetic modifications that account for differences in phenotypic expression of identical cells, and these epialleles can be affected by environmental factors.

Thagard & Findlay (2010) propose a potential epigenetic explanation schema. On their account, the explanation schema runs as follows:

> *Explanatory target*: Why does a patient have a disease with associated symptoms?
>
> *Explanatory Pattern*: The patient has an epimutation affecting the accessibility of gene(s) in a given region of chromatin.
>
> The improper expression of gene(s) produces the disease and its symptoms.

In this schema, "epimutation" refers to any epigenetic modifications to a particular region of chromatin, which can be genetic or environmental in origin. Genetic causes of epimutations usually occur when there is a mutation in a gene that would normally code for a component of epigenetic regulation, such as HAT or DNMT, or in MeCP2 in the case of Rett's disorder, which I discuss in a later section. Epimutations are well documented in disorders that are the result of genetic imprinting, such as Angelman's syndrome and Prader-Willi syndrome, which are discussed below. Further, the mutation in MECP2 in Rett's disorder is associated with the disruption of epigenetic processes, since MeCP2 functions to mediate interactions between DNA methylation and chromatin remodeling (Amir et al, 1999; also discussed in Thagard & Findlay, 2010).

5.3.2 Epigenetic Mechanisms: DNA Methylation, Histone Modifications, and Imprinting

Gene expression occurs because DNA wraps around octamers of histone proteins to form nucleosomes. The base pairs that make up DNA wrap around a core made with histone

proteins H2a, H2b, H3, and H4 to form the nucleosome. Nucleosomes of DNA and histone proteins are called chromatin, and changes to chromatin influences gene expression by either silencing or activating genes. When chromatin is condensed, genes are silenced, and when chromatin is open genes are activated. Changes in chromatin structure are regulated by epigenetic patterns of DNA methylation and histone modifications. Enzymes involved in these processes include DNA methyltransferases (DNMTs), histone deacetylases (HDACs), histone acetylases, histone methytransferases, and the methyl-binding protein MECP2 (Rodenhiser & Mann, 2006).

DNA methylation involves the addition of a methyl group (CH3) to a CpG base pair. This process is carried out by enzymes called DNA methyl transferases (DNMT). Areas in the DNA sequence where there are many CpG pairs clustered together are called CpG islands, and they are located in the upstream regulatory regions of many genes. Prior to their discussion of the potential epigenetic explanation schema, Thagard & Findlay (2010) give more detail about epigenetic changes to gene expression and epigenetic modifications to DNA. They state that hypermethylation of CpG islands is associated with transcriptional silencing of the downstream gene, while hypomethylation of these regions is associated with transcriptional activation of the downstream gene. These authors (2011) also note that "[r]esidues on the slim 'tails' that extend away from each of the core histone proteins can be modified by the addition of small chemical groups that affect the attraction between histones and DNA. Such modifications include acetylation, phosphorylation, methylation, ubiquitination, and sumoylation (p. 2)". These modifications work to either weaken or strengthen the attraction between DNA and the histone proteins. For instance, acetylation of lysine residues by an enzyme called histone acetyl transferases (HAT) weakens the interaction between DNA and histones to allow transcriptional activation. On the other hand, a class of enzymes called histone deacetylation (HDAC) removes a methyl group from a CpG pair, and thus represses transcription (Thagard & Findlay, 2010).

In addition to DNA methylation, changes to histone proteins can change DNA organization and gene expression. Histone-modifying enzymes ensure that a receptive DNA region is either accessible for transcription or silenced (Rodenhiser & Mann, 2006). Thus, active regions of chromatin have high levels of acetylated histones, and inactive regions of chromatin have methylated DNA and deacetylated histones. These regions act as an epigenetic 'tag' on targeted DNA that actives or silences genes. These epigenetic processes are reversible, and ensure that specific genes are activated or silenced in the presence of certain developmental and biochemical cues, which Rodenhiser & Mann (2006) state can include hormone levels, dietary components and drug exposures. These authors argue that DNA methylation patterns can fluctuate in response to changes in diet, inherited genetic polymorphisms and exposure to environmental chemicals like heavy metals and

hydrocarbons.

Epigenetic changes can also occur throughout the lifespan. Rodenhiser & Mann (2006) state

> [i]mmediately following fertilization, the paternal genome undergoes rapid DNA demethylation and histone modifications. The maternal genome is demethylated gradually, and eventually a new wave of embryonic methylation is initiated that establishes the blueprint for the tissues of the developing embryo. As a result, each cell has its own epigenetic pattern that must be carefully maintained to regulate proper gene expression (p. 343.)

Further, Crepaldi & Riccio (2009) note that recent evidence indicates that differentiated neurons in the central nervous system deploy epigenetic changes that produce behavioural responses. These authors state the expression of behaviour, either simple or complex, is the result of the complex interplay between neuronal networks connected within the CNS. Even though neurons are fully differentiated and do not produce daughter cells, chromatin within neurons retains a significant amount of plasticity.

There are two other epigenetic mechanisms most commonly discussed in the epigenetic literature: X-chromosome inactivation and genetic imprinting. The former affects females and occurs when one X chromosome is silenced in every cell. Imprinting refers to the hemizygous expression of certain genes in that is parent-of-origin dependent. Imprinting is regulated by DNA methylation and histone modifications, and allows a gene to 'remember' whether it was inherited from the mother or the father, so only one of the two parental alleles is expressed. As discussed in the previous section, congenital developmental disorders like Angelman's syndrome and Prader-Willi syndrome are related to ASD, and these disorders are the result of imprinting errors. In Angleman's syndrome, imprinting errors are inherited on the paternal allele, and in Prader-Willi syndrome, errors are inherited on the maternal allele.

5.3.3 An Epigenetic Explanation Pattern for Psychiatric Conditions

Theorists such as Nestler (2009) are investigating the epigenetic mechanisms involved in the development of psychiatric disorders with complex etiologies, such as addiction. Epigenetic research presents a potentially powerful explanatory account of the etiology of psychiatric disorders. For instance, epigenetic mechanisms give a partial account of how environmental

factors throughout the lifespan such as stressful life events can cause changes in the brain (see Moffitt, Caspi & Rutter, 2005; Nestler, 2009). In the case of addiction, many theorists have argued that there is a biological component to addiction, and that relatives of addicts often develop substance abuse problems as well. However, there are also environmental aspects to addiction, such as trauma, a home environment where addictive behaviour and substance abuse are modeled, and introduction to substance use at an early age. If the relationship between environmental and genetic risk factors for addiction could be elucidated, an epigenetic explanation of this condition could account for why some individuals become addicts and some do not, despite similar upbringings and genetic predispositions. Further, Thagard & Findlay (2010) argue that epigenetic explanations of diseases are powerful because they provide a mechanistic account of how such environmental and genetic factors interact to cause diseases including psychiatric disorders like addiction. If correct, epigenetic explanations of psychiatric disorders may provide a way to include causes that occur early in life as well as during later periods of socio-cognitive development, because epigenetic changes or mutations can occur any time during an individual's life.

If such epigenetic explanations of psychiatric conditions could be generated, the course, progression and presentation of symptoms of a complex disorder like addiction may be better predicted. On this account, the progression of symptoms could be predicted based on the nature of the subject's environment, the extent of genetic predispositions, and the interactions between these causes.

We can construct an epigenetic explanation schema for psychiatric disorders as follows:

> *Explanatory target*: Why does a patient develop the symptoms of a psychiatric disorder?
>
> *Explanatory Pattern*:
>
> The patient has multiple, interacting epimutations, that affect the accessibility of genes in given regions of chromatin.
>
> These epimutations cause the improper expression of genes.
>
> The improper expression of genes produces the multi-level impairments characteristic of a given psychiatric disorder.
>
> Impairments in neurological development and functioning result in particular socio-cognitive impairments.
>
> The improper expression of certain genes involved in neurological development and functioning cause particular cognitive and behavioural symptoms.

Thus, if further research continues to elucidate epigenetic causes for psychiatric conditions, an explanatory framework using a broad definition of epigenetics may be a powerful one for explaining the causes and progression of psychiatric conditions.

5.3.4 Re-Framing the Genetic Research: ASD as an Epigenetic Disorder

Epigenetic research is compatible with the genetic research discussed in the previous section, and presents the possibility to explain the role of genetics in ASD by understanding how the particular genes involved in these disorders are expressed. Epigenetic explanations of autism spectrum disorders identify the genetic, epigenetic, and *de novo* changes to the genes involved in the development of neurological structures and pathways associated with socio-cognitive processes like theory of mind and imitation. Thus, an epigenetic explanation pattern for autism spectrum disorders identifies the epimutations associated with the neurological impairments that cause the cognitive and behavioural symptoms characteristic of these disorders. Schanen (2006) states that

> despite considerable effort over the past decade, [the] underlying risk alleles [of ASD] have been remarkably elusive...[and] the obstacles encountered in mapping risk alleles have led a number of investigators to rethink the model of inheritance to include contributions of new mutations and/or epigenetic mechanisms such as genomic imprinting or epimutations in the underlying genetic susceptibility to ASD (p. 138)

Schanen (2006) argues that epigenetic factors are clearly at work in the development of ASD, since epigenetic regulatory mechanisms are central to the development of Rett's disorder and Fragile X syndrome. Further, in the case of autism spectrum disorders other than Rett's Disorder, there may be many factors that contribute to the development of the ASD phenotype, from maternal diet to epimtuations and *de novo* mutations and epimutations. For example, Schanen (2006) states DNA methylation can be modified by mutations, maternal exposures and postnatal experiences. Thus, research into the genes involved in the development of autism spectrum disorders indicates that a mixed genetic/epigenetic/*do novo* model is the most likely candidate for an accurate genetically based causal account of these disorders (Jiang, et al. 2004).

Researchers discovered that FMR1, an X-linked gene, is involved in the development of Fragile X syndrome (Persico & Bourgeron, 2006). Persico & Bourgeron (2006) state

that the "Fragile-X mental retardation protein (FRMP) is encoded by the FMR1 gene, the 5'-untranslated region (5'-UTR) of which contains a polymorphic CGG repeat that can undergo triplet-repeat expansion, resulting in promoter hypermethylation and FMR1 gene silencing. The clinical outcome is fragile X syndrome...(p. 350)". FMRP is an RNA-binding protein, located on the Xq28 region of the X chromosome. This particular genetic abnormality is associated with what Persico & Bourgeron (2006) call 'secondary' autism, or 'syndromic' autism, which refers to autistic behaviours associated with other developmental-neurological conditions.

Schanen (2006) states that genomic imprinting is an epigenetic mechanism that leads to parent-of-origin specific expression. Recall the discussion in the previous section regarding Angelman's and Prader-Willi syndromes, and the overlap between the symptoms of these syndromes and those that characterize ASD. Angelman's syndrome is caused by the duplication of 15q11-q13 region of chromosome 15. The Angelman syndrome gene, UBE3A, is maternally imprinted, which means that it is expressed only from the paternal allele. Prader-Willi Syndrome is caused by abnormalities in the same region of chromosome 15 as Angelman's syndrome. However, Prader-Willi is caused by a deficiency in the expression of the paternal allele, instead of the maternal allele and is not as strongly associated with ASD as Angelman's syndrome. Since AS and PWS share some genetic abnormalities with ASD, it is very likely that epigenetic mechanisms are at work in the development of ASD as well. Further, genetic studies investigating the origins of autism spectrum disorders also point to the role of imprinting in the pathogenesis of these disorders.

The genes on the 7q region of chromosome 7 and abnormalities on the X chromosome implicated in ASD exhibit epigenetic properties, and their functions can be interpreted using an epigenetic explanation pattern. Schanen (2006) notes that the genes at 7q32.2 show parent-of-origin effects on the sharing of alleles in this region. Further, epigenetic mechanisms have been proposed to explain the gender bias in ASD (Schanen, 2006). Recall that autism affects four times more boys than girls, and the girls who do have ASD usually have Rett's syndrome. A study of 80 females with Turner syndrome, also known as monosomy X, found parent-of-origin effects. Females with a paternally derived X chromosome performed better on measures of social cognition than females with a maternally inherited X chromosome. This indicates that an imprinted locus, tentatively mapped onto Xq or Xp, increases social behaviours in females, and the genes on this locus are hypothesized to insulate females from developing ASD (Schanen, 2006). We can develop an explanation schema for ASD, based on this epigenetic research. The schema is as follows:

> *Explanatory Target* Why do certain children develop ASD, which are disorders characterized by impairments in communication, interaction and stereotyped

behaviours?

Explanatory Pattern

ASD is caused by the interaction of multiple genes and epigenetic mechanisms.

These epigenetic abnormalities are thought to include the imprinted regions of the 15q11-13q region of chromosome 15, the q22-q33 region of chromosome 7, and Xq28 locus on the X chromosome.

These genes code for proteins that are involved in neural migration, the creation of synapses and other aspects of neurodevelopment.

These genetic abnormalities and epimutations interact with each other and with environmental causes to cause particular socio-cognitive impairments in communication, interaction, and motor behaviour during early phases of development.

This schema is very similar to the genetic schema discussed in the previous section. However, an epigenetic explanation pattern attempts to explain why the concordance rate between identical twins is not 100% despite strong evidence that ASD is one of the most heritable disorders in the DSM, and why there is a significant gender bias in individuals with ASD. Recall that the heritability of the broad ASD phenotype is approximately 92%, and several authors have argued that ASD are the result of genetic, epigenetic and *do novo* changes, where epigenetic changes are thought to be a significant aspect of the development of the ASD phenotype. On an epigenetic account of these disorders, one twin may not develop an autism disorder despite having identical genes, and thus identical risk alleles, because he or she does not develop certain epimutations which cause these risk alleles to be expressed in the ASD phenotype. Since by definition epigenetic changes are not coded in DNA, these changes are responsible for one twin developing ASD while the other does not. If the data just discussed are correct, and continue to provide clues as to the etiology of ASD, epigenetic mechanisms can help explain the patterns of inheritance and the development of the ASD phenotype, and thus may begin to elucidate the complex causation involved in ASD that cannot be explained by genetics.

Rett's Disorder: Single-Gene Mutations and Epigenetic Changes

Rett's disorder, or Rett's syndrome, is a pervasive developmental disorder currently included in the autism spectrum disorders listed in DSM-IV-TR. Rett's disorder, like the other disorders on the autism spectrum, is characterized by impairments in communication, interaction and motor behaviour. However, unlike autistic disorder, Asperger's

syndrome and pervasive developmental disorder not otherwise specified (PDD-NOS), the impairments in Rett's disorder appear after a period of at least 6 months of normal development, and previously learned socio-cognitive skills are lost. Also, unlike the other disorders on the spectrum, Rett's disorder mostly affects girls.

