University of Alberta

Family Experience of Differential Diagnosis when a Child has a Developmental Disability

by

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Abstract

This paper-based dissertation is an investigation of the parental experience of the diagnostic process when a child has a developmental disability. The first paper is a literature review on differential diagnosis and is followed by two research papers. The primary research question of the study was "What is the meaning of a differential diagnosis to parents of a child with a developmental disability?" A basic interpretive approach (Merriam, 2002) was employed to provide an in-depth look at this experience. Parents of children with identified diagnoses participated in the study as well as parents who are still seeking a differential diagnosis. Fourteen parents were interviewed, including six parents of children with identified genetic conditions, three parents of children with autism spectrum disorders, and five parents of children with unidentified disabilities. The first research paper addresses parents' encounters with medical professionals while seeking a diagnosis for their child. Parents reported tolerating intensive testing, sensing rigidity in thinking by professionals, perceiving pessimistic prognostic information, receiving multiple diagnoses, feeling dissatisfaction with the information provided, and encountering a range of professionals. The second research paper addresses the meaning parents ascribe to a differential diagnosis. Parents described searching for a diagnosis because of the importance of "knowing," understanding the cause for the disability, gaining knowledge about future expectations and appropriate interventions, and gaining access to funding or specific services. The dissertation concludes by bringing the current study together with the Family Adjustment and Adaptation Response (FAAR) model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) and provides recommendations for improving this process for families.

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CHAPTER 1: INTRODUCTION

"The most important thing that happens when a child with disabilities is born is that a child is born. The most important thing that happens when a couple becomes parents of a child with disabilities is that a couple becomes parents" (Ferguson & Asch, 1989, p. 108). This may seem like a self-evident statement, but when we look at the history of professional responses to the birth of a child with a disability, we find patterns of research and practice that have, until recently, assumed that the disability itself inevitably overwhelms all other considerations (Ferguson, 2002).

Further, most existing studies of family adaptation to disability have not specified the type of developmental disability, but family responses may vary based on the child's specific type of developmental disability (Hodapp, Fidler, & Smith, 1998). Breakthroughs in human genetics have led to increased numbers of researchers who are examining individuals with specific genetic diagnoses. Particular conditions predispose individuals to characteristic developmental patterns, specific behaviours, as well as strengths and weaknesses (Dykens & Hodapp, 2001). In addition, different labels may also indirectly influence parents, peers, and other surrounding individuals.

This study is an investigation of the parental experience of the diagnostic process. The present study extends the literature on family adaptation to diagnosis of a disability by using an interpretivist perspective (Merriam, 2002) to examine the ways in which families adapt to their child's differential diagnosis or lack thereof. I interviewed parents of children with a specific developmental disability, as well as parents of children who are still looking for a differential diagnosis. Following the interviews, I conducted a thematic analysis (Merrian, 1998).

Purpose

The purpose of this paper-based dissertation was to explore parental experience of the diagnostic process and the resultant impact on family adaptation. The primary research question was "What is the meaning of a differential diagnosis to parents of a child with a developmental disability?" Further, the study asked how parents experience the process of their child's differential diagnosis or lack of diagnosis. This process includes genetic counselling, psychoeducational assessment, dealing with professionals, and the feelings associated with receiving or failing to receive a differential diagnosis.

Rationale

Most research on family adaptation to disability has centered on families of children with mixed or non-specific disabilities, although family outcomes may differ based on the child's specific type of disability (Hodapp et al., 1998). There is a long history of family and disability research, but until recently, few studies have addressed the specific diagnoses of the children and have simply considered "developmental disability" as being the most significant factor (Ferguson, 2002) contributing to family stress. Most research has glossed over the situational complexities and has failed to look at specific stressors or strains, such as the diagnostic process or the specific type of developmental disability. Given that certain conditions predispose individuals to particular adaptive and maladaptive behaviours, intellectual or linguistic strengths and weaknesses, and characteristic developmental patterns (Dykens & Hodapp, 2001), an exploration of family responses to receiving specific diagnoses seems relevant. Individual diagnoses are also associated with different diagnostic processes and this experience also deserves attention. Providing a differential diagnosis has the potential to afford many

benefits to parents including appropriate intervention for caregivers and the individual; knowledge regarding the individual's learning challenges and strengths; and knowledge regarding medical or mental health risks and resiliencies (Carmichael, Pembrey, Turner, & Barnicoat, 1999; Gilman, Heyman, & Swain, 2000; Griffiths & Watson, 2004; Poelmann, Clements, Abbeduto, & Farasad, 2005).

Although differential diagnosis can be very valuable to parents and the individual, many individuals with developmental disabilities do not have a specific differential diagnosis (American Association on Mental Retardation, 2002). Differential diagnosis is often sought by parents in the belief that a specific label will enable support to the family or provide a greater understanding of the needs of the child. Nevertheless, little is known about what this process is like for families or how a failure to receive a differential diagnosis is perceived.

The current study applies an interpretivist perspective to study parental experience of diagnosis. Most research on family adaptation has used a positivist perspective, which does not provide a sufficient description of the family experience. Furthermore, although there has been a lot of research on parents and having a child with a disability, few studies have investigated variations in parental reactions as a function of differences in the child's disability (Poelmann et al., 2005) or in response to the diagnostic process specifically.

It is important to highlight the emphasis on meaning in current family response theory and how this applies to interpretive, constructionist research. An interpretivist perspective is uniquely suited to this undertaking because it seeks to describe, interpret, and understand another's perspective (Merriam, 2002). Moreover, it recognizes that

reality is a socially constructed phenomenon that can be shared through an individual's narrative accounts of experiences. This approach allows individuals to tell their story in their own words and to explain the meanings associated with their experience. The emphasis on meanings is reflected in current family theory on reaction to stressful events, specifically the Double ABCX model (McCubbin & Patterson, 1983, 1987) and the Family Adjustment and Adaptation Response model (FAAR; Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998).

Individuals in interaction with their world socially construct meaning. Experiences are not the fixed, measurable phenomenon that is assumed in positivist, quantitative research (Merriam, 2002). There are variations in meaning and interpretation of events, depending on the unique situations of individuals, and experiences change over time. Such variation is again reflected in current family theory. Consequently, it is only by asking parents about the diagnostic process that we can begin to understand the impact that this has on their lives. For these reasons, basic interpretive qualitative research (Merriam, 2002) has been chosen as the methodology.

Basic interpretive research exemplifies all of the essential characteristics of qualitative research in general (Merriam, 2002). In this approach, the researcher is interested in understanding how participants make sense of a situation and this meaning is mediated through the researcher. The strategy is inductive, aiming to gain understanding and develop concepts, rather than testing a theory as in deductive, positivist research (Merriam, 2002). Further, the outcome of basic interpretive qualitative research is richly descriptive, allowing the researcher to gain an understanding of the experience from the participants' perspectives.

Reflexivity

In interpretivist research, the researcher is the research instrument (Merriam, 1995; Patton, 1990). Denzin (1997) suggests that researchers must frame the research and reflexivity is a process by which writers articulate "what frames our seeing" (Lather, 1993, p. 675). Reflexivity is an ongoing process that saturates every stage of the research process (Guillemin & Gillam, 2004). To increase the credibility of the present study, information on my experiences, training, and perspective will be revealed, including personal and professional information that may affect data collection, analysis, or interpretation.

Personal and professional experiences. I have been working in the field of developmental disability for approximately ten years. I supported individuals with disabilities and their families at a community agency for five years, providing counselling and sexuality education training. In this role, I also conducted research on sexuality issues, abuse prevention, and human rights promotion. Another significant portion of my responsibilities was organizing an annual conference dealing with sexuality and rights issues.

Until recently, I was a board member at a local community agency providing supports to adults with developmental disabilities, strongly advocating community involvement of individuals with disabilities. We were also very involved with families and this organization values the contribution parents and extended family can make to the lives of the individuals to whom they provide support.

My education has also given me lots of opportunities to work with individuals with disabilities and to receive specific guidance on issues related to this population. In

my Master's degree, my thesis topic was "Sexuality Education for Individuals with Developmental Disabilities: The Need for Assessment." During my thesis research, I conducted several interviews and assessments of the sexuality knowledge of adults with developmental disabilities.

I have taken many courses on disabilities, both at the undergraduate and graduate level. In addition, I had the fantastic opportunity to receive training in mental health issues for individuals with disabilities through the Certificate Programme in Habilitative Mental Health, offered through NADD, Brock University, and Niagara University. It is through this program that I became interested in family research as well as diagnosis. In the same summer, I took two courses; one course on family-centred practice and another on genetic syndromes. After taking these courses, I started to think about the distinctive characteristics of individuals with specific genetic syndromes and how these might uniquely affect families. I hypothesized that the experience would be different for families who had a child with Fragile X syndrome, for example, than for parents of children with Angelman syndrome. Learning about specific disabilities and how they are formally diagnosed also allowed me to think about and clarify my own beliefs regarding diagnosis and disability. These will be discussed below.

Personal perspectives. From my work with individuals with developmental disabilities, it is quite evident that I have a strong disability advocacy perspective, recognizing the rights of individuals with disabilities to express themselves and have the same opportunities as individuals without disabilities. My experience with genetics courses also initiated a lot of thought about diagnosis and what I would do if I had a child with a disability. I began to ponder about the benefits and drawbacks to diagnosis, and

became deeply interested in the history of eugenics, the initial rationale behind genetic testing, and how the disability community views the diagnostic process. This resulted in a publication with my mentor, Dorothy Griffiths (Griffiths & Watson, 2004) about the benefits of genetic testing, yet recognizing some of the risks involved. After discussing these risks further, a second publication developed (Watson & Griffiths, in press) about the right to life and the ethical issues around prenatal testing and other medical interventions with individuals with disabilities. In the course of writing these chapters, I realized that I am very much in favour of prenatal testing as a means to prepare families for the experience of having a child with a disability and to have greater access to information about the specific manifestations of the disability, but I do not support testing as a means of preventing disability. I realize this is a slippery slope and have had to negotiate many of my thoughts regarding this controversial issue.