Van Acker, Loncola, & Van Acker (2005) state "Rett syndrome is a phenotypically distinct progressive X-linked dominant neurodevelopmental disorder...(p. 126)." These researchers state that next to Down Syndrome, Rett's disorder is one of the most common causes of mental retardation in females. Genetic research into the etiology of Rett's disorder discovered abnormalities at the Xq28 locus, and Schanen (2006) states that Rett's disorder "is a complex neurological disorder that arises from mutation in the gene that encodes the methyl-CpG-binding protein 2 (MeCP2)." MeCp2 plays an important role in silencing genes during certain critical periods of central nervous system development (Van Acker et al, 2005), and mutations in this gene results in the failure to produce the MeCP2 protein. Thus, unlike other disorders on the autistic spectrum, Rett's disorder is the result of the mutation of a single gene that is responsible for epigenetic changes during early development. Van Acker et al. (2005) state the MeCP2 protein

> binds to prescribed mehtylated cytosine nucleotides (CpG dinucleotides) on the DNA. The bound DNA-MECP2 complex then interacts with a histone deacetylase complex and the transcriptional co-repressor Sin3A. Together, these repressors alter the chromatin making the genes inaccessible to transcriptional activators - in essence, silencing the further transcription of that gene (p. 137).

Thus, the genes that would normally be silenced by the MeCP2 protein continue to engage in transcription. While the exact role and number of the genes MeCP2 is meant to silence is not known, Van Acker et al. (2005) state that these genes may be involved in the development and regulation of the brain and central nervous system.

Based on the discovery of the MeCP2 mutation, we can construct an epigenetic explanation schema for Rett's disorder as follows:

> *Explanatory Target* Why do certain females exhibit autistic symptoms, such as impairments in motor, social interaction and communication skills already acquired at six months of age?
>
> *Explanatory Pattern*
>
> Certain females have a mutation in the MECP2 gene.

> MECP2 encodes the methyl-CpG-binding protein 2.
>
> MeCP2 is involved in methylating DNA and silencing certain genes that are thought to be responsible for the degredation of already acquired socio-cognitive skills and autistic symptoms.
>
> Mutations in MECP2 causes autistic symptoms and the loss of motor and communication skills already acquired by six months of age.

Thus, Rett's disorder may have a different etiology than other disorders on the autism spectrum, since a mutation in a single gene is responsible for this condition and for the autistic symptoms, unlike the multiple genes likely involved in the other autistic disorders. Since MeCP2 is implicated in Rett's disorder, many researchers have argued that Rett's disorder should be considered an epigenetic disorder, since MeCP2 is involved in transcription and other epigenetic processes (see Rutter, 2005, in Volkmar et al, 2005a; Beaudet & Zoghbi, 2006). While the role of MeCP2 in idiopathic autism is less clear, defects in the MECP2 gene is thought to be involved in the development of these disorders as well.

5.3.5 Epigenetic Explanation Patterns and the Criteria for Adequate Explanation Patterns in Psychiatry

The epigenetic explanation pattern is a powerful one, and meets most of the criteria for adequate explanations of psychiatric disorders. First, although research into the epigenetic mechanisms involved in disorders like ASD, the data is far from conclusive and accounts of the epigenetic causes of most psychiatric conditions are highly speculative. An epigenetic explanation pattern of conditions like Rett's disorder is *internally consistent*, and has good *empirical support*. Further, understanding the causes of Rett's disorder can assist in the diagnosis of this condition in female children. While autism spectrum disorders is more common in boys, female children can also develop autism spectrum disorders, and determining which subtype of ASD a female child may have can be difficult based on symptoms alone. However, according to an epigenetic explanation pattern, a female child cannot be diagnosed with Rett's disorder unless she has the epimutation in the MeCP2 gene. If a female child does not have an epimutation in the MeCp2 gene, she should be diagnosed with another autism spectrum disorder besides Rett's disorder. Thus, an epigenetic explanation of Rett's disorder seems *universal* and *specific* enough to clearly demarcate this condition from the others on the spectrum, despite the similarities in neurological, cognitive and behavioural symptoms.

While an epigenetic explanation pattern for Rett's disorder seems to clearly demarcate that condition from others on the spectrum, and from other developmental disorders, the epigenetic causes of the other disorders on the spectrum are must less robust. As research continues, an epigenetic explanation pattern may be able to identify possible *primary causes* of autism spectrum disorders, even though these causes are both environmental and biological. Further, if correct, epigenetic explanations of conditions like ASD can help to fill in important details about the *interactions* between certain environmental factors and certain genes that result in particular neurological dysfunctions, which in turn cause particular cognitive and behavioural symptoms. Thus, epigenetic explanations could be mapped onto a *multi-level, mechanistic* representation of the interactions between the biological, cognitive and social levels of description, which produce changes in neurological functioning which results in the characteristic socio-cognitive and behavioural symptoms of ASD.

However, chapter 2 argued that the causation in psychiatric conditions such as those on the autism spectrum is complex and involves interactions between all of the levels, not just the environment and genetic of explanation. Epigenetic changes may be able to explain some of the interactions between primary causes, but it is less clear how epigenetic mutations are responsible for the cognitive aspects of psychiatric conditions, or how breakdowns and ongoing dysfunction in neurological and cognitive mechanisms can affect the genetic level. Thus, while the epigenetic pattern of explanation is a potentially powerful one, it may be not be powerful enough on its own to explain the complex, multi-directional and ongoing causation that is involved in the development of psychiatric conditions, such as most of the conditions on the autism spectrum.

5.4 Conclusion

Causal theories from epigenetics may be able to partially explain some of the complex causation involved in the development of psychiatric conditions such as ASD, and can be integrated into a multi-level, mechanistic explanation of the causes of a particular disorder. However, it seems that in the case of most of the conditions currently listed in the DSM, including most of the conditions on the autism spectrum, epigenetic explanations may not be robust and complex enough on their own to fully elucidate the causal structure of psychiatric conditions, and thus does not, at present, meet all of the criteria for adequate patterns of explanations for psychiatric conditions. However, as the next chapter argues, epigenetic explanations can be incorporated into an integrated explanation schema which I argue *does* meet the criteria for adequate patterns of explanation for psychiatric conditions

like those on the autism spectrum.

Chapter 6

Explanation Patterns for Autism Spectrum Disorder: An Integrated Schema

6.1 Introduction

In this section, I develop a sketch of an integrated schema for explaining the possible causes of psychiatric conditions such as autism spectrum disorders. To develop the integrated schema, I incorporate the more powerful schemas previously discussed into one that includes the genetic, epigenetic, cognitive neurological and cognitive levels of explanation. Admittedly, the account developed here is speculative. The actual merits of such an explanatory pattern are deeply contingent on the empirical evidence and theoretical changes that will emerge as research into the causes of psychiatric conditions continues. However, I argue that an integrated schema will likely be the most powerful one for elucidating the causal relations in psychiatric conditions like ASD for four reasons.

First, many theorists and clinicians discussed in chapter two argue that explanations of psychiatric conditions will likely be explained using a combination of all the explanatory patterns examined in this thesis. For instance, theorists such as Murphy (2006, 2008), Mitchell (2008a, 2008b) and Kendler (2008) argue that explanations of psychiatric conditions should incorporate multiple levels of description. Further, Mitchell (2008a, 2008b) and Kendler (2008) argue that social factors, such as stressful life events or trauma, are just as essential to explaining the causes and development of psychiatric conditions as biological and cognitive causes.

Second, as chapter one and two argued, emerging data from fields such as genetics, epigenetics, neuroscience, cognitive neuroscience and cognitive psychology indicate that psychiatric disorders are the result of complex interactions between multiple causes at different levels of description. Murphy (2006, 2008), Mitchell (2008a, 2008b), and Kendler (2008) have argued that adequate explanations of psychiatric conditions should include not just an account of the causes of psychiatric disorders, but how these causes interact across levels to produce the characteristic symptoms of conditions such as those on the autism spectrum.

Third, I have shown in the preceding discussion that none of the non-integrated explanatory frameworks seems to be powerful enough to explain the complex causation involved in the development of psychiatric conditions like those on the autism spectrum. Some of these schemas, such as the cognitive, cognitive neurological, genetic, and epigenetic schemas, meet some of the criteria outlined in chapter two, and thus have some explanatory and predictive power. However, while the conditions on the autism spectrum have biological, cognitive and social features, none of these features are powerful enough on their own to accurately and reliably identify cases of these disorders. However, I will argue in what follows that an integrated explanatory schema may be the most powerful for elucidating the causal structure of psychiatric disorders because such a schema meets all the criteria for adequate explanation frameworks in psychiatry. This fourth and most important reason for adopting an interactive approach for explaining the causes of psychiatric disorders will be elaborated upon in what follows.

6.2 Integrated Explanation Pattern for the Causes of Psychiatric Conditions

To help develop an integrated explanation schema, I examine two examples of integrated models of two very different psychiatric conditions discussed by Mitchell (2008a) and Kender (2008) respectively. Mitchell (2008a) uses Major Depressive Disorder to argue that an "integrated pluralist approach" (p....) is the most powerful way to represent the complexity of psychiatric conditions. DSM-IV identifies several symptoms that are characteristic of this disorder, such as anhedonia (the inability to feel pleasure from activities and experiences once enjoyed), thoughts of death and suicide, and profound feelings of sadness and guilt. Mitchell discusses a model of MDD developed by Kendler, Gardner & Prescott (2006), which includes both a "bottom-up" and a "top-down" analysis of the causal processes involved in the development of this condition. Kendler, Gardner & Prescott identify some of the biological factors associated with major depression, such as imbalances in the

neurotransmitters serotonin and noradrenaline, changes in brain structure and function in areas such as the frontal lobe, and changes to circulation and hormone levels in the brain. On the other hand, these theorists also identify a number of environmental and cognitive factors that are associated with the development of a major depressive episode, such as low-self esteem, childhood abuse, low social support, and traumatic events such as the loss of a loved one. However, none of these biological, cognitive or environmental factors are present in every patient, and none of these factors are necessary conditions for developing a major depressive episode (Mitchell, 2008a).

Mitchell also discusses the results of studies such as those conducted by Caspi et al. and Kendler et al. (2005) that indicate that individuals with two short 5-HTT alleles (as opposed to one long and one short allele, or two long alleles) are more susceptible to episodes of major depression because they are more sensitive to stressful life events. Mitchell states that "[w]hat is significant about the gene-environment interaction results found for the 5-HTT gene and stressful life events is that they entail a nonreductionist approach for explaining the complex causal network leading to MDD. In rejecting a purely molecular reductionist approach, Kendler et al. state 'our results argue against this as they suggest that understanding gene action in depression requires us to both "go down" to individual genetic polymorphisms and "go out" into the environment with detailed measurements of stressful experiences (Kendler et al. 2006, p. 534; in Mitchell, 2008a, p. 33). Mitchell states that "it is clear that depression is a complex behaviour of a complex system that depends on multiple causes and multiple levels...(2008a, p. 30)." She argues that "[i]n general, psychiatric disorders will not be amenable to purely or even partially reductive strategies. Because evidence suggests that they are behaviours of an integrative complex system, an integrative methodology is needed to understand the etiology and causally explain such behaviours (2008a, p. 33)." Thus, Mitchell argues that an "integrative pluralist approach" is the most powerful one for explaining the causal processes involved in complex phenomena like major depression (2008a, p. 35).

Kender (2008) uses the example of alcohol dependence to argue that that complex phenomena like addiction best understood using a multi-level mechanistic approach. He argues that a "range of compelling evidence indicates that [psychiatric] disorders involve causal processes...that act within and outside of the individual, and that involve processes best understood from biological, psychological, and sociocultural perspectives (p. 695)." To illustrate the power of multi-level mechanistic explanations for psychiatric conditions like addiction, Kendler (2008) develops a sketch of a multi-level mechanistic explanation of alcohol dependence. The characteristic symptoms of this condition listed in the DSM include continued use of alcohol despite physiological, financial and social problems, tolerance to increasing doses of alcohol, and withdrawal symptoms when alcohol is not consumed.

He discusses some of the data and theories of addiction and alcohol dependence, which identify possible causal factors at the genetic, psychological and social levels of description. Biological factors associated with alcohol dependence include genetic predispositions that can increase an individual's susceptibility to alcohol abuse and dependence. Kendler (2008) states that the genes associated with alcoholism can affect the way an individual metabolizes alcohol and the way alcohol interacts with receptors in the brain, which can in turn affect the individual's patterns of drinking and rate of intoxication. Psychological factors associated with alcohol abuse include personality traits such as impulsiveness, neuroticism and extraversion. Finally, social factors such as availability of alcohol, peer substance use, socio-economic status, and cultural acceptance of substance use are also associated with alcohol dependence.

Kendler (2008) states that each of these causal factors are vital to a complete understanding of a complex condition such as alcohol dependence. However, it is just as important to understand how these causes interact to produce the characteristic symptoms of this condition identified in the DSM. He argues that in studies using "twin designs, genetic effects on risk for drinking or alcohol dependence have been shown to vary as a function of religious beliefs, marital status, and social environment. Thus..[t]heir effects are dependent on both biochemical and psychosocial contexts (p. 697)." He elaborates on the ways in which genes and the environment interact to produce complex conditions like alcohol dependence. He states that genes "strongly influence the initial response to ethanol. At one extreme, individuals with a variant of aldehyde dehydrogenase metabolize acetaldehyde so slowly that they develop a dysphoric flushing reaction after significant ethanol consumption. This genetic effect substantially reduces the chances that such individuals will repeatedly reexpose themselves to the large doses of ethanol needed to develop dependence. At the other extreme, individuals who genetically have reduced sensitivity to ethanols effects are more likely to drink frequently and have an elevated risk of developing alcohol dependence. So genes influence subjective ethanol effects, which influence alcohol expectations, which in turn loop out into the environment, influencing consumption patterns, which in turn affect risk of alcohol dependence (2008, p. 697)."

However, although environmental factors influence maladaptive patterns of alcohol consumption characteristic of alcoholism, these environmental factors also affect the biological aspects of alcohol dependence. As Kendler explains, repeated and frequent "[e]xposure to ethanol produces physiological tolerance both from increased metabolic rates and decreased CNS sensitivity. This can produce a positive feedback loop in which early phases of heavy drinking permit an individual to better 'hold their liquor,' which in turn encourages yet greater consumption (2008, p. 697)." Further, psychological variables can also influence and exacerbate the biological and environmental risk factors for developing alcohol

dependence. He states that "[i]mpulsive, risk-taking adolescents seek out similar peers who provide support for and access to further antisocial and drug-taking behaviors. Genetic factors influence this process. So genetically influenced temperament causes individuals to select themselves into high-risk environments, which feed back on their risk for alcohol dependence by providing easy access to ethanol and encouragement for its excessive use (2008, p. 697)."

Despite the importance of understanding the cognitive and social factors in the development of alcohol dependence, Kendler also argues against purely cognitive or social explanations of psychiatric conditions. Without an understanding of how the genes that predispose an individual to alcohol dependence are affected by cognitive and social variables, clinicians are unable to explain why some individuals with this genetic predisposition develop alcoholism while others do not. On the other hand, a purely social or cognitive explanation account of conditions like alcohol dependence will also not be sufficient, since not all individuals who live in low socio-economic areas or who are exposed to substance use early develop alcoholism. Further, Mitchell (2008a) highlights the importance of knowing how complex phenomena like major depression or alcoholism interact with and are influenced by the environment in which they exist. Thus, a purely reductive approach will not be sufficient for explaining the causes of psychiatric disorders because of the importance of elucidating because the "type of interaction of causal components, not just the fact that there are other causal components...entails th[e] need for multiple levels in giving a causal explanation (Mitchell, 2008b, p. 128)" for psychiatric conditions.

Based on the discussion and the examples from Mitchell and Kendler above, we can generate a general integrated explanation schema for psychiatric conditions, which provides the general explanatory pattern for identifying the causes of conditions such as Major Depressive Disorder and addiction. While the discussions from Mitchell and Kendler do not include mention of epigenetic data for Major Depression or addiction, the previous section mentioned research into the possible epigenetic processes involved in the development of both these conditions (e.g. Nestler, 2009). Further, since an epigenetic schema was analyzed and evaluated in this thesis (and shown to have explanatory power), I will include the epigenetic level of description in the integrated schema I develop below. The general integrated schema for identifying the causes of psychiatric conditions is as follows:

> *Explanatory Target*: Why do certain people exhibit the symptoms of psychiatric conditions?
>
> *Explanatory Pattern*:
>
> The patient has particular patterns of interacting genetic and epigenetic abnormalities and defects.

Certain patterns of interacting genetic and epigenetic abnormalities and defects are associated with certain kinds of impairments in neurological development and functioning.

Impairments in neurological development and functioning result in breakdowns in neurological systems and mechanisms.

Breakdowns in neurological systems and mechanisms result in neurological malfunctions.

Neurological malfunctions cause disruptions in cognitive processes, such as the generation and implementation of attributions and beliefs.

Individuals with impairments in cognitive processes such as the generation and implementation of attributions and beliefs, develop disruptions in typical cognitive functioning and maladaptive patterns of social interaction.

Ongoing multi-directional and multi-level interactions between these biological and cognitive causes and conditions in the environment result in the characteristic symptoms of psychiatric disorders.

This schema identifies not just the causes at the biological, cognitive and social levels of description, but also attempts to identify the possible interactions between these causes to produce the symptoms of the psychiatric disorder in question. While this schema is speculative, it may be robust enough to accurately identify cases of psychiatric disorders in the population. Recall that emerging causal data on disorders like MDD, addiction, and ASD indicate that there is a heterogeneity of causal features across patients in a particular diagnostic category. In other words, different patients with the same characteristic symptoms may have some or all of the biological, cognitive and social causes in varying degrees and combinations. Thus, for most of the conditions contained in the DSM, no single-level or reductive causal schema will be able to identify all cases of a particular disorder. However, an integrated schema may be powerful enough to do so. On this account, although no two patients will have identical degrees, combinations, and interactions between the primary causes (and there is likely to be many) identified for each condition, all patients within a diagnostic category will have at least some of these causal features. For patients that seem to have the characteristic symptoms of a particular disorder, but do not have the primary causes identified by the explanation schema for that disorder, the patterns of biological, cognitive and social malfunctions and the interactions between these malfunctions will be better explained by the integrated schema for another disorder.

6.2.1 Proposed Integrated Explanation Pattern for Autism Spectrum Disorders

If multi-level interactive mechanistic explanations are indeed the most powerful for explaining the causes and development of psychiatric conditions, such explanations should also be a powerful tool for explaining the causes and development of the conditions on the autism spectrum. Like those that suffer from Major Depression and addiction, individuals with autism spectrum disorders also have a heterogeneous combination of the core symptoms, the degree(s) of severity of those core symptoms, and how those symptoms are affected by the surrounding environment. In this section I argue that the integrated schema could also provide a general framework for explaining the conditions on the autism spectrum.

Clinicians such as Boucher (2009) discuss the strengths of a multi-level explanation of ASD that incorporates biological and cognitive levels of description. She (2009) states "explaining autism is difficult for a number of reasons. These include the fact that a full explanation of autism will involve understanding the root causes, linking these to abnormalities of brain development and function, and linking these to psychological deficits that in turn cause the kinds of behaviour that are characteristic of people with ASD. In other words, there are at least three levels of explanation that have to be causally linked to each other (p. 112)." Like Mitchell (2008a, 2008b), Boucher (2009) also argues for the importance of understanding the role of the environment in the development of ASD and the presentation of the characteristic symptoms of these conditions. Further, Boucher (2009) argues against the feasibility of reductive explanations of autism spectrum disorders, and states that a multi-level explanation is necessary to understand the complex causation involved in the development of these conditions.

Although ASD are (at present) classified as a cluster of conditions with similar symptoms, each subtype currently identified as part of the autism spectrum has social, cognitive and biological features, and thus have a similar causal structure to the psychiatric conditions such as depression and alcoholism discussed above. However, it is very likely that each subtype within the autism spectrum will have a distinct causal structure, which may include different primary causes, or indicate that certain explanatory levels within the integrated schema should be emphasized. In those cases, certain subschemas can be developed that emphasize particular explanatory levels over others, depending on the nature of the causal evidence from ongoing autism research. For instance, Rett's disorder is caused by an epigenetic mutation. Thus, although Rett's disorder has cognitive and social features, the primary cause of this disorder is the mutation in the MeCP2 gene. Using the integrated framework developed in this section, Rett's disorder could be explained using a subschema of the integrated schema below where the primary cause it at the epigenetic level, and the

social and cognitive features of this condition are the result of this epimutation.

For the subtypes on the autism spectrum, i.e. Childhood Disintegrative Disorder, Autistic Disorder, Asperger's Syndrome, and Pervasive Developmental Disorder Not Otherwise Specified, the causes and progression of the symptoms are much more complex and are not completely explained by the epigenetic explanation pattern. Thus, for ASD other than Rett's disorder, we can generate an integrated explanation schema for autism spectrum disorders other than Rett's disorder as follows:

> *Explanatory Target*: Why do certain children develop the characteristic symptoms of autism spectrum disorders?
>
> *Explanatory Pattern*:
>
> The child has particular patterns of interacting genetic abnormalities and defects, such as those identified with 15q11-13q regions of chromosome 15, the q22-q33 region of chromosome 7, and Xq28 locus on the X chromosome.
>
> These genes code for proteins that are involved in neural migration, the creation of synapses and other aspects of neurodevelopment.
>
> The child also has certain patterns of interacting epimutations that cause the improper expression of genes involved in neurological development and functioning.
>
> These epigenetic abnormalities are thought to include the imprinted regions of the 15q11-13q region of chromosome 15, the q22-q33 region of chromosome 7, and Xq28 locus on the X chromosome.
>
> These genetic and epigenetic abnormalities cause impairments in neurological development and functioning, which result in breakdowns in neurological systems and mechanisms such as the mirror neuron system.
>
> Breakdowns in neurological systems and mechanisms such as the mirror neuron system are associated with cognitive impairments such as an underdeveloped theory of mind.
>
> Stereotyped behaviours, impairments in language and social interaction are associated with cognitive impairments such as an underdeveloped theory of mind.
>
> The characteristic symptoms of ASD are caused by the on-going interaction between genetic, epigenetic, neurological, and cognitive causes and features of the individual's environment.

The integrated schema for autism spectrum disorders has several strengths. Some of these strengths are inherited from integrating previously discussed schemas, whereas others are gained from the integration of these previous schemas. First, the integrated schema retains the ability of some of the previous schemas to differentiate between and identify cases of autism spectrum disorders and cases of unrelated disorders with similar symptoms. For instance, the theory of mind hypothesis developed by Baron-Cohen et al. (1985) reliably differentiates between typically developed children, children with Downs Syndrome and children with ASD. Further, epigenetic causal theories can help differentiate between cases of ASD and cases of Prader-Wili Syndrome and Angelman's Syndrome, and reliably identifies cases of Rett's disorder in females with autistic symptoms. However, with an integrated schema, we can 'fill in' some of the causal 'gaps' left by the other schemas, such as what biological dysfunctions are associated with theory of mind deficits, and how genetic and epigenetic changes can influence neurological and socio-cognitive development. Therefore, on this account, the disorders currently identified as the autism spectrum are best explained as complex conditions with biological, cognitive and social features, and are the result of complex interactions between causes at these levels of description. I argue in what follows that such an explanatory framework may be the most powerful for explaining and predicting the causes and progression of psychiatric conditions like those on the autism spectrum.

6.2.2 Integrated Explanation Schema and the Criteria for Adequate Explanation Patterns in Psychiatry

An integrated schema may better represents what the causal structures of psychiatric conditions seem to be, based on the limited causal data that has been obtained. That is, an integrated schema seems to fit better with the heterogeneous nature of the psychiatric disorders listed in the various categories of the DSM. Recall that some conditions listed in the DSM seem to be primarily biological in nature, such as ASD and schizophrenia, and thus biological primary causes such as genetic mutations, epigenetic mutations, and neurological malfunctions may bear most of the explanatory weight. However, some disorders seem to be primarily social in nature, such as eating disorders or post-traumatic stress disorder. In the case of these conditions, social or environmental primary causes may carry much of the explanatory weight. However, each of the specific schemas will still have the same basic explanatory pattern, and can be incorporated into a larger explanatory framework where the primary causes and their effects are linked to malfunctions or changes at the other levels identified in the integrated schema. Elucidating how the primary causes of each disorder influence the other levels identified in the schema will help differentiate

each disorder in terms of its primary causes, as well as its social and cognitive symptoms. Although each condition in the DSM likely has biological, cognitive and social features, and thus could potentially be explained using an integrated schema, the specific causal explanation for each disorder will be distinct, and may privilege or emphasize one or more levels over others, such as the explanation of Rett's disorder just discussed. Thus, although this integrated schema is complex and identifies multiple potential primary causes, it meets all the criteria for adequate explanation patterns outlined in chapter 2.

The first criteria that adequate schemas must meet is empirical support. While the causal data is still ongoing, there is good empirical support for most of the causal theories of ASD discussed in this thesis. If these theories continue to be supported by more research, clinicians and theorists can help to further identify and elucidate the parts of the inter-level mechanism, and how these parts interact. Causal theories such as the theory of mind hypothesis has good empirical support, as do the epigenetic data on Rett's disorder. Further, the mirror neuron research and the genetic studies discussed in chapters four and five may gain further support as research into the causes of ASD continues. If the causal theories included in the explanation pattern given below continue to gain good empirical support, this integrated schema may be the best way to capture the fundamental biological, cognitive and social features of these conditions. Even if continuing research reveals very different causes and processes in the development of each disorder on the autism spectrum, an integrated schema will likely be the most powerful for explaining these conditions and identifying each of them in the clinical population.

The second criterion is internal consistency. Using an integrated schema may help to link the various causal theories of ASD in a consistent and powerful way. Although the causal processes are complex, clinicians may be able to explain multiple primary causes could interact to produce the characteristic symptoms of conditions like ASD. The story goes something like this: certain genetic and epigenetic changes cause impairments in neurological development. These impairments in neurological development result in patterns of neurological dysfunction, such as a malfunctioning mirror neuron system. Certain patterns of neurological dysfunction like a malfunctioning mirror neuron system are implicated in the development of certain cognitive impairments, such as an underdeveloped theory of mind. Cognitive deficits like an underdeveloped theory of mind cause the characteristic symptoms of ASD, which are impairments in social interaction and communication, and stereotyped behaviours. On this account, the characteristic symptoms of ASD are caused by the interactions between causes at the biological, cognitive and social levels. Interactions between causes at multiple levels occur through processes such as genetic and epigenetic mutations, which can affect neurological development and functioning. The characteristic symptoms of ASD, impairments in social interaction and communication and stereotyped

behaviours, can be mitigated through intensive behavioural interventions, which are designed to scaffold the developmental precursors to theory of mind, such as imitation and pretense. Thus, while speculative, this schema is potentially internally consistent.

However, an explanation of psychiatric conditions like those on the autism spectrum must also have predictive validity. While there is still much that is unknown about the causal structures and causal processes that result in the development of psychiatric symptoms, the causal data that has been gathered seems to indicate that these phenomena are not syndromes with unity, but are the result of multiple causes interacting to produce particular symptoms. Thus, psychiatric conditions such as ASD are complex phenomena, and being able to identify their characteristic features at multiple levels is a more powerful way to identify these conditions, since there is no one primary cause or tell-tale sign that a patient has a particular psychiatric disorder. Therefore, as many theorists discussed in this thesis argue, psychiatric conditions are best explained as breakdowns in complex interactive mechanisms. The integrated schema may be powerful enough to be able to explain and predict the development and progression of disorders such as those on the autism spectrum, even though there is more than one primary cause involved (with the exception of Rett's disorder) and these primary causes occur at different levels of description. Thus, an integrated explanation pattern is *parsimonious*, even though it identifies more than one primary cause, and the primary causes it identifies are at different levels of description.

Since psychiatric conditions like those on the autism spectrum are not syndromes with unity, the causal processes and the development of the cognitive and behavioural symptoms do not have a law-like relationship. However, the casual processes involved in the development of conditions like ASD can be explained in terms of the relations between the parts of a mechanism, the activities of those parts, and the interactions between the parts of the mechanism to produce the phenomena in question, in this case the characteristic symptoms of ASD. Like most of the other schemas discussed in this thesis, the integrated explanation pattern is *mechanistic*.

However, like conditions such as major depression and alcoholism discussed above, the development and progression of the conditions on the autism spectrum (with the exceptio of Rett's disorder) is the result of complex interactions between the primary causes identified at the biological, cognitive and social levels. Thus, the fourth criterion for adequate explanation patterns is that not just a mechanistic explanation, but a *multi-level mechanistic explanation*. As indicated above, the integrated explanation pattern is multi-level, since it includes the biological levels (genetic and epigenetic causal theories), the cognitive level (cognitive causal theories like theory of mind), and also mentions the social or environmental levels of explanation as well.