I also have strong beliefs about the roles of families in the lives of individuals with disabilities. I acknowledge the professional prejudice that has existed against parents of children with disabilities. Furthermore, I appreciate the resilience shown by most families as they adapt to having a child with a disability. I also attest to a systems perspective, realizing that families are not alone in the rearing of their children. The larger community, professionals, and extended family significantly affect families as they adapt to the raising of their child. My recognition of this is evident in my application of the FAAR model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998), which will be discussed throughout this dissertation.

How my experience and perspectives may influence this study. It is significant to mention that when I began this study, I was not yet a parent, nor had I ever gone through

the diagnostic process. My perspective on parenting a child with a disability comes from my knowledge of the literature and my experience working with families at community agencies. I had to be open to listening to families as they described their experience because *they* were the experts on their experiences. Throughout the process, I had to be cautious about allowing my beliefs regarding family systems and how parents *should* react to disability to interfere with my observations and analysis.

I became pregnant with my first child during the process of my interviews with families and this was a very unique experience. I had hopes and dreams for my own child and tried to monitor those feelings throughout the process. At times I struggled to keep my feelings neutral when parents were discussing emotional issues and I did share a few tears with some parents. I feel that my pregnancy made parents feel at ease and put us at the same level. I was not a researcher coming in to examine them, but rather a fellow parent chatting about a challenging time in their lives.

Throughout the interviews, I had to be very open to families' reactions to having a child with a disability and tried not to become disturbed if families reported wishing to terminate pregnancies or wishing that their child had not been born. I had to recognize that these were normal family reactions and did not allow my own beliefs about eugenics to interfere with the study. By keeping an open mind and letting families tell their story, I tried to remain as receptive as possible during both the collection and analysis of the interview data.

Conclusion and Overview of Subsequent Chapters

This study presents an in-depth exploration of the parental experience of the diagnostic process. Basic interpretive qualitative inquiry (Merriam, 2002) was chosen as

the methodology in an attempt to provide a rich description of this experience. This approach was also chosen in reaction to the preponderance of positivist studies that may have failed to reflect the true experience of families. Differential diagnosis is often sought by parents in the belief that a specific label will enable support to the family or provide a greater understanding of the needs of the child with a developmental disability. Nonetheless, little is known about how families experience this process or how they perceive failing to receive a differential diagnosis. It is hoped that this dissertation and the three papers that follow will shed light on these issues to gain a greater understanding of families and their experiences when a child has a developmental disability.

Paper 1 is a literature review, presenting the debates around differential diagnosis and some of the issues faced by families and medical professionals throughout this process. This review highlights the need for interpretive research and critiques the limited research that has looked at family experience of differential diagnosis. Paper 2 is titled, "Parental Experiences Dealing with Professionals while Seeking a Differential Diagnosis" and addresses parents' interactions with medical practitioners in the search for a label for their child. Finally, Paper 3, entitled "Why do Parents Seek a Differential Diagnosis? A Search for Meaning and Support," looks at the meaning of a differential diagnosis and why parents seek such a label. The dissertation concludes with a brief summary of the studies conducted and brings the research together with the FAAR (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) model.

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Differential Diagnosis of Developmental Disability

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Abstract

This literature review presents the issues around families and differential diagnosis of developmental disability. The review begins with a brief overview of family research, followed by the arguments for and against labelling. The limited information known about the diagnostic process from family and medical professionals is discussed. Finally, a critique of the literature highlights the need for more interpretive studies of the diagnostic process and the reasons for seeking a differential diagnosis.

Differential Diagnosis of Developmental Disability

Labelling theory is concerned with how the self-identity and behaviour of an individual is created and influenced by how that person is categorized and described by others in society (Becker, 1963). As an application of phenomenology and originating in sociology and criminology, the theory focuses on the linguistic tendency of the dominant culture to negatively label minorities or those seen as deviant from norms. Labelling is associated with the concepts of a self-fulfilling prophecy, stigmatization, and stereotyping.

Scheff (1966) was the first to apply this theory to individuals with mental health issues, claiming that mental illness is manifested as a result of societal influence. Society views certain actions as deviant and in order to come to terms with and understand these behaviours, places a label on those who exhibit them. Consequently, Scheff argues that expectations are placed on such persons and over time, these individuals unconsciously change their behaviours to fulfill them.

Applied to developmental disability, differential diagnosis and the subsequent labelling that occurs is an issue that causes great debate in the literature (Gillman, Heyman, & Swain, 2000; Lauchlan & Boyle, 2007; McDermott, Goldman, & Varènne, 2006). Some theorists postulate that labels are not helpful (e.g., Lauchlan & Boyle; McDermott et al.), while others believe that differential diagnosis is essential for tailoring supports to the individuals with a developmental disability and their families (e.g., Dykens & Hodapp, 2001; Griffiths & Watson, 2004). Many families who have a child with a developmental disability seek a differential diagnosis in the belief that a label will result in treatment, intervention, and social support, ultimately leading to an improved

quality of life for the family and the individual (Gillman et al.). For the purposes of this literature review, the term *differential diagnosis* will refer to a specific diagnosis provided by a medical practitioner; examples might be Angelman syndrome or Fragile X syndrome, contrasted with a nonspecific diagnosis such as pervasive developmental disorder (PDD) or global developmental delay.

The following literature review begins with a brief overview of family research and then examines the debates about differential diagnosis. A discussion of the limited information regarding the diagnostic process from parent and professional perspectives follows. The review concludes with a critique of the current literature, highlighting the need for interpretivist research on family adaptation to diagnoses.

Family Research

Historically, there has been a general assumption that the stress of having a child with developmental disabilities has a "deleterious effect on parental functioning" (Crnic, Friedrich, & Greenberg, 1983, p. 127) and "a retarded child is unexpected, unpleasant, and a source of stress for the family" (Fotheringham, Skelton, & Hoddinott, 1972, p. 283). First-generation research on the impact of child disability and mental retardation on families (usually mothers) presented a bleak picture of stress, burden, depression, social isolation, and psychological dysfunction (Shapiro, 1983). However, as research became more refined, it was apparent that disability *per se* was not necessarily a predictor of parental dysfunction. More accurately, a host of variables appeared to influence the relationships between disability and adaptation or maladjustment of families. This line of investigation began to apply complex social, ecological, and stress-appraisal-coping models to the study of responses to disability in an effort to understand the interaction

between the presence of disability and the development of family dysfunction (e.g., McCubbin & Patterson, 1987; Patterson, 1989). In general, these models have moved away from solely deficit-based interpretations of adjustment and have recognized the possibility of positive adaptations to having a child with a developmental disability. Further, they have begun to emphasize the interactional and developmental nature of adaptations and are addressing a variety of intrapersonal factors, such as appraisal or evaluation of a situation, and external factors, such as resources and social support (Shapiro, Blacher, & Lopez, 1998).

Although we are seeing a shift in the emphasis of family research in reaction to disability, there are still methodological limitations to many of the recent studies, such as reliance on questionnaires and other positivist measures, which will be discussed later in this review. Most existing studies of family adaptation have focused on families of children with a variety of disabilities or the differential diagnosis is not mentioned, yet family stress and coping may be altered based on the child's specific type of disability (Hodapp, Fidler, & Smith, 1998). Furthermore, little is known about the effects on the family of nonspecific developmental delays or disabilities of unknown etiology (Keogh, Garnier, Bernheimer, & Gallimore, 2000). Approximately 30 – 40% of individuals with developmental disabilities have no clear etiology for their disabilities (American Association on Mental Retardation, 2002) and thus many families are never given a specific label for the disability of their child.

Researchers are increasingly examining specific genetic diagnoses, largely because particular conditions may predispose individuals to unique developmental patterns, behaviours, strengths, or weaknesses (Dykens & Hodapp, 2001). Specific

differences in development or health patterns exist whether or not a differential diagnosis is made, but the differential diagnosis may have another set of effects all its own. Accordingly, the differential diagnosis may influence families, peers, and other surrounding individuals. These benefits and disadvantages of differential diagnosis are discussed below.

The Debate Regarding the Utility of Differential Diagnosis

There are differing opinions about the role of labels and the function of differential diagnosis. Lauchlan and Boyle (2007) have succinctly highlighted several of the arguments for and against differential diagnosis and subsequent labelling. These lines of reasoning include access to treatment and resources, raised awareness, reducing ambiguities, providing comfort to families, and creating a sense of identity. Each of these arguments will be highlighted in turn.

Access to treatment and resources. The first and most significant reason in favour of differential diagnosis is that diagnosis, or labelling, leads to treatment and opens doors for resources (Dykens & Hodapp, 2001; Dykens, Hodapp, & Finucane, 2000; Griffiths & Watson, 2004; Hodapp et al., 1998). Differential diagnosis can provide access to specific interventions and community services and is often a requirement for funding dollars (Gillman et al., 2000; Leonard, 1999). A differential diagnosis can also allow practitioners to figure out what to look for in individuals to determine their unique needs and thus tailor supports. Dykens and Hodapp (1997), for example, recommend that individuals with Fragile X syndrome require emphasis on contextual learning and visual integration rather than focusing on auditory short-term memory and individuals with

Williams syndrome require tools such as computers and calculators that do not rely on the individual's written ability. Such specific strategies definitely have educational utility.

Clinically, it is important to know about genetic diagnosis because the underlying genetic mechanisms are related to phenotypic differences among individuals with different etiologies for a genetic syndrome or any other condition (Summers & Pittman, 2004) and many disorders are associated with specific medical conditions that can affect quality of life. Approximately 80% of individuals with Williams syndrome, for example, have congenital cardiovascular anomalies (Finucane, 2004). It is essential to recognize that these benefits are apparent, whether the differential diagnosis is of genetic origin or not. Differential diagnoses provide information and allow families to make more informed decisions about their child's health and supports.

Lauchlan and Boyle (2007) offer the counter argument that a label is often applied, but that there is a lack of consideration regarding the nature of intervention. They affirm that it is merely a case of "label equals more money" or "label equals placement at special school" (p. 36), without consideration about how the extra resources are helping the children's difficulties. A label can be useful if it leads to a specific intervention that facilitates learning, as discussed above, but labels themselves do not always provide the precise details regarding "what you do about it" (Ogilvy, 1994, p. 60). I would postulate that such is the case with newer rare diagnoses, where a label is given of Angelman syndrome, for example, because little is known about how best to teach a child with such a disability, and so the differential diagnosis does not lead to specialized intervention. However, with increased awareness regarding disability identification, researchers and practitioners may be able to address this need.

Raised awareness. The second case for differential diagnosis is that labels lead to increased knowledge and can promote understanding of particular difficulties (Lauchlan & Boyle, 2007). Diagnosis of a specific disability may lead to increased familial or teacher understanding of the child's behavioural uniqueness (Dykens & Hodapp, 2001; Griffiths & Watson, 2004; Gus, 2000). Gus describes a classroom where students were allowed to openly discuss autism and the challenges of having a peer with such a diverse learning need. This open discussion lead to an increased acceptance of the peer with autism and improved the classroom environment.

However, some theorists argue that labels can contribute to exclusion from society and social disadvantage (Gillman et al., 2000; Sutcliffe & Simons, 1993). Thus, labels lead to stigmatization. With specific reference to families, when we look at the history of professional responses to the birth of a child with a disability, we find patterns of research and practice that have, until recently, assumed that the disability itself inevitably overwhelms all other considerations (Ferguson, 2002). The notion persists that a family with a child who has a developmental disability is a disabled family.

Reducing ambiguities. A third line of reasoning for differential diagnosis is that labels lower uncertainties regarding behavioural challenges and uniqueness and provide clear communication devices for professional exchanges of information (Griffiths & Watson, 2004; Dykens & Hodapp, 2001). Research and practice has often treated individuals with developmental disabilities as homogeneous, without regard for individual differences.

On the other hand, Lauchlan and Boyle (2007) argue that there is no clear agreement amongst professionals about how diagnoses are decided, and labels can lead to

generalization of children's difficulties. Consequently, a label can lead to a neglect of specific individual issues. Furthermore, there is debate about whether professionals, when using labels, are talking about the same thing. One medical professional may be referring to something different when discussing "developmental disability," for example. This also speaks to cultural differences, where the United Kingdom uses the term "learning disability" to refer to what Canadians mean by an "intellectual disability." In Canada, a "learning disability" is a completely different diagnosis, usually referring to an average intelligence level and below average performance (Learning Disabilities Association of Canada, 2005). McDermott et al. (2006) also discuss the cultural context of labels and how the meanings for labels have changed throughout the years.

Providing comfort to families. A fourth claim in support of differential diagnosis is that labels offer reassurance to children and families by 'explaining' their difficulties (Poelmann, Clements, Abbeduto, & Farasad, 2005). This advantage will be discussed later in the review, but Lauchlan and Boyle (2007) argue against this line of reasoning, maintaining that differential diagnosis leads to a focus on within-child deficits and possibly lowered expectations. As Brechin (1999) so firmly stated, "If the whole problem, *by definition*, lies *with* the individual [due to the diagnosis], then our understandings and interventions start and stop with the individual" (p. 1). If one can blame the differential diagnosis for the behaviours of the individual, then we can use that as an excuse for not being able to do anything to ameliorate the challenging behaviour (Lauchlan & Boyle, 2007). Griffiths and Watson (2004) argue, however, that differential diagnosis allows not only identification of the challenges presented by the disability, but also the strengths and advantages associated with the diagnosis. Individuals with

Williams syndrome, for example, typically have social and musical strengths (Finucane, 2004) and individuals with Prader-Willi often excel at jigsaw puzzles (Dykens & Kasari, 1997), assets that can be useful when tailoring interventions and supports.

Creating a sense of identity. A final argument for the use of labels is that they can provide people with a sense of identity or a sense of belonging to a group. Differential diagnosis of children can give families access to many parent professional groups, which can provide emotional support to families and a sense of not being alone when dealing with their child's behavioural uniqueness (Griffiths & Watson, 2004; Simon, 2004). However, Lauchlan and Boyle (2007) counter that labels can contribute to teasing, low self-esteem, and bullying. These theorists conclude that labels are "more unhelpful than helpful" (p. 41).

There is thus disagreement in the literature about the value of labels. I would argue that in many cases differential diagnosis is helpful, but it can also do more harm than good. Regardless of the debates for and against differential diagnosis, there is a strong emphasis on disability identification in the literature and in practice. Many families who have a child with a developmental disability embark on a quest for a label in the conviction that this differential diagnosis will facilitate intervention and social support (Gillman et al., 2000). These family and professional issues around diagnosis are discussed below.

Family Diagnosis Research

A limited body of research has looked at families and their reasons for seeking a differential diagnosis. Families have reported perceived benefits of receiving a differential diagnosis, including knowing a cause for the disability (Burden, 1999;

Gillman et al., 2000); access to appropriate intervention (Carmichael et al., 1999; Gillman et al.; Poelmann et al., 2005); contact and support from other families (Carmichael et al., 1999; Wilcox, 1991); and help explaining the child's behaviour to their friends (Carmichael et al., 1999; Gillman et al.). It is vital to recognize, however, that seeking a differential diagnosis is rarely a positive experience. Several researchers describe parents' negative perceptions of dealing with professionals while seeking a diagnosis for their children (Brogan & Knussen, 2003; Goin-Kochel, Mackintosh, & Myers, 2006; Leonard, 1999; Poelmann et al.; Skotko, 2005; Watson, manuscript in preparation; Woolfe & Bartlett, 1996). Parents appear to be more satisfied with the diagnostic process if they are given written information about the disability at the time of diagnosis (Brogan & Knussen), see fewer professionals throughout the diagnostic process (Goin-Kochel et al.), and if professionals accept parents' first suspicions of disability (Brogan & Knussen).

A differential diagnosis can relieve the stress or ambiguity of the unknown (Gillman et al., 2000) and Trute (2005) further declares that the greatest stressor for parents is "not knowing", contributing to the fear and anxiety experienced by parents. Even when the formal information is "unpalatable" (p.12), Leonard (1999) found that parents prefer "knowing" to the frustration and anxiety associated with the uncertainty of not knowing. Not knowing may take several forms; including not knowing how or why the child has a disability, not knowing what to expect in the future, or not knowing if there is something useful that would help the child. Being informed also plays a part in parents' sense of control, which has been found to contribute to parents' positive adaptation to having a child with a disability (Knox, Parmenter, Atkinson, & Yazbeck, 2000). When parents do not know what is unique about their child, they feel a lack of

control over the situation because they do not have the pertinent information to help their child (Knox et al.). Poelmann et al. (2005) cite one mother's reaction to receiving a diagnosis of Fragile X for her child:

> When the testing was done, I felt like this major mission had been completed. I was just relieved - relieved because I knew what it was. Knowing what it was wasn't going to change it, but at least you could figure out a course of action. At least you could get some information to know what to expect and what you are dealing with, and maybe figure out what some better ways of handling it would be. (p. 262)

Burden (1991) proposes that one of the reasons parents may experience guilt in reaction to having a child with a disability is because they do not have an understandable cause for the disability. When the etiology of the disability can be satisfactorily explained and a label is received, the guilty feeling may be reduced, but as long as the etiology "remains shrouded in mystery, the likelihood of self-blame is greater" (Burden, p. 333). Burden studied three groups of mothers in London, England, including mothers who were recipients of service after the differential diagnosis of the child's disability, mothers whose children had been similarly diagnosed but did not have professional support until the child was 2 years of age, and a third group of mothers whose children's disabilities had not been formally diagnosed. Mothers of children without a differential diagnosis had higher levels of family crisis and had more difficulty working through their feelings of anger and guilt than mothers in the other two groups. It is important to note that none of the diagnoses for the children were due to maternal drinking or other parental factors so the diagnosis relieved guilt. Gillman et al. (2000) considered a hierarchy of disabilities,

discussing how some labels are more stigmatizing than others. For example, these authors postulate that a hearing or visual impairment is often perceived as less stigmatizing than a learning disability. Families may search for an alternative label that is regarded by the person or society as less disgraceful. It is also critical to recognize that some labels might be more stigmatizing to the child and some labels, such as Fetal Alcohol Spectrum Disorder, may be more stigmatizing to the parent. An important area of research would address such feelings in parents of children where the disability was due to a parental treatment factor or the disability was preventable.

Another significant reason for seeking a differential diagnosis is to gain information about medical vulnerabilities and treatment. In their questionnaire study of parents in London, England, Woolfe and Bartlett (1996) found that parents expressed frustration and anxiety in not receiving specific medical information about their child. Leonard (1999) found similar results in a study of 240 parents in London, England, and cites one parent's frustration at not receiving any differential diagnosis for her child:

> My only wish was that I had been told earlier in her life, as the doctor always made an excuse whenever I asked questions and never explained what was wrong with her apart from telling me she had brain damage which was left to my imagination. (p. 3)

This parent's narrative highlights the issue of timing of the diagnosis and the frustration that families can feel in failing to receive a label for the disability. The diagnostic process, which often occurs over a long period of time and consists of many visits to many professionals, is often traumatizing to families and the family system.

Factors cited by parents as contributing to a negative perception of the diagnostic process are dealing with too many doctors (Goin-Kochel et al., 2006; Wilcox, 1991; Woolfe & Bartlett, 1996); medical professionals who acted with pity or embarrassment at the birth of the child with a disability (Leonard, 1999; Skotko, 2004; Woolfe & Bartlett); doctors not listening to parents' concerns (Baird, McConachie, & Scrutton, 2000; Harrington, Patrick, Edwards, & Brand, 2006; Leonard; Woolfe & Bartlett); negative information regarding disabilities (Hedov, Wikblad, & Annerén, 2002; Poelmann et al., 2005; Skotko); and the lack of information regarding disabilities and interventions (Hedov et al.; Skotko; Sloper & Turner, 1993).

Timing of diagnosis is a specific factor that has been identified as contributing to family functioning in reaction to diagnosis. When there is a delay in finding a differential diagnosis, parents report frustration at the loss of valuable time early in the child's life, a time when they need to feel they are doing everything possible for their child (Baird et al., 2000; Leonard, 1999; Poelmann et al., 2005; Woolfe & Bartlett, 1996). Some researchers have found that parents are more in favour of being given information at an early stage, even if this was based on suspicions rather than certainties (Leonard; Sloper & Turner, 1991; Woolfe & Bartlett). In their study of mothers of children with Fragile X syndrome, Down syndrome, and autism, Abbeduto et al. (2004) discuss the timing at which these disabilities are diagnosed and how parents of children with Down syndrome can begin the process of adaptation to their child's condition when their children are only a few hours old, and in many cases, before the child is born. They conclude that the experience is different for parents of children with Fragile X and autism since these disorders are often not recognized until later in the child's life.