The final criterion for adequate explanation patterns for psychiatric conditions is that it must not just identify the primary causes at multiple levels, but explain the *interaction* between these primary causes. The integrated schema is interactionist, since the genetic, epigenetic, cognitive neurological, cognitive and social levels interact with each other to produce the symptoms of psychiatric conditions like ASD. If processes like epigenetics and epigenetic changes in neurological functioning were better understood, the link between epigenetic changes and neurological dysfunction can explain some of the causal interactions. Thus, processes like epigenetics can explain some of the interactions between causes at different levels of description, such as the interaction between causes at the neurological and genetic levels. If the interactions between neurological dysfunction and patterns of cognitive impairment were better understood, such as how a malfunctioning mirror neuron system is associated with a theory of mind deficit, that would also help to explain the interactions between the neurological and cognitive levels. If this integrated explanation schema does give the right basic structure to the explanation (identifying a stable cluster of causal properties), and these clusters of causal properties can be understood mechanistically, then further research can begin to elucidate the interactions between causes at multiple levels included in the mechanism.

While an integrated schema seems to have the advantage of including multiple levels, there are also concerns about such as what levels to include, and how each or all of these levels will be emphasized or integrated into the overall schema. In the case of psychiatric disorders such as ASD, alcoholism, and major depression, we can restrict the levels included in the explanation to those that seem to be more fundamental than others. For instance, the genetic, epigenetic, neurological, cognitive neurological, cognitive and social levels seem to be implicated in the development of most psychiatric disorders, including ASD. These levels of description are also identified in the more powerful explanation schemas discussed in this thesis. However, some psychiatric disorders may not have primary causes at all of the levels identified in the integrated explanation pattern generated here. For example, disorders such as Down's syndrome and Rett's disorder have primary causes at the genetic and epigenetic levels, which are a third 21st chromosome in the case of Down's syndrome and a mutation in the MeC2P gene in the case of Rett's. On the other hand, some psychiatric disorders, such as eating disorders, may be explained primarily in terms of social and cognitive causes, and thus the biological components of these conditions will not have as much explanatory weight in the schema. Finally, other disorders such as major depression, anxiety, and eating disorders may need all of these levels to full elucidate the causal processes involved in their development.

Thus, as more evidence comes in, more powerful and supported causal theories can help determine what the fundamental causes are, and thus what aspect(s) of the inte-

grated schema will be emphasized in the explanation of a particular condition. All psychiatric disorders can be explained in terms of biological, cognitive and social features, but certain causal explanations will be more robust or powerful than others, and thus these explanations will be emphasized when explaining and identifying these conditions, while the other aspects will flesh out the complete picture of the causal structure and processes involved in the development of a particular psychiatric condition. There are also levels within the levels included in the integrated schema, for instance the neurological level can be further broken down into levels such as neurological systems, individual neurons, neurotransmitters, neuromolecular, etc. These more specific levels may/can be integrated and/or emphasized in explanations of particular psychiatric disorders, depending on what causal evidence is found for a particular condition. Therefore, the integrated schema generated above may be the most powerful explanation schema for psychiatric disorders in general, and the causal evidence from ongoing research can also help inform what levels to include in an explanation of a particular disorder.

I have argued that integrated schema is the most powerful explanation of conditions such as those on the autism spectrum, because it may be able to differentiate between psychiatric conditions better than previous schemas discussed in this thesis. Thus, if the integrated schema is a more discriminating explanation pattern than previous schemas, it may help improve the explanatory and predictive power of psychiatric diagnosis. If this integrated schema is indeed the most powerful, this shows that a cause-based diagnostic system for psychiatry is feasible, even though the causation in psychiatric conditions is complex and heterogenous. It helps to look at more ambiguous or tougher diagnostic cases to show that the integrated schema is the most powerful one for explaining the complex causation involved in the development of psychiatric disorders. If an integrated schema can help with these ambiguous or tougher cases, it may be robust enough to inform a cause-based diagnostic and classification framework in psychiatry, and may help improve the explanatory and predictive power of psychiatric diagnosis and classification. Diagnostic categories may have better validity and reliability if diagnostic categories were informed by integrated causal schemas such as this one.

Chapter 7

The Ethical Implications of Cause-Based Diagnosis of Psychiatric Conditions

7.1 Introduction

The preceding chapters addressed the metaphysical and epistemological issues that a cause-based diagnostic and classification system for psychiatry present. I investigated whether the explanatory and predictive power of the diagnosis and classification of psychiatric conditions such as those on the autism spectrum could be improved if psychiatry adopts a cause-based framework in place of a symptom-based framework. The causation involved in the development of psychiatric conditions is complex and multi-directional, and thus only some explanatory patterns will be powerful enough to inform a cause-based diagnostic and classification system. I examined the difficulties in generating causal explanations of psychiatric conditions such as those on the autism spectrum, and developed a preliminary list of criteria for adequate explanation patterns in psychiatry. I then used these criteria to evaluate different explanatory patterns of ASD from past and present conceptual frameworks in psychiatry and clinical psychology. I argue only explanatory patterns that are mechanistic and identify primary causes interacting at multiple levels will be able to adequately capture the complex and multi-directional causation involved in the development of conditions such as ASD. Chapter 6 argued that an integrated explanatory pattern informed by fields such as epigenetics, genetics, cognitive neuroscience and cognitive psychology may meet the criteria for adequate explanation patterns outlined in chapter 2,

and thus may be able to help inform a cause-based diagnostic and classification system in psychiatry.

I also briefly argued that explanatory patterns that meet the criteria for adequate explanation patterns in psychiatry, such as an integrated explanatory pattern, may help to further justify two-stage realist accounts of psychiatric conditions, such as Wakefield's Harmful Dysfunction analysis (e.g. Wakefield, 1992, 1997, etc.). While developing a realist account of psychiatric conditions is beyond the scope of this thesis, chapter 1 argued that psychiatric disorders may have the same causal structure as many diseases and disorders treated by other branches of medicine. If this is correct, psychiatric conditions are as "real" as conditions such as lung cancer and type-II diabetes.

However, there are several ethical issues that arise if psychiatry adopts a cause-based diagnostic framework. These ethical issues are of two types: those that concern the diagnostic practices themselves, and those that concern the policy and social implications of changes to classification and diagnosis. I will discuss each of these ethical concerns, both with respect to autism spectrum disorders, but also with respect the treatment and diagnosis of psychiatric conditions in general. Many of the arguments put forward in this thesis can be applied to the classification and diagnosis of other psychiatric disorders, and a shift to a cause-based diagnostic framework will affect the legal and social status of these conditions as well.

In what follows, I examine whether explaining the conditions on the autism spectrum using an integrated explanation pattern can help to address the ethical issues of resource allocation, meeting and managing health care needs of individuals with ASD in a more effective way, and reducing the harmful false beliefs about these conditions. This thesis argues that basing diagnostic categories on causal theories from fields like epigenetics and neuroscience may make diagnostic boundaries less arbitrary, increase the explanatory and predictive power of diagnosis, and increase the effectiveness of treatment.

First, I discuss the ethical dilemmas that can arise in diagnostic practices themselves. If the disorders on the autism spectrum are best explained as the result of breakdowns in complex biologically-based inter-level mechanisms, a better understanding of the genetic, neurological and developmental characteristics of these conditions may re-draw diagnostic boundaries. If diagnostic categories are based on causal theories from fields such as epigenetics and neuroscience, diagnostic categories may have increased explanatory and predictive power, which increases the reliability and validity of psychiatric diagnosis. More accurate diagnosis may reduce the amount of time and resources needed to obtain a definitive diagnosis of ASD, and may reduce the costs involved in clinician and physician visits in the diagnostic process. Further, more accurate diagnosis may allow children with ASD

and their families to have access to treatment programs earlier in the child's development, which can make treatment more effective. Finally, I analyze how the costs of diagnosis and treatment of autism spectrum disorders will be affected by a cause-based system. I argue that while some of the costs involved in diagnostic procedures will increase, the overall costs over the individual's lifespan will be reduced.

The second issue I discuss is how the possible changes to diagnostic practices discussed in this thesis will influence legislative and policy decisions regarding the allocation of resources for the treatment of ASD. There have been several cases brought before the provincial and Supreme Courts of Canada petitioning for increased funding for Intensive Behavioural Intervention/Applied Behavioural Analysis treatment programs. Most of the provinces provide some funding for IBI/ABA programs, but the amount and extent of funding varies between the provinces. I discuss the possible impact of a shift to a cause-based diagnostic system and a better understanding of the causes of ASD using two recent Canadian cases: *Auton v British Columbia* and *Wynberg v. Ontario*. I suggest that explaining ASD as a complex biologically-based disorders may help support for the plaintiffs' claims that the deficits and characteristic symptoms of these conditions are severe enough and unique enough that treatment should be considered medically necessary under the *Canada Health Act*, and the provinces of British Columbia and Ontario should expand their funding of treatment programs.

The third issue I discuss is the social implications of a cause-based classification and diagnostic framework for psychiatric conditions. I discuss the potential changes to lay concepts of both ASD and psychiatric conditions in general. I argue that explaining ASD as the result of breakdowns in biologically-based inter-level mechanisms may help to further reduce the prevalence of 'myths' regarding these conditions that still have a significant impact on lay views of these conditions. A better understanding of the causes and progression of these conditions and how treat them may help to finally dispel the false, but still popular, beliefs that autism is caused by the MMR vaccine or poor parenting. Understanding ASD as biologically-based conditions may also help to alleviate feelings of responsibility parents might have for the diagnosis of autism their child receives. Finally, if other psychiatric conditions are also best explained using an integrated explanation pattern, such causal explanations may help to further reduce the stigma associated with other conditions in the DSM.

A shift to a cause-based diagnostic and classification framework will not automatically resolve the many ethical issues involved in the diagnosis and treatment of psychiatric disorders, and in the allocation of resources for diagnosis and treatment. However, I argue that while some direct medical costs involved in diagnosis and treatment may increase, the overall amount of time, resources, financial costs, legal boundaries, and social stigmas

associated with ASD and other psychiatric conditions are reduced. Thus, a shift to a cause-based diagnostic and classification framework may have positive consequences not just in the possible improvements in diagnosis and treatment, but in the legal and social realms as well.

7.2 Ethical Issues in the Diagnosis and Treatment of Autism Spectrum Disorders

As stated in earlier chapters, the current classification and diagnostic system is plagued with problems regarding diagnostic reliability and validity. Currently, ASD represent a spectrum of conditions included in one diagnostic category, based on the similarity of symptoms across these conditions. Chapter 1 discussed the five disorders currently identified as part of the autism spectrum: autistic disorder, childhood disintegrative disorder, Rett's disorder, Asperger's syndrome, and pervasive developmental disorder not otherwise specified (PDD-NOS).

Chapter 2 discussed the current diagnostic criteria for ASD. These criteria overlap with the criteria for other disorders of childhood, such as mental retardation and childhood coordination disorder, since ASD share a number of symptoms with these conditions. Thus, there is much ambiguity in the diagnostic boundaries of ASD as a spectrum, and in the boundaries between disorders on the spectrum. Children with autistic disorder often have IQs and behavioural impairments similar to children with mental retardation, and share language and motor impairments with other disorders of childhood. On the other end of the spectrum, children and adults with Asperger's syndrome may seem simply eccentric, shy or awkward, and may not be diagnosed with a disorder at all if the symptoms are mild. Such blurring at the ends of the continuum that represents ASD can make proper diagnosis and effective treatment difficult.

Further, much of the costs associated with autism spectrum disorders involve the diagnostic process. Misdiagnosis can incur more costs for the parents of children with ASD, for clinicians, and for society at large. An incorrect diagnosis can lead to the wrong treatment being prescribed, which may not be designed to address the unique deficits that are characteristic of ASD. Also, a re-diagnosis requires more interviews with clinicians and physicians, and in the case of ASD, a reassessment of what sort of special education a child will receive. Thus, re-diagnosis can be costly, both in terms of direct medical costs and the costs of educational assessment and assistance.

7.2.1 Diagnostic Accuracy and Better Access to Treatment

Integrated patterns of explanation can be used to elucidate the causal structures of many different types of psychiatric disorders, since the different explanatory levels can carry more or less weight depending on the nature of the primary causes. For instance, some disorders may have more or most primary causes at the social level, rather than the biological or cognitive, such as post-traumatic stress disorder or adjustment disorders. On the other hand, conditions like alcoholism discussed in chapter 6, can have primary causes at the biological, cognitive and social levels. Integrated explanation patterns for psychiatric conditions may generate more refined diagnostic criteria, and may help to draw sharper diagnostic boundaries around these conditions.

Causal theories from epigenetics, genetics, cognitive neuroscience and cognitive psychology may help to better determine whether a child should be diagnosed with autism spectrum disorder or another disorder of childhood even if the symptoms are similar. Further, causal theories can distinguish between the disorders on the spectrum. Chapter five discussed the example of Rett's disorder, an autism spectrum disorder affecting only female children. Rett's disorder shares the same pattern of deficits in the same three socio-cognitive domains as the other disorders on the spectrum, but shares only one aspect of the epigenetic mutations thought to cause the other conditions currently identified as the autism spectrum. Rett's disorder has a single epimutation as its cause, while the other disorders on the autistic spectrum have a more complex origin. Thus, identifying the epimtuation involved in the development of Rett's disorder could help clinicians to diagnose female children exhibiting the characteristic symptoms of ASD with more certainty. More accurate diagnostic categories for the disorders on the autism spectrum can ensure that children with these conditions are diagnosed and start treatment as soon as possible. If female children with autistic symptoms are diagnosed as having Rett's disorder in a more efficient and certain manner, affected females can enter behavioural intervention programs earlier in their socio-cognitive development, thus reaping more benefit from these treatment approaches.

The most effective and popular treatment for children ASD is Lovaas-style Intensive Behavioural Intervention/Applied Behavioural Analysis programs. These programs use learning theory and reward contingencies to help the children develop some ability to communicate and interact, and to reduce self-injurious or maladaptive behaviours. These treatment programs are designed to scaffold and develop socio-cognitive skills like imitation and verbal communication, which many researchers argue are important milestones in the development of theory of mind (Rogers & Williams, 2006; Baron-Cohen et al, 2000). These skills are delayed or malfunctioning in children with ASD, and interventions are

designed to develop these skills through the techniques of behavioural management, reward contingencies and structured interactions with a therapist.

However, there are varying results regarding the efficacy of Lovaas-style intervention programs, and the overall success achieved by such programs has been debated. Higher estimates suggest that this style of therapy may be effective in as many as 40% of cases (Lovaas, 1993, 1987; Motiwala, Gupta, Lilly, Ungar and Coyte, 2006). However, there are more conservative estimates of the success rate of such programs indicating that sometimes very limited progress is made, and there are significant individual differences between the progress made by children in these programs (Motiwala et al 2006). However, Lovaas style therapy remains the most popular and effective treatment, even if its success is limited to a smaller number of cases.[1]

Lovaas (1993; 1987) stated that the earlier the child receives treatment for ASD, the more effective in ameliorating the severity and progression of symptoms. He (1987; 1993) states that applied behaviour analysis is effective in improving IQ, language skills, social interaction and overall levels of functioning if the child receives 40 or more hours a week for more than 2 years. In the discussion of his results, Lovaas (1987) argues that the younger the child when the intervention is started, the less severe the symptoms, and the closeness of the protracted mental age to the child's chronological age predict greater outcomes for treatment and a better prognosis. Motiwala et al (2006) also found that individuals with ASD who received treatment early in life needed less long-term care as adults, and gained more dependency-free years in their adult lives.