When looking at diagnosis of pervasive developmental disorders such as autism, most children are not diagnosed until about 5 years of age (Brogan & Knussen, 2003; Goin-Kochel et al., 2006) and the average time between first referral and diagnosis for autism spectrum disorders is approximately 9 months (Harrington et al., 2006). Some autism spectrum disorders are also diagnosed with more delay. Asperger syndrome, for example, is typically diagnosed later than autism or Pervasive Developmental Delay - Not Otherwise Specified (PDD-NOS; Goin-Kochel et al.). In their interpretive study of 21 mothers of children with Fragile X syndrome and Down syndrome, Poelmann et al. (2005) found that the average time between the beginning of the diagnostic process and receiving a diagnosis for Fragile X syndrome ranged from 2 to 11 years (mean = 6.1 years), while mothers of children with Down syndrome were informed of the diagnosis at the child's birth or the following day. Furthermore, the mothers of children with Fragile X syndrome experienced more distress than the mothers of children with Down syndrome, largely due to misdiagnoses and the increased uncertainty associated with Fragile X syndrome. Delays in receiving a differential diagnosis have lead parents to doubt the quality of the medical care their child was receiving (Harrington et al.; Woolfe & Bartlett, 1996). Consequently, many families direct frustration toward medical practitioners.

Medical Professional Issues

Some researchers have postulated that the delays in receiving a diagnosis are due to the lack of information regarding autism spectrum disorders (Goin-Kochel et al., 2006) and newer genetic diagnoses (Lee et al., 2005). Most medical professionals learn about developmental disabilities on a case-by-case basis through their experiences with

individual children (Lee et al.) and have little formalized education regarding differential diagnoses. There is a lack of professional awareness and diagnostic expertise about autism spectrum disorders (Brogan, 2001; Shah, 2001) and confusion regarding diagnostic criteria for some of the autism subtypes (Wing, 1999). Researchers have also cited lack of awareness regarding genetic syndromes. In their study of teacher and pediatrician awareness regarding Down, Fragile X, and velo-cardio-facial (VCFS) syndromes, Lee et al. found that significantly much more was known about Down and Fragile X syndromes than VCFS, a rarer genetic disorder. For pediatricians, the number of years of medical experience only increased their knowledge of the physical features of Down syndrome, but did not affect their knowledge level regarding the other two disabilities. Lee et al. concluded that professionals make critical decisions regarding the treatment of individuals with these disorders and would benefit from professional development.

The more a disability can be located in the body rather than the mind (e.g., a genetic syndrome versus an autism spectrum disorder), the more confident a medical professional will be in offering a differential diagnosis (Gillman et al., 2000). Autism spectrum disorders are diagnosed based on a child's behaviour (Lord & Rissi, 2000), contributing to an uncertainty about the correctness of the differential diagnosis. However, genetic diagnoses are not without ambiguity. Angelman syndrome, for example, is a diagnosis that can be genetically confirmed in 80% of cases, but 20% of cases are diagnosed clinically and there are several mimicking conditions for this disorder, including Rett syndrome, PDD, and childhood autism (Williams, Lossie, Driscoll, & the R.C. Phillips Unit, 2001). Behavioural difficulties are common

developmental concerns for several differential diagnoses as well as normal concerns for families of children between the ages of 1 and 3, factors that lead medical professionals to be cautious about incorrect labelling or diagnosis (Filipek et al., 2000). However, the potential harm of an incorrect diagnosis must be weighed against the frustration of a delayed diagnosis (Goin-Kochel et al., 2006).

A few researchers have looked at medical professionals' diagnostic practices. Hasnat and Graves (2000) found that pediatricians scored relatively low on an index based on recommended disclosure practices. Pediatricians in this study reported lack of time and lack of experience or training as the most significant hindering factors in their ability to provide strong diagnostic support to families. Sices, Feudtner, McLaughlin, Drotar, and Williams (2004) looked at physicians' referral practices when a developmental disability was suspected. Using fictionalized clinical vignettes, these researchers found that the expression of parental concern did not increase the probability of referral for diagnostic services or intervention programs and many physicians preferred to "watch and wait" (p. 279).

Pediatricians have reported a preference for formalized medical school education as a means of learning about differential diagnoses (Lee et al., 2005). However, Ralston (2000) talks about the training he received in medical school about disabilities and how when disability was discussed, it was a bad thing "to be avoided at all costs" (p. 335). His education was geared toward describing medical vulnerabilities of specific differential diagnoses and that the value of the individuals' lives was "hardly recognized, must less stressed" (p. 335). Moreover, Ralston states that medical professionals have very little

contact with individuals with developmental disabilities. As a result, they are unaware of the strengths of these individuals.

Methodological Considerations and Concluding Remarks

Despite the growth of qualitative techniques in family research over the last decade, the need continues for more extended narrative accounts from parents and other family members that capture the full range of their experience (Ferguson, 2002). Until recently, most research on families of children with disabilities tended to gloss over the situational complexities and cultural variables that surround all of us, in the interest of making global claims about the inevitable and often negative responses of parents to having a child with a disability (Ferguson, 2002).

Of the literature cited in this review addressing families and the diagnostic process, only a few papers employed a qualitative approach (e.g., Gillman et al., 2000; Leonard, 1999; Poelmann et al., 2005; Woolfe & Bartlett, 1996). Given that labelling is inherently a phenomenological issue, dealing with what we call things and how we construct our attitudes and behaviour, more qualitative research is needed to look at this issue from a disability perspective. Further, of the qualitative studies that have been conducted, little information was provided regarding the specific methodology or the data analysis procedures. Only Poelmann et al. described their thematic analysis procedures, employing a grounded theory approach for analysis, but not for the entire study. Some studies included an open-ended component to their questionnaires (e.g., Baird et al., 2000; Skotko, 2004; Sloper & Turner, 1993), but most of the research focused on parental satisfaction with the diagnostic process (e.g., Brogan & Knussen, 2003; Goin-

Kochel et al., 2006; Hedov et al., 2002; Skotko; Sloper & Turner), employing Likert-type questionnaires.

There is a call for research questions that help elicit the true experiences of families of children with developmental disabilities. We know relatively little about the process of facilitating family involvement and less still about how improved relationships between family members of individuals with disabilities and professionals might affect either individual well-being or family functioning (Gersten, Irvin, & Keating, 2002). We also do not understand the impact of the diagnostic process on families and how receiving a differential diagnosis or failing to receive a differential diagnosis impacts family adaptation (Keogh et al., 2000). There is a need for direct accounts from parents and other family members. There is the tendency to obtain information on a single occasion, with few attempts to conduct longitudinal studies or to genuinely get to know families and understand their experiences. In fact, some researchers have conducted their studies without even meeting parents, relying on mailed surveys (e.g., Hedov et al., 2002; Skotko, 2004) or web-based designs (e.g., Goin-Kochel et al., 2006).

Collaboration between parents and professionals in research has also been recommended (Turnbull, Friesen, & Ramirez, 1998; Turnbull & Turnbull, 1997). Turnbull et al. advocate for participatory action research when conducting family research and Singer (2002) encourages the use of focus groups and structured interviews to provide additional information. When researchers begin to treat parents as valued and contributing members of the research team, parents may view professionals as support (Lipsky, 1985). Such collaboration will encourage the family system to maintain as much adaptation as possible with as little stress as possible.

Collaboration between parents and professionals is also required during the diagnostic process. Sices et al. (2005) found that parental distress did not increase the likelihood of pediatrician referral for diagnostic services, but if medical professionals listened to parental concerns, then this may address some of the constraints limiting their diagnostic abilities such as lack of time and lack of information regarding disabilities (Hasnat & Graves, 2000). Such an approach would also address a significant parental difficulty, which is that doctors do not listen to their thoughts or worries during the diagnostic process (Baird et al., 2000; Harrington et al., 2006; Leonard, 1999; Woolfe & Bartlett, 1996). Parental concerns have been found to reveal much about their children's developmental and behavioural needs, are easy to elicit, and take only a few minutes of professional time (Glascoe, 1999). Asking for parental feedback also takes less time than a formal screening instrument (Glascoe; Sices et al., 2004), and facilitates a collaborative, family-focused approach to addressing differential diagnoses.

Medical practitioners have expressed a desire for more professional development opportunities regarding disabilities (Hasnat & Graves, 2000; Lee et al., 2005; Ralston, 2000). If medical professionals' only education regarding disabilities focuses on the negatives, then that is going to be the message translated to families when providing diagnostic and prognostic information. Medical practitioners require more than updated medical and scientific literature on differential diagnoses; they also require information on the educational and social potentials of individuals with disabilities (Skotko, 2004).

The above literature review has provided information on the debates regarding the value and concerns of differential diagnosis and has provided an overview of the limited research on families' experiences of the diagnostic process. This review also addressed

medical professionals' concerns regarding differential diagnosis and provided a brief look at diagnostic practices. The review concluded with a methodological critique of the diagnostic literature, highlighting the need for more interpretive, qualitative studies to elucidate what the diagnostic process is truly like for families.

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Parents' Experiences Dealing with Professionals when Seeking a Differential Diagnosis

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Abstract

This is a study of the diagnostic process when a child has a developmental disability. Fourteen parents were interviewed about their experiences searching for a differential diagnosis for their child. Participants included parents of children with identified disabilities as well as parents who were still seeking a differential diagnosis. Parents reported tolerating intensive testing, sensing rigidity in thinking by professionals, perceiving pessimistic prognostic information, receiving multiple diagnoses, feeling dissatisfaction with the information provided, and encountering a range of professionals. Recommendations are provided for making this process better for families.

Parents' Experiences Dealing with Professionals when Seeking a Differential Diagnosis

"We are the lucky ones. We now have a diagnosis..." (Wilcox, 1991, p. 94). These are the final words of Wilcox's (1991) article on her family's long search for a diagnosis for their daughter, Heather. This mother of a child with Angelman syndrome discusses their experiences dealing with doctors and the 5 year process that ensued to finally receive a diagnosis of Angelman syndrome for Heather. She also recounts the number of labels given to Heather, ranging from "developmentally delayed due to unknown causes" (p. 92) to cerebral palsy.

It is striking that Wilcox (1991) states that her family is lucky to have received a diagnosis of Angelman syndrome for Heather. Much of the literature on parenting a child with a disability is negative and developmental disability has been described as "probably the most dreadful diagnosis a parent can receive" (Fewell, 1986, p. 209). Perhaps it is the receipt of a vague or nonspecific diagnosis that contributes to the negative familial reaction. Many families who have a child with a developmental disability embark on a major quest for a differential diagnosis, seeking this more specific diagnosis in the belief that identification and classification of a set of symptoms will result in treatment, intervention, and social support that will ultimately lead to an improved quality of life for the family and the individual (Gillman, Heyman, & Swain, 2000). Importantly however, The American Association on Mental Retardation (2002) affirms that 30 – 40% of individuals with developmental disabilities have no clear etiology for their disabilities. Consequently, many families search for a differential diagnosis for prolonged periods and some are never given a specific label for the disabilities of their children.

There are several sources of advice available to medical professionals on how to present parents with an early diagnosis of disability in their child (e.g., Cottrell & Summers, 1990; Cunningham, 1994), but parents of children with various disabilities continue to report dissatisfaction with the diagnostic process. There is a substantial body of literature on parental dissatisfaction with the diagnostic process (Baird, McConachie, & Scrutton, 2000; Brogan & Knussen, 2003; Goin-Kochel, Mackintosh, & Myers, 2006; Pianta, Marvin, Britner, & Borowitz, 1996; Skotko, 2005; Sloper & Turner, 1993; Woolfe & Bartlett, 1996), but not on what families face as they live through this experience. Research shows that parents are unhappy with this process, but little is known about what contributes to the unpleasant experience. Moreover, research has been conducted in Sweden (Hedov, Wikblad, & Anneren, 2002), the United Kingdom (Baird et al.; Brogan & Knussen; Gillman et al., 2000; Leonard, 1999), and the United States (Goin-Kochel at al.; Poelmann, Clements, Abbeduto, & Farasad, 2005), but a Canadian perspective is lacking in the literature. Given that medical systems and policies differ internationally, a look at the Canadian experience is pertinent.

This study investigates the family experience of the diagnostic process and specifically focuses on parents' experiences dealing with professionals throughout that process. The present study is part of a larger study addressing the family experience of differential diagnosis when a child has a developmental disability and the meaning that such a diagnosis provides. The study extends the literature on families and disability diagnosis by using an interpretivist perspective (Merriam, 2002) to examine the ways in which families experience dealing with professionals in seeking such a differential diagnosis. The term *differential diagnosis* is employed to refer to a diagnosis of a specific

condition such as Angelman syndrome, Down syndrome, or Fragile X syndrome, as contrasted with a general or nonspecific diagnosis such as pervasive developmental disorder (PDD) or global developmental delay. I interviewed both families of children who had received differential diagnoses and families of children seeking differential diagnoses. Following the interviews, I conducted a thematic analysis (Merriam, 1998).

Literature Review

With advancements in human genetics and diagnostic techniques, there has been increased emphasis on determination of increasingly specific differential diagnoses. In addition, access to many services or funding dollars is based on the individual's specific disability. Thus, seeking a differential diagnosis has important implications for service, as well as financial support to families.

Given that certain conditions predispose individuals to specific adaptive and maladaptive behaviours, intellectual or linguistic strengths and weaknesses, and characteristic developmental patterns (Dykens & Hodapp, 2001), an exploration of family responses to receiving different diagnoses is relevant. Providing a differential diagnosis potentially affords many benefits to families including appropriate intervention for caregivers and the individual; knowledge regarding the individual's learning challenges and strengths; and knowledge regarding medical or mental health risks and resiliencies (Carmichael, Pembrey, Turner, & Barnicoat, 1999; Griffiths & Watson, 2004). Although differential diagnosis can be very valuable to families and the individual, many individuals with disabilities do not have a specific diagnosis (AAMR, 2002). Differential diagnosis is often sought after by families in the hopes that a specific label will provide support to the family or offer a greater understanding of the needs of the child.

It is vital to recognize, however, that seeking a differential diagnosis is rarely a positive experience. Several researchers have described the negative perceptions of parents in dealing with professionals while seeking a diagnosis for their children (Baird et al., 2000; Brogan & Knussen, 2003; Goin-Kochel et al., 2006; Leonard, 1999; Pianta et al., 1996; Poelmann et al., 2005; Skotko, 2005; Woolfe & Bartlett, 1996). The diagnostic process, which often occurs over a long period of time and consists of many visits to many professionals, is often traumatizing to families and the family system.

Uncertainty is a major contributor to stress for many families and some researchers believe that "not knowing" can be *the* major stressor (Trute, 2005). Not knowing may take several forms; this could include not knowing how or why the child has a disability, not knowing what to expect in the future, or not knowing if there is something useful that would help the child. Not knowing is also a concern for parents of children with diagnoses such as autism or pervasive developmental disorder (PDD) because these diagnoses are based on behavioural observations (Lord & Rissi, 2000), leaving parents wondering whether the diagnosis is "correct." The following literature review will address the issue of receiving a differential diagnosis, which includes dealing with a large number of professionals, the delays in receiving a diagnosis, and the lack of positive, up-to-date, or useful information about the diagnosis.

Number of Professionals

Research has shown that parents are more satisfied with the diagnostic process when they have seen fewer professionals and when the children received the diagnosis at a younger age (Goin-Kochel et al., 2006). However, parents report a large number of visits to doctors, psychologists, hospitals, and therapists to try to understand their child's

problems (Baird et al., 2000; Schall, 2000; Woolfe & Bartlett, 1996). The large numbers of visits often result because physicians and other medical professionals do not respond to parents' initial concerns about their child's development. It is important to note that the specific diagnosis might dictate the number of doctor visits. Down syndrome, for example, is easily recognized and typically diagnosed right at birth or even prenatally, resulting in families having fewer interactions with medical practitioners. Angelman or Fragile X syndrome, however, may not be as obvious until later in the child's life, and diagnosis is not as easy, leading to a lengthier process.

Parents are frequently faced with a slow and frustrating period of uncertainty and worry, and find themselves in the position of trying to convince their children's doctors that there is a need for a specialist assessment (Harrington, Patrick, Edwards, & Brand, 2006; Goin-Kochel et al., 2006; Woolfe & Bartlett, 1996). Moreover, parents report that they are often met with cold indifference and a lack of respect when dealing with professionals, a pattern that is repeated throughout the life of the child (Schall, 2000; Woolfe & Bartlett). In a vignette study where physicians were presented with case studies of children with various delays, the expression of parental concern did not increase the probability of referral to diagnostic or treatment services (Sices, Feudtner, McLaughlin, Drotar, & Williams, 2004). This tendency is unfortunate since it has been found that parents may be more able to withstand the lengthy process of assessment if they feel that their opinions, observations, and fears are taken seriously by professionals (Brogan & Knussen, 2003). Parental concern about a child's developmental status is also a strong predictor of an actual developmental delay (Glascoe, 1999; Sices et al.).

Many parents go from doctor to doctor in the hopes of receiving a differential diagnosis. However, more doctor visits, especially to new doctors, mean more hassle and a greater financial burden and increased numbers of doctor visits also contribute to a delay in receiving specific diagnostic information (Goin-Kochel et al., 2006).

Delay in Diagnosis

Timing of diagnosis is a specific factor that has been identified as contributing to family functioning in reaction to diagnosis. When there is a delay in finding a differential diagnosis, parents report frustration at the loss of valuable time early in the child's life, a time when they need to feel they are doing everything possible for their child (Leonard, 1999; Poelmann, 2005).

The timing at which different disabilities are identified has also been discussed in the literature (Abbeduto et al., 2004; Woolfe & Bartlett, 1996) and may contribute to parents' adaptation or difficulty with the diagnosis. Parents of children with Down syndrome, for example, can typically begin the process of adaptation to their child's condition when their children are only a few hours old and in many cases, before the child is even born. The experience is different for parents of children with cerebral palsy, Fragile X, and autism since these disorders are often not recognized until later in the child's life (Abbeduto et al.; Woolfe & Bartlett). When looking at diagnosis of pervasive developmental disorders such as autism, most children are not diagnosed until about 5 years of age (Brogan & Knussen, 2003; Goin-Kochel et al., 2006). The average delay in receiving a diagnosis for Fragile X syndrome is 6.1 years (Poelmann et al., 2005), while the average time between first referral and diagnosis for autism spectrum disorders is approximately 9 months (Harrington, Patrick, Edwards, & Brand, 2006), which is

significantly better than in the past (Howlin & Moore, 1997), where delays were 1 to 5 years. There are also differences within autism spectrum disorders with regard to timeliness of diagnosis, with Asperger syndrome typically being diagnosed later than autism or Pervasive Developmental Delay- Not Otherwise Specified (PDD-NOS; Goin-Kochel et al). Such concerns point to the difference between receiving diagnoses of specific physiological disorders versus less specific disorders such as pervasive developmental disorders. The latter are diagnosed exclusively on behavioural grounds and are considered a category of disorders, rather than a diagnostic label (Lord & Rissi, 2000).

Although dealing with delays in receiving a differential diagnosis has been reported to be difficult for parents, perhaps the wait or memory of the delay would not be so difficult to endure if it resulted in helpful information for families. Unfortunately, however, parents report that even if a differential diagnosis is identified, this diagnosis does not result in information or supports for the child or the family (Watson, manuscript in preparation).

Perceived Lack of Information

When parents are successful in receiving a differential diagnosis for their child, parents express disappointment with the amount of information that is provided; specifically, parents express frustration and anxiety in not receiving specific medical information about their child (Brogan & Knussen, 2003; Hedov et al., 2002; Skotko, 2005; Skotko & Bedia, 2005; Woolfe & Bartlett, 1996). Some families report receiving no information, good or bad, from the doctor of the hospital (Skotko & Bedia, 2005),

while others state that when information is provided about Down syndrome for example, it is often out of date (Skotko, 2005; Skotko & Bedia, 2005).