Thus, early and accurate diagnosis for young boys and girls may help to ensure that the predicting factors for successful interventions are in the child's favour. Further, a more accurate account of the underlying causes of the deficits in communication, social interaction and behaviour can help design more targeted and effective treatments. In turn, a better understanding of the development of the impairments in the three socio-cognitive domains can help to determine what the goals of the intervention should be, and what cognitive-behavioural techniques are best suited to ameliorating the symptoms of ASD.

7.2.2 Parent and Educator Interactions with Children with ASD

A better understanding of the causes and development of the symptoms of the conditions on the autism spectrum may help parents of children with ASD to interact with their

[1]it should also be noted that there are newer systems of IBI/ABA, such as the Denver Start Model developed by Sally Rogers and her colleagues that also attempt to scaffold socio-cognitive and language skills. For instance, see Rogers & Dawson (2009).

children in less stressful and more effective ways. First, understanding these conditions as complex conditions involving causes at the biological, cognitive and social levels can help parents have a better idea of the nature of their children's symptoms, why they occur, and how best to deal with their manifestation. Teaching parents how to manage the symptoms of these conditions in the home can reduce the psychological and emotional stress for both parents and children. Second, parents can gain the knowledge that the condition is the result of a complex interaction between genetic, neurological, cognitive and environmental factors, and not the result of bad parenting or their choice to give their child the MMR vaccine. Third, increased understanding of the causes of these conditions may reduce the amount of alternative therapy approaches parents try, which reduces the direct non-medical costs and stresses associated with ineffective approaches.

A more powerful explanation of the causes of ASD may also affect educational costs and services associated with these conditions. First, explaining these conditions using an integrated explanatory pattern may help to prevent anti-immunization behaviours on the part of parents worried about the link between autism and the MMR vaccine. If anti-immunization behaviours are reduced, the administrative problems and costs that arise with unvaccinated children in the school system are also reduced, as is the chance of a spread of dangerous childhood diseases. Second, a better explanation of the causes of ASD may also change the way these disorders are understood in relation to other disorders of childhood, and the nature of educational resources available for children with ASD. Causal theories from fields such as epigenetics and neuroscience may provide more insight into the nature of the socio-cognitive deficits that define the conditions on the autism spectrum. Thus, educators may gain a better understanding of what sort of resources should be allocated to manage the symptoms of ASD in special education and the regular classroom. Further, a child with ASD who enters treatment in early childhood treatment may improve enough in IQ, language and social interaction to be integrated into the regular classroom environment. More children with ASD who are able to function in the regular classroom reduces the number of children needing special education services, thereby reducing some of the costs of these services. Earlier access to IBI/ABA programs depends in part on a more accurate diagnosis of these conditions, which I argue may result from a shift to a cause-based framework informed by adequate patterns of explanation for psychiatric conditions.

7.2.3 Managing the Costs of Diagnosis and Treatment for Autism Spectrum Disorders

A final report was submitted to the Standing Senate Committee on Social Affairs, Science and Technology in March of 2007 regarding the care and treatment currently available for Canadians with ASD and their families. The report (2007) states that the Committee "heard repeatedly during its hearings that ASD families often have a single income earner because the second parent must stay home to care for the affected child or children. Frequently, the earning parent must also take on a second job in order to pay the high cost of ASD care and treatment. Witnesses explained that this results in pushing the income earner into a higher tax bracket, reducing the effect of the medical expense tax credit (p. 27)." The report (2007) further states that "The Committee fully supports the view expressed by families with autistic children and autistic individuals themselves that governments must pay now; otherwise, they will pay later. We believe that the latter is simply not an option (p. 30)."

Parents of children with ASD face expenses in several areas, both financial and emotional. Financial expenses individuals with ASD and their families face are in the form of direct medical costs, non-direct medical costs, productivity costs in terms of loss of pay, and the obstacles to achieving higher-paying employment. Direct medical costs include things like physician and specialist visits, drugs, transportation to physician visits and therapy sessions, and behavioural interventions. Direct non-medical costs include things like out-of-home services, developmental services, day programs, and child care. Indirect costs include the loss of wages and opportunities for individuals with ASD, which occur as a result of the symptoms of these conditions. The parents and family members of people with these disorders also experience a loss of wages and employment opportunities, since much of their time is devoted to caring for these individuals (Ganz, in Moldin & Rubenstein, 2006).

Ganz (2006) highlights several other areas in which there are heavy costs for individuals with ASD and their families. These areas include 1) legal costs to secure services, 2) value of lost productivity of individuals other than parents, 3) level of psychological stress that people with ASD and their families endure 4) costs of genetic testing, 5) full costs of alternative therapies, such as special diets or vitamin regimens, 6) costs associated with adverse outcomes of potentially dangerous treatments, and 7) effects of changes in immunization-avoidance behaviours. In this section, I argue that while a cause-based framework may increase some costs involved in the treatment of these disorders, it will reduce costs in several areas. The brief cost-benefit analysis I give here indicates that a shift to a cause-based diagnostic framework is a viable solution to some of the financial

problems related to diagnosis and treatment of these disorders.

Interventions are intensive behavioural modification programs that are expensive and time consuming. Jacobson, Mulick, & Green (1998) estimate that it costs approximately $37, 537 US per child per year for behavioural interventions. Hildebrand (1999) estimates that the cost for Canadian citizens seeking treatment for ASD is approximately $45, 053 CAN in 1999, which Ganz estimates to be closer to $65,000 CAN in 2000. Currently, IBI/ABA programs are partially funded by the provinces, but parents and family members still pay a significant amount for such treatment. However, the high costs of the interventions are not the only financial obstacle individuals with ASD and their families face.

As stated in the report to the Standing Senate Committee on Social Affairs, Technology and Science (2007), parents of autistic children often have to switch to lower-paying but more flexible employment in order to care for them. Such an occupational change can place financial constraints on the amount and type of resources parents are able to secure for their child. Ganz (2006) estimates that the average income loss for parents of children with ASD to be up to $46,033, depending on the level of the child's disability. However, ASD are pervasive disorders that will continue to manifest throughout the individual's life time, and there is a pressing need to provide services for individuals with these disorders as they reach adulthood. However, care of adults with ASD can also be expensive. For instance, Ganz (2006) argues it can cost up to $3.2 million dollars US to care for an individual with autism spectrum disorders over the lifespan, and about $35 billion dollars to care for all individuals with ASD in the United States across their lifetimes. For Canadians, Hildebrand et al (1999) estimate that the cost of day programs for adults with ASD to be $13, 168 CAN to $25, 937 CAN, depending on the individual's level of disability. They (1999) also argue that special education of children with ASD can cost up to $28, 216 dollars, which Ganz adjusts to up to $39, 599 in Canadian rates from the year 2000.

The figures discussed above are based on the current state of diagnostic practices, and on the type of treatment typically available to individuals with autism spectrum disorders. If a cause-based framework is informed by causal theories from fields such as epigenetics, there will likely be a larger role for genetic testing and neuroimaging technologies in the diagnostic process. The cost of genetic or neurological testing procedures is likely to be great, and the availability of such resources for diagnostic and treatment practices may place additional strain on the already scarce resources allocated to mental health care and the services focused on those with developmental delays. Thus, a cause-based diagnostic system informed by an integrated explanation pattern may increase direct and non-direct medical costs for individuals with ASD and their families. However, a cause-based diagnostic framework can help to reduce the costs of autism treatment in several of

the areas Ganz (2006) discusses.

I argue that cause-based diagnosis of ASD may have better explanatory and predictive power, which may make diagnosis more accurate. If diagnosis is more accurate, individuals with ASD may save costs in terms of re-diagnosis, re-assessment of one's legal rights based on re-diagnosis, and the amount of care individuals with these conditions may require over their lifetimes. Further, more accurate diagnosis may help to secure earlier access to behavioural interventions, which may reduce the amount of treatment and resources these individuals need as adults. Motiwala et al (2006) conducted a cost benefit analysis of the financial gains and therapeutic benefits of IBI/ABA programs in Ontario. They (2006) argue that even if such therapy has limited success, government funding of IBI/ABA programs saves significant costs, not just for the individuals with ASD and their families, but for the provincial government as well. Motiwala et al (2006) argue even the current amount and duration of IBI/ABA treatment funded by the Ontario government can result in a gain of 'dependency free years' where individuals with ASD need less health care and social assistance from the province as adults. Further, they argue that expanding IBI care in Ontario past the age of 6 results in more dependency free years, and more improvements in cognitive and social functioning across individuals with these conditions. A gain in dependency-free years saves costs for the Ontario government in the long-term care of individuals with ASD over their lifetimes. These theorists (2006) estimate that expanding current the funding of ABA/IBI programs in Ontario could result in a savings of $45,133,011 in 2003 Canadian dollars, based on gains in dependency free years by individuals with ASD treated with these programs.Thus, early access to behavioural intervention programs may reduce the number of physician visits, reduce the amount of time and resources parents must spend to help manage the symptoms as the child develops, and the total cost of care for individuals with ASD throughout their lifespans.

7.3 Legal Issues and the Social Status of ASD and Psychiatric Conditions

The last section argued that a cause-based diagnostic framework may save costs and resources involved in the treatment and care of individuals with ASD, their families, and the health care and education systems. However, the legal system plays a large role in how much access one has to the diagnostic and treatment resources. A cause-based diagnostic classification and diagnostic framework may change the boundaries and clustering of disorders drastically based on a better understanding of the causes of psychiatric conditions. Re-drawing diagnostic boundaries can sometimes lead to a change in one's legal status,

which may change the amount of resources and access to treatment programs and social services. This section explores two areas in which theories and data from fields such as epigenetics, genetics, cognitive neuroscience and cognitive psychology could influence legal decisions regarding the treatment and care of individuals with ASD in Canada. First, I discuss the possible impact causal theories of the conditions on the autism spectrum on legal claims for increased funding for treatment programs. Causal theories of autism spectrum disorders could play an evidentiary role in legal cases such as those discussed below. Plaintiffs in Canadian legal cases such as *Auton v British Columbia* and *Wynberg v Ontario* are fighting for increased resources from these provinces for the treatment of these conditions, on the grounds that ASD involve impairments severe enough, and unique enough to qualify for expanded funding for IBI/ABA treatment programs. An increased understanding of the causes of autism spectrum disorders could be used as evidence of the unique needs of children with these conditions and the importance of early access to treatment.

Second, I discuss the possible impact of cause-based diagnosis and a better understanding of the causes of ASD on lay beliefs about these conditions and their causes. The beliefs that ASD are caused by bad parenting or vaccinations still influence lay concepts and explanations of these conditions. I argue that using an integrated explanation pattern to identify the causes and progression of ASD can further reduce the tenability of explanations such refrigerator mothers or vaccination, which may reduce the influence of these false causal theories in popular culture. While the refrigerator mother hypothesis has been discredited by psychiatrists and clinicians, some parents still believe that their parenting skills are part of the reason for their child's symptoms. Feelings of responsibility or guilt on the part of parents can create more psychological and emotional stress in already difficult situations, and can negatively affect the interactions between these parents and their children. If parents had a better understanding of the causes of these condition, feelings of responsibility and stress levels may be reduced. Also, disproving the vaccine hypothesis may reduce the amount of anti-immunization behaviours on the part of parents and family members, which saves both financial and emotional costs that result from not vaccinating children. Further, a reduction in anti-immunization behaviours may reduce the risk of the spread dangerous childhood diseases such as measles, mumps and rubella, which can be a danger if children are unvaccinated.

7.3.1 Improving the Treatment of Autism Spectrum Disorders in Canada

The final report to Standing Senate Committee on Social Affairs, Science and Technology states "[f]amilies with autistic children in Canada are facing a crisis (2007, p. 11)." Although the treatment and management of the symptoms of ASD is crucial for increasing the quality of life for individuals with these disorders, the report states "families must often pay out of their own pockets for a very large portion of expensive autism therapy whose cost may reach $60,000 per year because provincial and territorial jurisdictions offer only limited financial assistance (p. 11)." The report also discussed the need for specialized care and resources devoted to the treatment and care of individuals with ASD. Several parents and advocacy groups argued that the funding allocated for children and adults with disabilities is not sufficient for the "diverse and substantial needs [individuals with ASD] have (p. 18)" and further "that ASD is not always eligible for tax credits or deductions intended for disability or medical expenses (p. 18)." Further, the committee also heard testimony regarding the wait time involved in diagnosis and assessment, which further delays access to treatment programs. Sometimes, parents have to wait months, or even years, for a diagnosis of ASD only to find that their child was now too old to qualify for government funded programs.

Because of the extent of the wait times and expenses associated with treatment programs, parents and family members of individuals with ASD have filed lawsuits arguing that treatment with IBI/ABA programs should be deemed 'medically necessary' under the *Canada Health Act*, and that increased funding for these programs should be required of provinces such as Ontario and British Columbia. For instance, in 2000, four families from British Columbia filed a lawsuit against the province, stating that the treatment of autism spectrum disorders in the form of intensive behavioural interventions (IBI) should be considered 'medically necessary' under the Canada Health Act. Plaintiffs in *Auton v. British Columbia* argued that not funding IBI programs for children with ASD constituted discrimination under sections 7 and 15 of the *Charter of Rights and Freedoms* (see *Auton et al. v. British Columbia* 2000), since funding for treatment and services for children with other disabilities is provided by these provinces. British Columbia Superior Court Madam Justice Allan found that the province's narrow interpretation of medically necessary treatment was discriminatory to individuals with ASD on the basis of their disability (Auton, 2000 paras 126, 129; Tiedemann, 2008). Madam Justice Allan concluded that the province's decision to *not* fund IBI programs was based on the premise that treatment for children with ASD was of limited success and effectiveness. However, she found that access to early IBI programs was necessary to reduce the marginalization and exclusion of

children with ASD in society (Auton, 2000 para 127; Tidemann, 2008).

The province of British Columbia appealed the British Columbia's Superior Court's decision, and the case was heard by the Supreme Court of Canada in 2004. Madam Chief Justice McLachlin found that while the *Canada Health Act* requires that provinces fund 'core services' that are medically necessary, the *Act* does not require that all medically necessary treatments are funded. Currently IBI/ABA programs for the treatment of ASD lies outside the 'core' services covered under the CHA (Auton, 2004 paras. 35,36; Tiedemann, 2008). Madam Chief Justice McLachlin found "[t]here is no evidence suggesting that the governments approach to ABA/IBI therapy was different than its approach to other comparable, novel therapies for non-disabled persons or persons with a different type of disability. In the absence of such evidence, a finding of discrimination cannot be sustained (Auton, 2004, para 62)." Thus, the Supreme Court of Canada found that the province of British Columbia was not required to fund IBI/ABA programs under the *Canada Health Act*.