Summary

This literature thus demonstrates that families report dissatisfaction with the diagnostic process when a child has a developmental disability. It is of course easy to blame medical professionals because they are often giving disappointing news, but it is important to understand why families are so negative about their experiences with medical practitioners. The following study examines families' lived experiences of the diagnostic process.

Methodology and Methods

Methodology

Basic interpretive inquiry was chosen to address the research question, "what is the experience of receiving a differential diagnosis like for families?", because the overall purpose of this approach is to understand how people make sense of their lives and their experiences (Merriam, 2002). A basic interpretive study draws from phenomenology and symbolic interactionism in particular. Researchers employing this approach are interested in how people interpret their experiences, how they construct their worlds, and what meaning they attribute to their experiences (Merriam, 2002). Constructionism underlies this methodology and recognizes that individuals construct reality in interaction with their social worlds (Merriam, 2002).

Participants

Fourteen parents from thirteen families of children with developmental disabilities participated in the study. A family was defined as anyone that parents considered to be a part of their family and all family members were invited to participate.

Although extended family or other individuals were welcome to take part in the interviews, parents, specifically mothers, made up most of the participants. Of the 14 total parents who participated in the study, 12 were mothers and 2 were fathers. Two additional fathers said that they would be willing to participate, but we were never able to meet due to time demands. All families lived in Canadian urban settings and had experienced the diagnostic process in an urban centre.

Participants included parents of children with an identified genetic disorder or specific differential diagnosis, as well as parents of children with unidentified developmental delay. Both groups were recruited to investigate the diversity of experience and to look at for commonalities in their experience of the diagnostic process among different types of disabilities. Participants had children with a range of developmental disabilities. Five families had children with specific genetic disabilities: Stuart and Francine are the parents of Michaela, who has Angelman syndrome; Gillian and Shannon also have children with Angelman Syndrome; Tom and Kathy have children with rare chromosomal abnormalities. Three families had children with diagnosed autism spectrum disorders, ranging from PDD to Asperger syndrome; Collette and Ingrid each have two children with autism spectrum disorders. Phoebe and Deborah suspect autism spectrum disorders, but a differential diagnosis has never been given. Finally, Judy, Nina, and Theresa have children with unidentified disabilities or nonspecific developmental delay. For those families receiving a differential diagnosis, timing since diagnosis ranged from 2 years to 25 years.

Differentiating between the types of disabilities became more complicated than I had anticipated; I originally thought it would be clear about how to categorize families,

but with the different kinds of diagnoses or behavioural versus genetic diagnoses, this distinction between groups was not as clearcut as I had previously thought. For example, I struggled with how to categorize a suspected autism spectrum disorder or families who disagreed with the differential diagnosis of their child. Nina, for example, has a son who presents several developmental delays, but his official diagnosis is Systemic Onset Juvenile Arthritis. Nina believes there is more going on with her son, Carl, and is still seeking a more specific differential diagnosis, thus I categorized her with the parents of children with unidentified disabilities. The above categorization represents the manner in which I thought it best to classify families and corresponded with families' own beliefs about the label for their child. The one exception is Phoebe, who believes her son has Asperger syndrome, but this is not yet official; I placed her with the other families of children with unidentified disabilities. A summary of participants and their children can be found in Table 1.

Table 1

Participant Descriptions

Name	Child Name	Differential Diagnosis	Categorization
Stuart	Michaela	Angelman syndrome	specific disability
Francine	Michaela	Angelman syndrome	specific disability
Shannon	Eric	Angelman syndrome	specific disability
Gillian	Christa	Angelman syndrome	specific disability
Kathy	Rebecca	extra chromosomal matter	specific disability
Tom	Grace	chromosomal deletion	specific disability
Collette	Claude	autism spectrum disorder	specific disability
	Sam	autism spectrum disorder	specific disability
Farah	Sangeeta	autism spectrum disorder	specific disability
Ingrid	Simon	autism spectrum disorder	specific disability
	Martin	autism spectrum disorder	specific disability
Deborah	Franklin	communication deficit	unidentified disability
Nina	Carl	juvenile diabetes	unidentified disability
Theresa	Curtis	developmental delay	unidentified disability
Judy	Michelle	developmental delay	unidentified disability
Phoebe	Matthew	ODD; ADHD; possible Asperger's	unidentified disability

Procedures

I accessed families through disability support organizations such as the Association for Community Living and the Canadian Angelman Syndrome Foundation. Word of mouth was also used, whereby I contacted individuals who might know families interested in participating in the study. This technique is sometimes called respondent-driven or snowball sampling and is often used in hidden populations that are difficult for researchers to access (Salganik & Heckathorn, 2004).

I invited participants to take part in individual semistructured interviews. In the case where both a mother and father participated in the study (Stuart and Francine), parents were interviewed separately. Trute (1995; 2005) has found gender differences in mothers' and fathers' reactions to disability and recommends that each parent be allowed his or her chance to speak. Interviewing parents separately allowed for a more in-depth analysis of each parent's recollection of the experience.

The process began with an orienting interview where the participant and I went over the study description and consent was discussed. On many occasions, this orienting interview took place over the phone. Following this initial orienting interview, a semistructured interview was conducted, blending more and less structured questions (Merriam, 2002), and lasting approximately one hour. These took place at the convenience of participants and in locations agreed upon by both the participant and myself, such as coffee shops, family homes, and participants' places of employment. Follow-up questions were asked of the participant, using e-mail and telephone conversations.

Each interview involved three segments. The first segment was open-ended and involved asking parents to describe their son or daughter and to tell a favourite family experience. The goal of this was to establish rapport while allowing the parent to discuss either strengths or weaknesses of their child and how these characteristics have affected the family. The second part of the interview focused on the process of learning the child's diagnosis. This portion of the interview was semistructured and involved asking the participant to recount the process of receiving the child's diagnosis, including the initial perceptions and feelings that there might be a concern about their child's development; recalling any emotional experiences related to receiving or failing to receive a differential diagnosis; and asking parents to assess any changes in their thoughts or feelings between the initial perception of developmental differences and a differential diagnosis. For families who had failed to receive a differential diagnosis for their child, questions were adapted to reflect this. It is important to stress here that the questions were open-ended; I asked parents to "describe the diagnostic process" and parents were encouraged to speak freely. The analysis will reveal that parents had quite negative experiences, but I did not prompt this. I was actually quite shocked at the negative tone to the interviews and tried to bring out positive memories, asking what was helpful or positive about the experience. However, I did not want to lead the participants too much and if parents were unable to think of anything positive after prompting or rephrasing the question, I did not persist.

The final segment of the interview was also open-ended and allowed parents to discuss their child's developmental progress, including any experiences that have helped or hindered the family. This also provided an opportunity for parents to make recommendations for making the diagnostic process better for families.

Consent and Ethical Issues

Informed consent was received from all participants and consent was revisited throughout the research process. Munhall (1989) suggests the use of "process consenting," which allows the researcher and participant to assess consent throughout the research process. This provides more protection and freedom of choice for participants since unforeseen issues inevitably arise and risk can never fully be anticipated. This is especially true when dealing with sensitive issues. Process consenting allows the participant the freedom to withdraw from the interview or study at any time and ensures that the participant has a say in confidentiality throughout the research process.

Absolute confidentiality cannot be guaranteed, but reasonable measures of ensuring privacy were undertaken. Participants' names and any other identifying information were changed for data presentation, plus interview transcripts and recordings were kept in a locked filing cabinet to ensure safety of the information. Furthermore, since participants were consulted throughout the study, they were able to make changes as needed to ensure confidentiality to the best extent possible.

Analysis of Interview Data

Interviews were digitally recorded, transcribed verbatim, and rechecked for accuracy prior to analysis. I reviewed digital recordings and transcripts several times to familiarize myself with the content of the interviews. Because I was both the researcher and interviewer, I had more insight and in-context knowledge and was thus able to establish a variety of important links between the research questions and the data gathered (Litosseliti, 2003).

I conducted a thematic analysis of the interview transcripts (Merriam, 1998) by reviewing transcripts, making notes and comments throughout. I followed Merriam's levels of analysis, beginning with a descriptive account, followed by category construction, then making inferences. A descriptive account is a narrative that conveys the meaning the researcher has derived from studying the topic of interest. The second level of analysis is the construction of categories or themes. However, before themes may be created, codes must be organized as a way to organize the data. According to Merriam, "category construction *is* data analysis" (p. 180). Ryan and Bernard (2003) recommend looking for repetitions in the data, as well as looking for transitions and linguistic connectors. In speech, pauses and changes in voice tone may indicate a transition and connectors may indicate a new theme. To document my decision making, I kept a codebook, which included a detailed description of the code, inclusion and exclusion criteria, and exemplars of real text for each theme (MacQueen, McLellan, Kay, & Milstein, 1998).

DeSantis and Ugarizza (2000) make the important distinction between the terms categories, factors, variables, and themes, critiquing much qualitative research that confuses these terms, thus compromising methodological rigor. Themes are described as implicit, implied, and tacit rather than explicit, declared, and easily expressed (Patton, 1990) and a theme "captures and unifies the nature or basis of the experience into a meaningful whole" (DeSantis & Ugarizza, p. 362). Once themes emerged from the data, inferences were made and data were summarized.

Findings

Participants focused on two main issues, including the meaning the diagnosis provided to the family and their experiences dealing with professionals. Analysis of the meaning data will be presented elsewhere (Watson, manuscript in preparation). The present paper will focus on families' experiences dealing with professionals. The main themes discussed by parents were tolerating intensive testing, sensing rigidity in thinking by professional, perceiving pessimistic prognostic information, receiving multiple diagnoses, feeling dissatisfaction with the information provided to families, and encountering a range of professionals. Each of these themes will be discussed in turn, but this section will begin with Shannon's experience, the mother of a son with Angelman syndrome, which exemplifies many of the aforementioned themes:

> I was very concerned about Eric's development. He could not control his movements, his limbs moved constantly, his eyes could not focus, and he had very low muscle tone. I was told he was completely normal… [then] I decided to get a second opinion and saw an older pediatrician who immediately said Eric had brain damage. When I asked how he knew this, he said because Eric had tremors and that is always linked to brain damage…he said the results showed Eric had a very small brain, microcephaly… [We then] saw a neurologist… [She said] with respect to the MRI, although the findings were not completely normal, she did not consider the findings striking or specific… Eric did not meet the definition of microcephaly. She found Eric to be slightly hypermotor for age, but [she was] not convinced of movement disorder per se.