Tiedemann (2008) states that while the plaintiffs in *Auton v. British Columbia* lost their case, the case did bring out a number of positive consequences. First, she states

> the trial motivated the province of British Columbia to expand a small pilot treatment program into policy. That province now delivers autism intervention services through three programs: Autism Funding, Under Age 6; Autism Funding, Ages 6-18; and Early Intensive Behavioural Intervention (EIBI). Under the first program, families are allocated up to $20,000 per year to purchase autism intervention. Under the program for children aged 6-18, families are allocated up to $6,000 annually; and under the EIBI program, some treatment and intervention services for children under age 6 are delivered through contracted agencies (p. 9).

Second, the case brought the issue of government funding of ASD treatment into the public eye. Tiedemann (2008) notes that an Ipsos-Reid poll taken after the conclusion of *Auton* trial indicated that 84% of Canadians support public funding of IBI programs. Third, there were a number of petitions placed before Parliament to expand the CHA to include IBI/ABA as medically necessary treatment. Fourth, the *Auton* case motivated other families in provinces such as Ontario to petition the courts for increased funding for the treatment of ASD.

For instance, the Ontario Superior Court of Justice heard *Wynberg v. Ontario*, where the plaintiffs argued that access to IBI/ABA programs was a right under the *Education Act* (*Wynberg v. Ontario*, 2006; Tiedemann, 2008). While the province of Ontario does fund

IBI/ABA programs, the funding is only provided for children aged 2-5. Before and after ages 2-5, parents and families must bear the cost of treatment and services. The plaintiffs in *Wynberg v Ontario* argued that the cut-off age of 5 discriminated against children with ASD outside of that age bracket based on their age. Madam Justice Kiteley ruled in favour of the plaintiffs, and rewarded damages to compensate the plaintiffs for the costs of past and future IBI/ABA treatment (*Wynberg,* 2006 paras. 4, 792, 808, 871-872; Tiedemann, 2008). However, the government of Ontario successfully appealed the ruling by Madam Justice Kiteley in the Ontario Court of Appeals. On July 7 2006, the Ontario Court of Appeals found that plaintiffs in *Wynberg v Ontario* had failed to adequately establish their claims of age discrimination. The families involved in the *Wynberg* case attempted to have the decision of the Ontario Court of Appeal reversed in the Supreme Court of Canada, but the Supreme Court refused their application for an appeal on April 12 2007 (Tiedeman, 2008).

While the families in both *Auton* and *Wynberg* ultimately lost their cases in the Supreme Court, these cases are interesting to examine for a number of reasons. These lawsuits were filed to change policies regarding the allocation of resources to children with ASD based on these disorders' severity, and on their similarity and differences of the impairments in ASD compared to other developmental disorders. Causal theories and data regarding the conditions on the autism spectrum may play an evidentiary role in any future lawsuits filed on grounds similar to those in *Auton* and *Wynberg*. Explaining the conditions on the autism spectrum using an integrated pattern may help to provide support for the pressing need for children with ASD to enter IBI programs as soon as possible, for which an accurate diagnosis is necessary.

First, an integrated explanation pattern for ASD may help to identify the interactions between the biological and environmental aspects of these conditions, which emphasizes the importance of the early environment in the development of these conditions. If ASD are best explained as conditions that result from breakdowns in complex interactive mechanisms, the early environment plays an important role in the development, progression and severity of the symptoms of these conditions. Second, a better understanding of the causes and progression of these conditions may help to make distinctions between the impairments and needs of individuals with ASD compared to other developmental disorders. A better understanding of the similarities and differences to other developmental disorders provide more data that can inform legal decisions when comparing the needs of individuals with ASD to those with other conditions, to help determine whether treatment for ASD should be considered 'medically necessary.' According to the autism research discussed in earlier chapters, the conditions on the autism spectrum are similar to other developmental disorders in that they are all biologically-based disorders with strong genetic components

in their etiologies. Both ASD and other developmental disorders have an early onset, and all of these conditions persist throughout the lifespan. However, if these conditions are best explained using an integrated pattern, ASD are unique in terms of characteristic symptoms and the progression of these symptoms, which are the result of complex interactions between genetic, neurological, cognitive and environmental causes. Although some of the genes may be shared between the conditions on the autism spectrum and other developmental disorders, if they are best explained using an integrated pattern, they are a distinct set of conditions with primary causes and interactions between these causes that may be distinguished from the interactions and primary causes in the development of other developmental disorders.

7.3.2 False Beliefs, Myths and Stigmas Associated with ASD and Psychiatric Conditions

The final report to the Standing Senate Committee on Social Affairs, Science and Technology (2007) discussed above also highlighted the importance of public awareness of the causes and symptoms of ASD and awareness of the special needs of individuals with these conditions. The report (2007) states:

> "Throughout the course of the hearings on this difficult subject, witnesses identified a clear need for a national public awareness campaign. The Committee agrees that there is a general lack of understanding among Canadians about autism and its spectrum of disabilities and feels that a greater understanding of ASD by all Canadians could help to reduce the stress experienced by these individuals and their families. The general population should be made aware of the associated early signs and symptoms in order that parents might pursue assessment of their child at the youngest possible age (p. 22-23).

Several myths and misconceptions continue to misrepresent and exacerbate the difficulties individuals with ASD and their families face, and generate stigma and misunderstanding of these conditions in Canadian society.

Herbert,Sharp & Gaudiano (2002) argue that "several factors render autism especially vulnerable to etiological ideas and intervention approaches that make bold claims, yet are inconsistent with established scientific theories and unsupported by research (Herbert & Sharp, 2001) (citation in original, para 3)." These theorists argue that since the diagnosis of ASD can be emotionally and financially devastating for families of individuals with these

conditions, "[p]arents are typically highly motivated to attempt any promising treatment, rendering them vulnerable to promising 'cures.' The unremarkable physical appearance of autistic children may contribute to the proliferation of pseudoscientific treatments and theories of etiology...The normal appearance of autistic children may lead parents, caretakers, and teachers to become convinced that there must be a completely 'normal' or 'intact' child lurking inside the normal exterior (para 3)." Believing that ASD can be prevented or cured by dubious or untested methods (such as avoiding vaccination or special diets) can be detrimental to accessing the proper care and treatment that individuals with these conditions need.

Herbert et al (2002) discuss several of the 'myths' associated with the causes of autism spectrum disorders and how best to treat them. They (2002) argue that some of these 'theories' of ASD are pseudoscientific as best, and harmful to individuals with these conditions and their families at worst. For instance, although there is no evidence that poor parenting on the part of the mother causes ASD, the psychoanalytic hypothesis of 'refrigerator mothers' still remains in popular culture. As discussed in chapter 3, the 'refrigerator mother' hypothesis was popular during the era of psychoanalysis, but continued in the mental health literature and lay concepts of these conditions long after psychoanalysis fell out of favour. Analysts such as Bettelheim (1967) and Mahler (1952; 1958) argued that ASD were caused by frigid, emotionally unresponsive mothers, and the symptoms of these conditions were defense mechanisms against emotionally unavailable mothers. Bettelheim (1967) argued that a 'parentectomy' was necessary for the improvement of children with these conditions, advocating the removal of the child from the parent's care. Blaming the parents of children with ASD for their condition is unfair, unwarranted given the empirical evidence to the contrary, and harmful to the relationship between these parents and their children.

Another myth that can be detrimental to individuals with ASD, their families, and to accurate lay concepts of these conditions is the 'vaccine hypothesis.' This myth, although false, is pervasive and remains a popular theory of the causes of the conditions on the autism spectrum. The supposed evidence for this hypothesis comes from the increase in the prevalence of ASD over the last two decades or so, which as roughly coincided with the widespread use of the measles, mumps, rubella (MMR) vaccine starting in 1979 (Herbert et al, 2002). Also cited as evidence is the fact that the administering of the MMR vaccine coincides with the age when diagnoses of ASD are usually made in children. Finally, the MMR vaccine was identified in a now-discredited study as the cause of autism spectrum disorders. Originally published in the scientific journal *Lancet*, a study by Andrew Wakefield and his colleagues (1998) argued that they found cases of ASD in children after they had received the MMR vaccine. However, no medical evidence has ever been found

that supports a causal link between the MMR vaccine and ASD. Further, the evidence Wakefield et al (1998) presented has been greatly debated, and recently recanted by *The Lancet* in 2010. However, despite the lack of medical evidence, the vaccine hypothesis was reported and discussed in popular media, and was adopted by many people in the lay community.

The pervasiveness and popularity of the vaccine hypothesis has resulted in many parents refusing to vaccinate their children for fear of 'causing autism.' However, not vaccinating children causes significant administrative problems when the child enters school, since admission into preschool and elementary school requires that children be vaccinated. Not vaccinating children against harmful diseases like measles, mumps and rubella can also result in financial costs and significant loss of time for parents attempting to be exempted from the vaccination requirement, which are usually not granted on the grounds that vaccines are associated with the development of ASD. However, Herbert et al (2002) argue that "the real harm [from the vaccine hypothesis] is the public health concern raised by encouraging parents to avoid vaccinating their children from serious diseases that can easily be prevented (para 15)." Thus, the vaccine hypothesis is harmful to individuals with ASD, since false theories of the causes of these conditions will not reduce, and can exacerbate the stigma and misconceptions associated with these conditions. Further, this hypothesis is harmful to parents and families of individuals with these conditions, since believing that vaccination is the cause of ASD may incur guilt from parents or family members for the existence of these conditions in their loved ones. Finally, the vaccine hypothesis is harmful to society at large, both for perpetuating false theories of these conditions, and for potentially exposing the public to infectious diseases that would be prevented by vaccination.

However, ASD are not the only psychiatric conditions associated with myths, misconceptions and unwarranted stigmas. Torvino (2008) states that the National Mental Health Association, recently renamed Mental Health America, estimate that 71% of Americans still believe that mental disorders are caused by mental weakness, 65% of people believe they are the result of poor parenting, and 35% believe that mental disorders are a form of retribution for sinful behaviour. These statistics indicate a significant amount of the population believes that mental disorders are the result of moral failings or character flaws, and that and individuals who suffer from these conditions are blameworthy in some way. However, if psychiatric conditions have the same causal structure as conditions treated by other branches of medicine, the stigma attached to them may be reduced.

A better understanding of the causes of psychiatric disorders may help to eliminate the notion that these conditions mental disorders are the product of a weak will, moral failings, or poor upbringing. The belief that individuals with mental disorders are blameworthy for

their conditions may be partially based on the view that mental disorders do not have physical causes. However, if psychiatric conditions are best explained using an integrated pattern, they are the products of breakdowns in neurological mechanisms, which are the result of ongoing interactions between genes and the environment (see Nestler, 2009 for a discussion of epigenetic explanations of psychiatric conditions such as addiction and major depression). As I argued in chapter 1, if psychiatric conditions are best explained as the result of breakdowns in complex, biologically-based interactive mechanisms, they have the same causal structure of conditions such as cancer, type-II diabetes, obesity and heart disease, which are also the result of complex interactions between biological and environmental factors. If psychiatric conditions are best explained using an integrated explanation pattern, they are biologically-based conditions with complex causal interactions, like heart disease and cancer. Thus, an integrated explanation pattern may provide a better understanding of the causes of psychiatric conditions, and thus may help collapse the social distinctions between 'medical' and 'mental' disorders.

7.4 Conclusion

Changes in the classification and diagnosis of psychiatric conditions provide an opportunity to examine some longstanding epistemological issues in the philosophy of psychiatry. Several philosophers and clinicians have argued that classification and diagnosis may be more accurate if psychiatry adopts a causal framework based on theories from fields such as neuroscience, genetics, and epigenetics. This thesis used the disorders on the autism spectrum as a case study to investigate the relationship between patterns of explanation, diagnosis and classification in psychiatry. I examined the debate regarding whether psychiatric classification and diagnosis should continue to be based on symptoms, or whether psychiatry should adopt a cause-based framework. I discussed the conceptual and methodological problems with the current symptom-based framework identified by theorists who argue for a shift to a cause-based framework, and identify the classification and diagnostic issues with ASD as a result of a symptom-based framework. This thesis argued that the diagnosis and treatment of ASD could be improved by a shift to a cause-based framework.

However, there is much disagreement and debate regarding what sorts of causal explanations are best for psychiatric conditions, given the complexity and heterogeneity of causes in most psychiatric conditions. I identified and analyzed several obstacles to generating adequate casual explanations in psychiatry, and discuss arguments by theorists and clinicians regarding to how to address these obstacles. Through this analysis, I identified criteria that adequate explanation patterns in psychiatry may need to meet. I apply these

criteria to various explanation patterns for the disorders on the autism spectrum, based on the conceptual frameworks in which they were developed, and show that some explanation patterns can improve the reliability and validity of the diagnosis of these autism spectrum disorders if the criteria are met. However, this analysis also shows that inadequate explanation patterns for the causes of autism spectrum disorders limit the explanatory and predictive of the diagnosis of these conditions, which reduces the effectiveness of treatment. Thus, only some explanation patterns for psychiatric conditions and the conditions on the autism spectrum will be viable candidates for informing a cause-based diagnostic and classification system. I argued that an integrated explanation pattern may be the most powerful for explaining the causes and progression of ASD, since this pattern seems to meet the criteria I identified.

While this thesis mostly concentrated on the epistemological and ethical issues regarding a shift to a cause-based system, the types of explanation patterns that inform this system could influence metaphysical issues such as realism about psychiatric conditions. Debates regarding how best to explain, classify and diagnose psychiatric conditions has significant impact on the development and justification of realist theories of mental disorders. If the causes of psychiatric conditions are poorly understood, it is difficult to argue that these are real, biologically-based conditions and not social constructions (Murphy, 2006). However, the limited causal information we have about psychiatric conditions indicates that they are biologically-based conditions that are just as severe and harmful as so-called medical disorders. Thus, increasing understanding of the causes of psychiatric conditions, such as those on the autism spectrum, may help to justify realist accounts of these conditions. For instance, if psychiatric conditions are best explained using an integrated pattern, they have the same causal structure as medical conditions like heart disease and cancer. If psychiatric conditions are best explained using an integrated explanation pattern, they are just as 'real' as medical disorders.

These metaphysical and epistemological questions can inform ethical issues such as the type and extent of treatment resources for ASD that ought to be publicly funded, and how causal theories from fields such as epigenetics can further reduce misconceptions and stigmas associated with the disorders on the autism spectrum and psychiatric conditions in general. This chapter argued that a cause-based diagnostic framework may help address these ethical questions. I argued that while a cause-based framework may increase some costs of the diagnosis of these conditions, the overall costs of diagnosis and care of individuals with ASD will likely be reduced. Further, if the accuracy of diagnosis does improve with a cause-based system, the overall costs of treatment and care across the lifespan of individuals with ASD may be reduced. Next, I argued that a better understanding of the causes of ASD may provide support for legal cases petitioning for more government funding

for the treatment of ASD in Canada. Families of individuals with ASD often have to pay for the expenses of treatment and care themselves, which places a significant financial and emotional burden on the family members. Finally, I argued that a better understanding of the causes of ASD may help to further reduce the prevalence of myths and misconceptions about these conditions in society. Thus, adopting a cause-based diagnostic and classification framework may help to address ethical issues such as how to reduce the stigma and misinformation associated with ASD and other psychiatric conditions, and how best to allocate resources to treat these conditions.

References

Amir, R. E., Van den Veyver, I. B., Wan, M., Tran, C. Q., Francke, U & Zoghbi, H.Y. (1999). "Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics 23, (2)*: 185-88.

Arpaly, N. (2005). "How it's 'not just like diabetes:' Mental disorders and the moral psychologist. *Philosophical Issues, 15(1)*: 282-298.

Asperger, H. (1944). 'Autistic Psychopathy' in childhood. *In Autism and Asperger Syndrome.*. (1991) Trans. Firth, U. (Ed). Cambridge: Cambridge University Press.

———— (1979). "Problems of infantile autism" *Communication, 13*, 45-52.

Auton et. al v Attorney General, British Columbia, [2000] BCSC 1142

Auton (Guardian ad litem of) v. British Columbia (Attorney General), SCC 78, [2004] 3 S.C.R. 657

American Psychiatric Association (1952). *The Diagnostic and Statistical Manual, First Edition.* Washington: American Psychiatric Association

———————————— (1968). *The Diagnostic and Statistical Manual, Second Edition.* Washington: American Psychiatric Association

———————————— (1980). *The Diagnostic and Statistical Manual, Third Edition* (1980). Washington: American Psychiatric Association

———————————— (1987). *The Diagnostic and Statistical Manual, Third Edition, Revision.* Washington: American Psychiatric Association

———————————— (1994). *The Diagnostic and Statistical Manual, Fourth Edition, Text Revision.* Washington: American Psychiatric Association

———————————— (2013) *The Diagnostic and Statistical Manual, Fifth Edition.* Washington: American Psychiatric Association

Baker, D & Bhabha, F. (2006). *Health Law Review, 13, (1)*, 25-35.

Bailey, A., Le Couteur, A., Gottesan, I., Bolton, P., Simonoff, E., Yudza, F. Y., et al. (1995). "Autism as a strongly genetic disorder: Evidence from a British twin study." *Psychological Medicine, 25*: 63-77.

Bailey, A., Philips, W., & Rutter, M. (1996). "Autism: Towards an Integration of Clinical, Genetic, Neuropsychological, and Neurobiological Perspectives. *Journal of Child Psychiatry and Psychology, 37 (1)*, 89-126.

Baron-Cohen, S. (2000). "Is Asperger's syndrome necessarily a disability?" *Development and Psychopathology, 12*: 489-500.

Baron-Cohen, S, Leslie, A.M., & Frith, U, (1985) Does the autistic child have a "theory of mind?" *Cognition, 21*, 37-46.

Baron-Cohen, S. Tager-Flusberg H., & Cohen, D. (2000). *Understanding Other Minds: Perspectives from Developmental Cognitive Neuroscience, 2nd Edition.* Oxford: Oxford University Press.

Beaudet, A. L. & Zoghbi, H. Y. (2006). In *Understanding Autism: From Basic Neuroscience to Treatment.* Moldin, S. O. & Rubenstein, J. L. R. (Eds). Boca Raton: Taylor & Francis Group.

Bechtel, W. (2007). Reducing psychology while maintaining its autonomy via mechanistic explanation. In M. Schouten and H. Looren de Jong (Eds.). The Matter of the Mind: Philosophical Essays on Psychology, Neuroscience and Reduction. Oxford: Basil Blackwell.

Bechtel, W. & Richardson, (1993). *Discovering Complexity.* New Jersey: Princeton University Press.

Bechtel, W. & Abramson, A. (2005). A mechanist alternative. *Studies in History and Philosophy of the Biological and Biomedical Sciences , 36*, 421-441.

Beck, A. T. (1967). *The diagnosis and management of depression.* Philadelphia: University of Pennsylvania Press.

Beck, A. T. (1972). *Depression: Causes and Treatment.* Philadelphia: University of Pennsylvania Press.

Beck A.T., Rush A.J., Shaw B.F., Emery, G. (1979) emphCognitive Therapy of Depression, New York: Guilford Press

Belinger, L. J., & Smith, T. H. (2004). "A review of subtyping in autism and proposed dimensional classification model." *Journal of Autism and Developmental Disorders, 31 (4),* 411-422.

Bettelheim, B. (1967). *The Empty Fortress.* Toronto: Collier-MacMillan Ltd.

Birenbaum, A., Guyot, D., & Cohen, H. J. (1990). *Health Care Financing for Severe Developmental Disabilities*. Washington: American Association on Mental Retardation.

Bird, A. (2007). "Perceptions of epigenetics." *Nature, 447*, 396-398.

Bishop, D. V. M. (1989). "Autism, asperger's syndrome, and semantic-pragmatic disorder: Where are the boundaries?" *British Journal of Disorders of Communication, 24*, 107-121.

Bishop, D. V. M. & Norbury, C. F. (2002). "Exploring the borderlands of autistic disorder and specific language impairment: A study using standardized diagnostic instruments." *The Journal of Child Psychology and Psychiatry, 43 (7),*, 917-929.

Bleuler, E. (1916). *Lehrbuch der Psychiatrie*. Trans. A. A. Brill, (1951) *Textbook of Psychiatry*, New York: Dover.

Bonora, E., Lamb, J. A., Barnby, G., Bailey, A. J. & Monaco, A. P. (2006) "Genetic basis of autism." In *Understanding Autism: From Basic Neuroscience to Treatment*, Moldin, S. O. & Rubenstien, J. R. (Eds). Boca Raton: Taylor & Francis Group.

Boucher, J. (2009). *The autistic spectrum: Characteristics, causes and practical issues.*. Los Angeles: Sage.

Clark, L. A., Watson, D., & Reynolds, S. (1995). "Diagnosis and classification of psychopathology: Challenges to the current system and future directions." *Annual Review of Psychology, 46*, 121-53.

Crepaldi, L. & Riccio, A. (2009). Chromatin learns to behave. *Epigenetics, 4, (1)*, 23-26.

Elkisch, P. & Mahler, M. "On infantile precursors of the 'influencing machine.'" In *The Selected Papers of Margaret S. Mahler, Volumes One and Two.* (1979) M. S. Mahler, (Ed). New York: Jason Aronson.

Eysenck, H, J. (1985). Behaviourism in clinical psychiatry. *International Journal of Social Psychiatry, 31*, 163-169.

Ferster, C. B. (1961). "Positive reinforcement and behavioural deficits of autistic children." In *Classic Readings in Autism*, Donnellan, A. M (Ed), (1985). New York: Teachers College Press.

Ferster, C. B. & DeMyer, M. K. (1961). "The development of performances in autistic children in an automatically controlled environment." *Journal of Chronic Diseases, 13.* 312-345.

"Final Report on: The Enquiry on the Funding of the Treatment of Autism. Pay now or pay later: Autism Families in Crisis." (2007). The Honourable Art Eggleton, P.C. (chair) & The Honourable Wilbert Joseph Keon (deputy chair) *Final Report to Standing Senate Committee on Social Affairs, Science and Technology*, Canada.

Folstein, S. E. & Rutter, M. (1977a). "Genetic influences and infantile autism" *Nature, 256*: 726-728.

Folstein, S. E. & Rutter, M. (1977b). Infantile autism: A genetic study of 21 pairs." *Journal of Child Psychology and Psychiatry, 18*: 297-321.

Francis, A. First, M. B, & Pincus, H. A. (1995). *DSM-IV Guidebook.* Washington: American Psychiatric Association.

Frith, U. (1991). *Autism and Asperger Syndrome.* Cambridge: Cambridge University Press.

Gallese & Goldman (1998). Mirror neurons and the simulation theory of mind-reading. *Trends in Cognitive Sciences, 12*, 493-501.

Ganz, M. L. (2006). "The costs of autism." In *Understanding Autism: From Basic Neuroscience to Treatment*, Moldin, S. O. & Rubenstein, J. L. R. (Eds). Boca Raton: Taylor & Francis Group.

Goldberg, Allis, & Bernstein, (2007). Epigenetics: A landscape takes place. *Cell, 128*, 635-638.

Graham, G. & Stephens, G. L. (1994). *Philosophical Psychopathology.* Cambridge: MIT Press.

Harris, J. C. (1995). *Developmental Neuropsychiatry.* New York: Oxford University Press.

Herbert, J. D., Sharp, I. R, & Gaudiano, B. R. (2002). "Separating fact from fiction in the etiology and treatment of autism." *The Scientific Review of Mental Health Practice, 1, (1)*, 23-43.

Holliday, R. (1987). "The inheritance of acquired epigenetic variations." *Science, 238*, 163-170.

———— (2006). "Epigenetics: A historical overview." *Epigenetics, 1, (2)*, 76-80.

Hildebrand, D. C. (1999). "Cost-benefit analysis of Lovaas treatment of autism and autism spectrum disorders (ASD). Vancouver: Columbia Pacific Consulting.

Illes, J. & Bird, S. J. (2006). Neuroethics: A modern context for ethics in neuroscience. *Trends in Neuroscience, 30* (10), 1-7.

Insel, T. R. & Collins, F. S. (2003). "Psychiatry in the genomics era." *American Journal of Psychiatry, 160 (4)*, 616-620.

Jablesky & Kendel, (2002). "Criteria for assessing a classification in psychiatry." In *Psychiatric Diagnosis and Classification*, Maj, M., Gaebel, W., Lopez-Ibor, J. J., & Sartorious, N (Eds). New York: John Wiley & Sons, Ltd.

Jacobson, J. W., Mulick, J. A., & Green, G. (1998). "Cost-benefit estimates for early intensive behavioral intervention for young children with autism -general model and single state case. *Behavioral Interventions, 13*, 201-226.

Jaenisch, R. & Bird, A. (2003). "Epigenetic regulation of gene expression: how the genome integrates intrinsic and environmental signs." *Nature Genetics Suppliment, 33*, 245-254.

Jiang, Y., Sahoo, T., Michaelis, R.C., Bercovich, D., Bressler, J., Kashork, C. D., Lui, Q., Shaffer, L. G., Schroer, R. J., Stockton, D. W., Spielman, R. S., Stevenson, R. E., & Beaudet, A. L. (2004). "A mixed epigenetic/genetic model for oligenic inheritance of autism with a limited role for UBE3A." *American Journal of Medical Genetics, 13*, 1-10.

Junaid, M. A., Kowal, D., Barua, M., Pullarkat, P. S., Brooks, S. S. & Pullkarkat, R, K. (2004). "Proteomic studies identified a single nucleotide polymorphism in glyoxalase I as autism susceptibility factor." *American Journal of Medical Genetics, 131*, 1-17.

Kandel, E. R. (1998). "A new intellectual framework for psychiatry." *American Journal of Psychiatry, 155, (4),* 457-469.

————- (1999). "Biology and the future of psychoanalysis: A new intellectual framework for psychiatry revisited." *American Journal of Psychiatry, 156, (4)*, 505-524.

Kanner, L. (1943). "Autistic disturbances of affective contact." In *Classic Readings in Autism*. Donnellan, A. M. (Ed). (1985). New York: Teachers College Press.

———— (1949). Problems of nosology and psychodynamics in early infantile autism. *American Journal of Orthopsychiatry, 19,* 416-426.

Kelly, G. A. (1955). *The psychology of personal constructs*. New York: Norton. Reprinted by Routledge (London), 1991.

Kendell, R. E. (2002). "Five criteria for an improved taxonomy of mental disorders." In *Defining Psychopathology in the 21st century*, Helzer, J. E., & Hudziak, J. J. (Eds). Washington: American Psychiatric Association Publishing.

Kendler, K. S. & Parnas, J. (2008). *Philosophical Issues in Psychiatry: Explanation, Phenomenology and Nosology*. Baltimore: John Hopkins University Press.

Kendler, K. S., Gardner, C. O. & Prescott, C. A. (2006). "Toward a comprehensive developmental model of major depression." Richmond: Virginia Institute for Psychiatry and Behavioral Genetics and the Department of Psychiatry, Medical College of Virginia of Virginia Commonwealth University.

Kendler, K. S. (2008). "Explanatory Models for Psychiatric Illness." *American Journal of Psychiatry, 165 (6)*, 695-702.

Kendler, K. S. (2009). "An historical framework for psychiatric nosology." *Psychological Medicine, 39*, 1935-1941.

Kennedy, (2004). "Why we must end insurance discrimination against mental health care." *Harvard Journal on Legislation, 41, (2)* 363-375.

Kupfer, D. J., First, M. B., & Regier, D. A. (2002). *A research agenda for DSM-V.* Washington: American Psychiatric Assocation.

Leahy, R. (1996). *Cognitive-Behavioral Therapy: Basic Principles and Applications.* New York: Jason Aronson.

Leichtman, M. (1990). "Developmental psychology and psychoanalysis: I. The context for a revolution in psychoanalysis." *Journal of the American Psychoanalytic Association, 38*: 915-950.

Levinson, B. M. & Osterweil, L. (1984). *Autism: Myth or Reality?.* Springfield: Charles C. Thomas.

Levy, N. (2007). *Neuroethics.* Cambridge: Cambridge University Press.

Lovaas, O. I. (1987). Behavioral treatment and normal educational and intellectual functioning in young autistic children. *Journal of Consulting and Clinical Psychology, 55*, 3-9.

————- (1993). The development of a treatment-research project for developmentally disabled and autistic children. *The Journal of Applied Behavior Analysis, 26, (4)*, 617-630.

Lovaas, O. I., Schreibman, L. & Koegel, R. L. (1974). "A behavior modification approach to the treatment of autistic children." *Journal of Autism and Childhood Schizophrenia, 4*, 111-129.

Machamer, P., Darden, L, & Craver, C. (2000). Thinking about mechanisms. *Philosophy of Science, 64*, 1-25.

Mahler, M. (1952.) "On child psychosis and schizophrenia: Autistic and symbiotic infantile psychosis." In *The Selected Papers of Margaret S. Mahler, Volumes One and Two.* (1979) M. S. Mahler, (Ed). New York: Jason Aronson.