"This test, that test, that test" - Tolerating Intensive Testing with Multiple Professionals

The actual diagnostic testing was a very long and difficult process for families, with parents using words such as "mission" or "saga" to describe the experience. Such undertakings meant long waits for referrals or diagnostic tests, dealing with multiple professionals, and watching their child go through sometimes painful testing.

All families reported intensive testing and dealing with multiple professionals. Testing lasted from 1 year to 7 ½ years, with some parents never receiving a differential diagnosis for their child. Parents described the frustrating process of seeing specialist after specialist, extensive hospital stays, and going back and forth between hospitals. The following excerpt from Francine, the mother of a daughter with Angelman syndrome, illustrates the number of medical professionals that families dealt with in their search for a differential diagnosis:

> Well, there was the pediatrician, the neurologist, there was the eye specialist, the cardiologist, the physical medicine doctor, the dietician, the gastro-intestinal specialist, and kidney specialist, I think that's it… Oh yeah, physiotherapist and occupational therapist, this is all leading up to the diagnosis.

Dealing with multiple professionals was a "very, very frustrating process", reports Farah, the mother of a daughter with an autism spectrum disorder. Much of the frustration was due to waiting for appointments, then being disappointed when more testing was required. Kathy, the mother of a child with a rare chromosomal abnormality, recalls:

> You go into these appointments, you're hoping, you're thinking, "OK, this is the appointment that is going to tell me what she has", and then they say, 'no, it's

> negative. She doesn't have that'. So now we have to do more testing and you always have to wait because you can only see the geneticist once a year.

For the children of nine parents, the diagnostic process resulted in extensive hospital stays and intensive, often invasive procedures. These parents described their child enduring blood tests, skin samples, muscle testing, EEGs, and other procedures. Nina, the mother of a son with unidentified developmental delay, remembers, "We went back to the [hospital] and stayed there approximately 8 weeks. In that time period he was tested for everything imaginable and after many rough days and nights the diagnosis of Systemic Onset Juvenile Arthritis was made." Judy, the mother of a daughter with unidentified developmental delay, also recalls:

> So then I finally got myself a pediatrician and we started the saga. She went into the hospital at 13 -14 months old and she was in for 10 days, ran every possible test they could possibly think…They did X-rays, chest X-rays and all that type of stuff, they did genetic testing, had blood drawn, they did developmental tests.

After the intensive testing, Phoebe, Ingrid, Deborah, Gillian, and Judy grew tired of the process and just wanted to get on with life and deal with the presenting issues. They may have felt pressure from professionals to get a diagnosis or wanted to discontinue the process due to guilt for putting their child through an arduous experience. "I don't want his whole life spent in and out of doctor's offices", states Phoebe, the mother of a son with an unidentified autism spectrum disorder. Judy recounts her decision to stop the diagnostic process:

> She must have been 2 ½ when finally I said, that's enough you know, none of this running back and forth because we had the [occupational therapist],

> physio[therapist], psychologist, doctors, speech therapist, whole gamut. Finally I thought about this and I thought, "You know, never mind what it is that she's got, what it's called, or whatever, we just have to get on with raising this child!"

Although Francine, Stuart, Deborah, Tom, Kathy, Phoebe, Judy, and Nina reported that their child coped well with the testing, seeing their son or daughter fearful or in pain was very stressful for families. Parents asserted the importance of going through the process, but felt tremendous guilt. Kathy, the mother of a daughter with a rare chromosomal abnormality, expresses her concern and recounts that she would ask herself:

> "What am I putting my child through? Getting poked, getting probed." Some of the testing was so hard on her, like they're freezing and snipping her muscles. You feel sorry for them, but yet you have to do it. It was nice to finally get it over with and not put her through any more torture.

Collette, Shannon, Phoebe, Ingrid, and Farah also struggled observing their children dealing with IQ tests or other developmental batteries. As Collette, the mother of two children with autism spectrum disorders, remembers,

> The most difficult was watching Sam struggle and fail on the most obvious tests. The opening to the clear plastic box was on the side. The tester put the car in the box in front of Sam and he couldn't figure it out. She showed him again and again and he couldn't figure out how to get the car. That was very hard to watch.

Six parents also said that they saw their children being treated poorly by medical professionals, and this was difficult. Phoebe, the mother of a child with an undiagnosed

autism spectrum disorder describes one instance where a pediatrician was insensitive to her son:

> He was quite nasty for the first 10 minutes, almost mean, very sharp and short with Matthew… He said, "This kid sounds like a nightmare! My kids would never behave like this." As soon as he said this, you could see Matthew wince. He insulted me and he insulted Matthew. What this did essentially was destroy all of the work that we had done with Matthew, with that one careless sentence.

Parents also felt that doctors limited themselves by relying on diagnostic tools. Stuart, Ingrid, Farah, Phoebe, Ingrid, and Gillian discerned that doctors were not looking for new information or approaches to disability identification, but rather relied on "checklists", without completing a thorough observation of the child. Phoebe, the mother of a son with an unidentified autism spectrum disorder, describes the diagnostic process:

> The doctors ask the same questions over and over… There was a piece of paper at the table that I filled out, and the psychologist didn't even talk to Matthew… she has spent 5 minutes alone with him in the 2 years we have been seeing her… He has only taken one test with a psychologist.

It is important to point out that this experience seemed to be more common in children with autism spectrum disorders. Farah, Phoebe, Deborah, Ingrid, and Collette, who are parents of children with diagnosed or suspected autism spectrum disorders complained about "checklists", while Francine, Gillian, Stuart, Judy, Shannon, Kathy, and Nina, who have children with genetic disabilities or without communication deficits discussed the invasive testing. This variation in experience is a reflection of the

differences between differential diagnoses and will be addressed in the conclusion section of the paper.

It is important to point out that Tom, Francine, Judy, and Collette acknowledged that some medical professionals were very helpful, as will be discussed below, but nearly all parents reported that by and large, doctors were not sensitive to their needs. Such insensitivity took the form of rigidity in thinking and tactlessness when providing diagnostic information.

"The doctor told me I was crazy" - Sensing Rigidity in Thinking by Medical Professionals

Parents said that some professionals did not listen to parental concerns and relied more on diagnostic tools such as "checklists." Parents perceived this as "being rigid" (Farah). Nine parents reported discontent with professionals' receptiveness to their anxieties about their child, which was often reflected in the manner in which the diagnostic process was conducted.

All parents suspected that something was wrong with their child before the medical professionals; Francine and Kathy even suspected difficulties prenatally. "The diagnosis wasn't all that big of a deal. I knew there was a developmental delay, I figured that out, I was just waiting for other people to figure it out," recalls Judy, the mother of a daughter with unidentified developmental delay. Nine parents described having to fight to get doctors to take their concerns seriously. As Judy remembers, "The doctor not listening to me was the most difficult part of the process and I'm sure every parent has their own version of that story."

Two families reported doctors responding immediately to family concerns. Francine and Stuart's doctor, as well as Tom's daughter's pediatricians acted quickly when the parents were worried about their child's development. Michaela and Grace were both hospitalized right at birth and these parents maintained close contact with professionals throughout their daughters' infancies. At 3 months, Michaela's pediatrician was concerned that she had a head lag and supported by Francine's and Stuart's concerns, initiated an intensive diagnostic process at 6 months. Grace was diagnosed with a terminal chromosomal deletion at approximately 18 months of age quite quickly after Tom and his wife noticed developmental delays.

Eight parents, however, reported doctors telling them they were overreacting, even when children were presenting significant developmental delays. Ingrid, the mother of two sons with autism spectrum disorders described years of doctor visits and bringing articles on autism, but "The family doctor told me I was crazy" and she was offered parenting books. When she asked for a referral to a psychologist, the doctor would not provide one. Because of the lack of credence given to her concerns, Judy, the mother of a daughter with unidentified developmental delay, recounts doubting her intuition and speculations of disability, even though she suspected for quite a while that something was going on with her daughter:

> You know some of those early developmental things that should have been happening by then at 5-6 months. So then I voiced my concerns to the doctor and he said, "oh, go home, she'll be fine. If you keep worrying, you're going to make yourself old before your time. I'm thinking, "OK, New Mom, I am crazy. I'll take her home and I'll just fumble through some more."

Because parents felt that doctors were not listening to concerns, Nina, Gillian, Deborah, Shannon, Phoebe, and Ingrid grew frustrated and in order to initiate the diagnostic process, they believed they had to be persistent. Unfortunately, it often took significant medical issues before medical professionals would get the process started. Nina, the mother of a son with unidentified developmental delay, recalls, "Once he was so sick he couldn't move, people listened. Before that he was ill quite often for about a year and I was told it was just a virus, time and time again that [it] would go away. Some virus." Shannon, the mother of son with Angelman syndrome, also describes her discouragement during the route to differential diagnosis:

> I was tired of being told there was nothing wrong with him or he was colic [sic]. When you are a mother of two, you know when something is wrong with your child. I just kept coming back until they agreed to do some testing.

Such persistence often led to conflict between parents and medical professionals. Phoebe, the mother of a son with an unidentified autism spectrum disorder describes one such encounter where the doctor said, "You're just one of those hysterical mothers who will take your child from doctor to doctor until you get the diagnosis that you want." Shannon, the mother of a son with Angelman syndrome recounts similar treatment:

> The most difficult part of the process was being treated as a parent who worried over nothing. I felt every time I walked into the doctors' office or the emergency room, they thought, "Oh god, here she comes again!"

Statements such as these unfortunately gave rise to alienation between doctors and families.