———— (1958). "Autism and symbiosis: Two Extreme Disturbances in Identity." In *The Selected Papers of Margaret S. Mahler, Volumes One and Two* M. S. Mahler, 1979.

Mandlawitz, M. R. (2005). "Educating children with autism: Current legal issues. In *Handbook of autism and pervasive developmental disorders, Volume One*. Volkmar, F. R., Paul, R., Klin, A. & Cohen, D. (Eds). New Jersey: John Wiley & Sons.

Marshall, B. J. & Warren, J. R. (1984). "Unidentified curved bacilli in the stomach of patients with gastritis and peptic ulceration." *Lancet, 1*: 1311-1315.

Martin, J. B. (2002). "The integration of neurology, psychiatry, and neuroscience in the 21st century." *Americal Journal of Psychiatry, 159 (5)*, 695-704.

Mayes & Horwitz (2005). "DSM-III and the revolution in the classification of mental illness. *Journal of the History of Behavioural Sciences, 41 (3)*: 249-267.

Merikangas, K., R. (2002). " Implications of genetic epidemiology for classification." In *Defining Psychopathology in the 21st Century* Helzer, J. E., Hudziak, J. J. (Eds). Washington: American Psychiatric Association.

Mesibov, G. B., Adams, L. W., & Klinger, L. G. (1997). Autism: Understanding the Disorder. New York: Plenum Press

———— (1959). "On infantile precursors of the "influencing machine." In *The Selected Papers of Margaret S. Mahler, Volumes One and Two* M. S. Mahler, 1979.

Mitchell, S. D. (2008a). "Explaining complex behaviour." *Philosophical issues in psychiatry*. Kendler, K. S. & Parnas, J. (Eds). Baltimore: The Johns Hopkins University Press.

————. (2008b) "Taming causal complexity." *Philosophical issues in psychiatry*. Kendler, K. S. & Parnas, J. (Eds). Baltimore: The Johns Hopkins University Press.

Moffitt, T. E., Caspi, A. & Rutter, M. (2005). "Strategy for investigating interactions between measured genes and measured interactions. *Arch Gen Psychiatry, 62*, 473-481.

Molden, S. O. & Rubenstein, J. L. R. (2006). *Understanding Autism: From Basic Neuroscience to Treatment.* Boca Raton: Taylor & Francis Group.

Motiwala, S.S., Gupta, S., Lilly, M. B., Ungar, W. J., & Coyte, P.C. (2006). "The Cost-Effectiveness of Expanding Intensive Behavioural Intervention to All Autistic Children in Ontario." *Healthcare Policy, 1 (2),* 135-151

Muhle, R., Trentacoste, S. V., & Rapin, I. (2004). "The genetics of autism." *Pediatrics, 113*, 472-486.

Murphy, D. (2006). *Psychiatry in the scientific image* Cambridge: MIT Press.

———— (2008). "Levels of explanation in psychiatry." *Philosophical issues in psychiatry.* Kendler, K. S. & Parnas, J. (Eds). Baltimore: The Johns Hopkins University Press

Nadesan, M. H. (2005). *Constructing Autism: Unraveling the 'Truth' and Understanding the Social.* Abingdon: Routledge

Nakao, M. (2001). "Epigenetics: Interaction of DNA methylation and chromatin." *Gene, 278*, 25-31.

Narayanan, U. & Warren, S. T. (2006). "Neurobiology of related disorders: Fragile X syndrome" In *Understanding Autism: From Basic Neuroscience to Treatment.* Molden, S. O. & Rubenstein, J. L. R. Boca Raton: Taylor & Francis Group.

Nestler, E. (2009). "Epigenetic mechanisms in psychiatry." *Biological Psychiatry, 65, (4),* 189-190.

Norbury, C. F. & Bishop, D. V. M. (2010). "Inferential processing and story recall in children with communication problems: A comparison of specific language impairment, pragmatic language impairment and high-functioning autism." *International Journal of Language and Communication Disorders, 37 (3),* 227-251.

Paluszny, M. (1979). *Autism: A practical guide for parents and professionals.* New York: Syracuse University Press.

Pennington, K. Cotter, D. & Dunn, M. J. (2005). "The role of proteomics in investigating psychiatric disorders." *British Journal of Psychiatry, 187*, 4-6.

Persico, A. M. & Bourgeron, T. (2006). "Searching for ways out of the autism maze: Genetic, epigenetic and environmental cues." *Trends in Neuroscience, 29 (7),* 349-358.

Poland, J., Von Eckhardt, B., & Spaulding, W. (1994). "Problems with the DSM approach to classifying psychopathology." In emphPhilosophical Psychopathology. Graham, G, & Stephens, G. L. (Eds). Cambridge: MIT Press.

Reimer, M. (2008). Psychopathology without (the language of) disorder. Neuroethics. Doi 10.1007/s1152-008-9017-5.

Riesman, J. A. (1991). *A History of Clinical Psychology, 2nd Edition.* New York: Hemisphere Publishing Corporation

Rimland, B. (1964). *Infantile Autism.* United States: Meredith Publishing Company

Ritchie, & Richards, (2002). In *Psychiatry as a Neuroscience.* Lopez-Ibor, J. J., Gaebel, W., Maj, M., & Sartorious, N. (Eds). Chichester: John Wiley & Sons, Ltd.

Rizzolatti, G. & Arbib (1998). Language within our grasp. *Trends in Neuroscience, 21*, 188-194.

Rizzolatti, G. & Craighero, L. (2004). "The mirror neuron system." *Annual Review of Neuroscience, 27*, 169-192

Rodenhiser, D. & Mann, M. (2006). "Epigenetics and human disease: Translating basic biology into clinical applications." *174 (3)*: 341-348.

Rogers, S. J. & Williams, J. H. G. (2006). *Imitation and the Social Mind.* New York: Guildford Press.

Rogers, S. J. & Dawson, G. (2009). Play and Engagement in Early Autism: The Early Start Denver Model. Volume 1: The Treatment. New York: Guilford Press.

Rutter, M. (1968). "Concepts of autism: A review of research." In Donnellan, A. M. (1985). (Ed). *Classic Readings in Autism.* New York: Teachers College Press.

——— (1978). "Diagnosis and definition of childhood autism." *Journal of Autism and Developmental Disorders, 8, (2)*: 139-161.

Rutter, M. (2005). "Genetic influences and autism" In *Handbook of autism and pervasive developmental disorders, Volume One.* Volkmar, F. R., Paul, R., Klin, A. & Cohen, D. (Eds). New Jersey: John Wiley & Sons.

Rutter, M., & Schopler, E. (1978). *Autism, A Reappraisal of Concepts and Treatment.* New York: Plenum Press.

Samaco, R. C., Hogart, A., &LaSalle, J. M. (2005). "Epigenetic overlap in autism-spectrum neurodevelopmental disorders: MECP2 deficiency causes reduced expression of UBE3A and GABRB3." *Human Molecular Genetcs, 14 (4),*: 483-492.

Schaffner, K. F. (2008). "Etiological models in psychiatry: Reductive and non-reductive approaches." *Philosophical issues in psychiatry.*

Kendler, K. S. & Parnas, J. (Eds). Baltimore: The Johns Hopkins University Press

Schanen, N. C. (2006). "Epigenetics of autism spectrum disorders." *Human Molecular Genetics, 15, 2*: 138-150.

Seifert, C. D. (1990). *Theories of Autism.* New York: University Press of America.

Shorter, E. (1993). *A History of Psychiatry.* Toronto: John Wiley & Sons Inc.

Sirgiovanni, E. (2009). "The mechanistic approach to psychiatric classification." *Dialogues in Philosophy, Mental and Neurosciences, 2(2)*: 45-49

Steffenburg, S., Gillberg, C., Hellgren, L., Anderson, L., Gillberg, I. C.,Jakobsson, G. et al, (1989). "A twin study of autism in Denmark, Finland, Iceland, Norway and Sweden" *Journal of Child Psychology and Psychiatry, 30* 405-416.

Tager-Flushberg, H. (2007). "Evaluating the theory of mind hypothesis." *Current Directions in Psychological Science, 16 (6)*, 311-315.

Thagard, P. (1999). *How Scientists Explain Disease*. New Jersey: Princeton University Press.

———— (2008). "Mental illness from the perspective of theoretical neuroscience." *Perspectives in Biology and Medicine, 51*, 335-352.

Thagard, P. & Findlay, S. (2010). "Conceptual change in medicine: Explanations of mental illness from demons to epigenetics." In W. J. Gonzalez (Ed.), Conceptual revolutions: From cognitive science to medicine. A Corua: Netbiblo.

Tiedemann, M. (2008). "Health Care at the Supreme Court of Canada I: *Auton (Guardian ad litem of) v British Columbia (Attorney General)*". Ottawa: Parliamentary Information and Research Service

Tidmarsh, L. & Volkmar, F. R. (2003). "Diagnosis and epidemiology of autism spectrum disorders." *Canadian Journal of Psychiatry, 48 (8),* 517-525.

Torvino, S. A. (2008). The impact of neuroscience on health law. *Neuroethics, 1 (1)*, 101-117.

Turner, A. J. (2008). Are disorders sufficient for reduced responsibility? *Neuroethics, 1 (1),* doi 10.1007/s12152-009-9041-0.

Van Acker, R., Loncola, J. A., & Van Acker, E. Y. (2005). "Rett Syndrome: A Pervasive Developmental Disorder." In *Handbook of Autism and Pervasive Developmental Disorders, Volume One*. Volkmar, F. R., Paul, R., Klin, A. & Cohen, D. (Eds). New Jersey: John Wiley & Sons.

Veenstra-VanderWeele, J. & Cook, E. H. (2004). "Molecular genetics of autism spectrum disorder." *Molecular Psychiatry, 9*, 819-832.

Vincent, N. A. (2008). Responsibility, dysfunction and capacity. *Neuroethics, 1 (1)*, 199-204.

Volkmar, F. R., Paul, R., Klin, A. & Cohen, D. (Eds). (2005a). *Handbook of Autism and Pervasive Developmental disorders, Volume One*. New Jersey: John Wiley & Sons.

———————————————— (2005b). *Handbook of Autism and Pervasive Developmental disorders, Volume Two*. New Jersey: John Wiley & Sons.

Volkmar, & Klin, (2005). "Issues in the classification of autism and related conditions." In *Handbook of Autism and Pervasive Developmental Disorders. Volume I.* Volkmar, F. R., Paul, R., Klin. A, Cohen, D. (Eds). Hoboken: John Wiley & Sons, Inc.

Volkmar, F. R. State, M. & Klin, A. (2009). "Autism and autism spectrum disorders: Diagnostic issues for the coming decade." *Journal of Child Psychology and Psychiatry, 50 (1)*: 108-115.

Waddington, C. H. (1942). "The epigenotype." Endeavour 1, 18-20.

Wakefield, A. J., Murch, S. H., Anthony, A., Linnell, J., Casson, D. M., Malik, M., Berelowitz, M., Dhillon, A. P. Thomson, M. A., Harvey, P., Valentine, A., Davies, S. E., & Walker-Smith, J. A. (1998). Ileal-lymphoid-nodular hyperplasia, non-specific colitis, and pervasive developmental disorder in children. *The Lancet, 351* 637-641.

Wakefield, J. C. (1992). "Disorder as harmful dysfunction: A conceptual critique of DSM-III-R's definition of mental disorder." *Psychological Review, 99 (2)*, 232-247.

————————- (1997). Diagnosing DSM-IV, part 1: DSM-IV and the concept of disorder. *Behaviour Research and Therapy, 35*, 633-649.

———————— (1999a). "Philosophy of science and the progressiveness of the DSM's theory-neutral nosology: Response to Follette and Houts, part 1. *Behavior Research and Therapy, 37*, 963-999.

———————— (1999b). "Philosophy of science and the progressiveness of the DSM's theory-neutral nosology: Response to Follette and Houts, part 2. *Behavior Research and Therapy, 37*,1001-1027

————————- (2007). "The concept of a mental disorder: Diagnostic implications of the harmful dysfunction." *Analysis,*

Wakefield, J. C. & First, M. B. (2003). "Clarifying the distinction between disorder and non- disorder: Confronting the overdiagnosis (false positives) problem in DSM-V." In Advancing DSM: Dilemmas in Psychiatric Diagnosis. Phillips, K. A., First, M. B., & Pincus, H. A. (Eds). Washington: American Psychiatric Association Publishing.

Warren, J. R. & Marshall, B. J. (1983). "Unidentified curved bacilli on gastric epithelium in active chronic gastritis. *Lancet, 1,*: 1273-1275.

Watson, J. B. (1916). "Behavior and the concept of mental disease." *Journal of Philosophy, Psychology, and Scientific Methods, 13*, 589-597.

Weaver, I. C. G. (2007). "Epigenetic programming by maternal behavior and pharmacological intervention." *Epigenetics, 2 (1)*, 22-28.

Widiger, T. A, & Clark, L. A. (2000). "Toward DSM- V and the classification of psychopathology. *Psychological Bulletin, 126 (6),* 946-963.

Williams, J. G. H., Whiten, A., Suddendorf , T & Perrett, D. I. (2001) "Imitation, mirror neurons and autism." *Neuroscience and Biobehavioral Reviews, 25, (4)*, 287 -295.

Willick, M. S. (1990). "Psychoanalytic concepts of the etiology of mental illness." *Journal of the American Psychoanalytic Association, 38 (4)*: 1049-1081.

Wing, L. (1976). *Early Childhood Autism: Clinical, Educational and Social Aspects, 2nd Edition.* Oxford: Pergamon Press

———- (1991). "The relationship between Asperger's syndrome and Kanner's syndrome." In *Autism and Asperger Syndrome*, U. Frith (Ed). Cambridge: Cambridge University Press.

———- (1997). "The history of ideas on autism: Legends, myths and reality." *Autism, 1, (13)*, 13-23

——— (2005). "Problems with categorical classification systems." In Volkmar, F. R., Paul, R., Klin, A. & Cohen, D. *Handbook of Autism and Pervasive Developmental Disorders* (Eds). New Jersey: John Wiley & Sons.

Woodward, J. F. (2008). "Cause and explanation in psychiatry: An interventionist perspective." *Philosophical issues in psychiatry.*, Kendler, K. S. & Parnas, J. (Eds). Baltimore: The Johns Hopkins University Press

Wright, C. & Bechtel, W. (1997). Mechanisms and psychological explanation. In P. Thagard (ed.), *Philosophy of Psychology and Cognitive Science* (Volume 4 of the Handbook of the Philosophy of Science). New York: Elsevier. *Wynberg v. Ontario*, [2006] CanLII 22919

Zilbovicius, M., Meresse, I., Brunelle, F., Samson, Y., & Boddaert, N. (2006). Autism, the superior temporal sulcus and social perception. *Trends in Neuroscience, 29 (7),* 359-366