Parents also talked about medical professionals being close-minded when conducting the diagnostic process. Ingrid, Phoebe, Deborah, Stuart, and Gillian objected to an overreliance on diagnostic tools or "checklists," while Farah and Phoebe complained about filling out form after form, with the doctors rarely spending time with the child being diagnosed. Farah, the mother of a daughter with an autism spectrum disorder, narrates her experience:

> The most difficult part of this process was in having to face doctors who could not accept individuality as a part of autism and in being sent from one specialist to the next because she didn't fit the descriptions, as they knew them. The fact that formal testing was not conducted on her; that she was not interviewed or my notes even glanced at, led me to believe we would never get a diagnosis. Most doctors let her sit in while I was being interviewed and after a few minutes of informal observation, usually declared her normal… With regard to the term autism, it was me who brought in the studies, documentation and checklists in order to convince doctors she had autism. When confirmations were finally given, each doctor consulted his or her own textbook and gave me a different diagnosis of autism. I was very, very frustrated.

Throughout the diagnostic process, families lived through extensive and often invasive testing, felt that medical professionals were not listening to their concerns, and perceived doctors as narrow in their approach to disability identification. These experiences were all leading up to receiving the actual differential diagnosis. Unfortunately, receiving the differential diagnosis and prognostic information was also painful for families and parents spent a lot of time discussing their recollections of the

receipt of the actual differential diagnosis and the accompanying prognostic information. The difficulty was felt either in the manner in which the diagnosis was given, or in receiving the actual information presented regarding the disability. If a differential diagnosis was provided, most families received the information in person, although three families received the differential diagnosis in a letter that was mailed to the home. Regardless of the mode in which the diagnosis was given, right from the start, a negative construction of disability was presented. Two salient themes emerged: dealing with an overabundance of pessimistic information provided about the disabilities and receiving multiple and incorrect diagnoses. Each of these themes will be discussed next.

"He would be placed in a home" – Perceiving Pessimistic Prognostic Information

When medical professionals delivered diagnostic information, the conversations often began with negative language. Deborah, the mother of a son with an unidentified developmental delay, reports being told by her doctor, "something was terribly wrong." This attitude continued when the medical professionals provided information regarding prognosis. Families felt that doctors either were pessimistic regarding the child's future functioning, might have provided information that was not deemed relevant for the time, or made hasty speculations regarding the differential diagnosis; these experiences were frightening for families.

When presenting the family with what to expect regarding the differential diagnosis, Francine, Stuart, Shannon, Gillian, Collette, and Tom felt that doctors were overly pessimistic regarding the child's future functioning. This was not perceived as helpful or sensitive when families were first presented with diagnostic information. Tom, the father of a daughter with a rare chromosomal deletion, remembers, "He turned to us

and said, 'Oh, I think she will walk' (Tom laughs). We were fully expecting that she would walk, what did he mean, YOU THINK SHE WILL WALK"!? Collette, the mother of two sons with autism spectrum disorders, also recalls, "He talked to us for almost 2 hours and the part that stuck with me the most was him telling us that Claude would talk through a computer, would be difficult to potty train and that our goal was to keep him from being institutionalized." As Collette's quote demonstrates, when prognostic information was provided to parents, it often dealt with issues later on in the child's life, concerns which were considered not relevant for the time, especially when families were dealing with an unexpected diagnosis or difficult news. For example, seven parents were provided with information regarding reproductive issues or future placement in group home settings. Stuart, the father of a daughter with Angelman syndrome, recalls:

> We went to see the medical geneticist and she is talking about this stuff and all of a sudden she starts talking about any offspring she will have. No, that Michaela will never have children because any offspring she would have would also have the defective gene.

Information regarding reproduction was not deemed relevant to the family, considering their daughter was 10 months old at the time of diagnosis.

Finally, nine parents discussed a perceived lack of sensitivity when doctors provided tentative or speculative diagnoses. As Theresa, the mother of son with unidentified developmental delay, remembers:

> He was going to a pediatrician at the time and he said, "I just don't know why he isn't developing, maybe he has muscular dystrophy." Can you imagine telling a parent that? I cried the whole way home.

Nine parents felt that the provision of spontaneous theories was "insensitive" and "frightening." Such speculative diagnoses bring us to a final issue, the communication of multiple and incorrect diagnoses.

"Every week it was something different"- Receiving Multiple and Incorrect Diagnoses

The above theme name is taken from an interview with Kathy, the mother of a daughter with a rare chromosomal abnormality. Farah, the mother of a daughter with an autism spectrum disorder also recalls, "all in all, my daughter received five or six different diagnoses, including schizophrenia. One well-known pediatric specialist told me I had too many children and that was all that was wrong with my child." All parents reported multiple diagnoses for their child, which were very stressful for families. The following quote from Collette, the mother of two sons with autism spectrum disorders, exemplifies this experience:

> At 1 year we were told he absolutely and definitely did not have autism, at 2 he had an expressive and receptive language disorder, at 3 he had complex behavioural developmental disorder and at 4 we were told that "even though he is more social than typically seen in children with autism, given the fact that he has a brother with autism I suspect he is within the spectrum."

The receipt of multiple diagnoses or theories was described as an "emotional roller coaster" by some parents, and led to anger for others. Francine, the mother of Michaela, remembers:

> There were lots of different theories. I think at one point they told us her problems were due to lack of white matter in the brain and then yeah, every week it was something different. It was an emotional roller coaster. People would tell you, "oh

> yeah, she might have some physical problems, but cognitively she is bright as a button and totally fine that way." We would be really worried, then we might be relieved about something. Some tests would rule out something bad and we would be happy, then someone else would bring up another concern they had about her and we would be upset again.

Stuart, Francine's husband, also described his frustration with the theory of lack of white matter, "What made me really mad was that neurosurgeon who said that he could 100% guarantee this is the problem and he was completely wrong." He further discusses the conflicting information their family received:

> I mean a week before we had taken her into the doctor and the same doctor who is telling me that she is severely cognitively impaired was telling me that regardless of what it is, we know that she is very bright.

Phoebe, the mother of a son with an unidentified autism spectrum disorder, also expressed her frustration at the theorizing by professionals. Her son, Matthew, has received diagnoses of oppositional defiant disorder (ODD) and attention deficit hyperactivity disorder (ADHD) over the years. Phoebe complains, "Funny how they threw diagnoses around, let's put him on drugs. You can't just throw these diagnoses around like that!" It thus appears that parents resented speculative diagnoses, feeling they were provided with very little grounding.

Phoebe's comments also bring up another family concern, which was medication. Because multiple or speculative diagnoses were provided, diverse medications were often prescribed, including antipsychotics. "Doctors were always willing to give us psychiatric drugs without any reasons for the problems," reports Nina, the mother of a son with an

unidentified developmental delay. Phoebe, Stuart, Deborah, Shannon, and Nina were very uneasy medicating their child, especially when a differential diagnosis was not given. This provision of medical intervention leads us to the final area of parental concern, which was the dearth of information provided to families about disabilities and how to provide remediation for their child.

"Give diagnosis, wish you well, and then you go home" - Dissatisfaction with the Information Provided to Families

The above theme name came from an interview with Ingrid, the mother of two sons with autism spectrum disorders. All parents reported receiving minimal information regarding intervention programs or supports. Farah, Ingrid, Shannon, Gillian, Stuart, and Phoebe felt that each doctor gave their own advice, each conflicting and contradicting the next, and that none of the advice was grounded in recent research. The two main areas of disappointment were in the provision of details about the disabilities and information about disability services and supports.

Lack of disability information. The first area of dissatisfaction for families was in the details communicated regarding disabilities. Parents were hoping for recent research about what to expect about their child's functioning and how to provide the best possible supports, but the medical professionals who presented the diagnostic information were able to present very little. Francine, the mother of a daughter with Angelman syndrome, describes her experience:

> So she gave us a bunch of journal articles that she printed off and I don't think that was very good either because they were older journal articles from the 60s and 70s and they had these kind of unattractive pictures of people with mental and

> physical disabilities in the article, that really scared us and I don't think that was a great time to be given that. I mean, she wanted to give us information and there is not a lot out there so she went to her medical books.

Francine was thus disappointed with information being provided right after the diagnosis, when she was dealing with quite a shock, and was also disconcerted by the lack of recent descriptions of developmental disability. For diagnoses such as Angelman syndrome, which is quite rare, some medical professionals appear unable to give many details to families. Tom's doctor was also unable to give appropriate information:

> And they said to us, this isn't like Williams, it isn't like Down syndrome where we can tell you that this is likely going to be, we just had to go day by day. They gave us one article, they said this is very rare… they said that's it, we can't tell you anything else... So they said you're going to have to take it day by day, so away we went.

Tom's description brings us to the final issue recounted by parents. Families were seeking access to disability services and were looking for guidance about where to turn. However, parents felt that doctors did not appear to have this information.

Lack of information regarding services or supports. Parents were seeking access to funding and services in order to best support their child, but once a diagnosis was presented, medical professionals appeared ill-prepared to deliver support information. As Francine recalls,

> We thought that as soon as we got the diagnosis, people would be knocking on our door saying, "OK, now this has happened, this is what we are going to hook you up with, these are the services, and someone will be calling you," but there

> was nothing. They gave me a phone number for [community agency] and tell you to phone them. We didn't even know what they were - what they could do for me…

"Some people are good and some people are bad" – Encountering a Range of Professionals

Despite the pervasive negative tone to the interviews, Tom, Francine, Judy, and Collette discussed the positive encounters they had with medical professionals. That being said, the above theme name, taken from an interview with Tom, demonstrates how parents discussed the positive aspects of the diagnostic process. There always appeared to be a qualifier to the positive statements. As Collette, the mother of two sons with autism spectrum disorders recalls, "the nurse was of some support, but it was really fleeting."

Francine, the mother of Michaela, who has Angelman syndrome, detailed how the doctor who provided the differential diagnosis "tried to put a positive spin" on the diagnosis. She recalls,

> I remember her telling us that Michaela would be able to participate in family life to some degree… and she said, "oh, she will be able to watch TV with you." And she thought that was a good thing but I remember thinking how horrible that that was the best thing she could say… That's not really comforting to me. I'm sure she meant it to be, she was trying to be helpful.

Tom, the father of Grace, who has a rare chromosomal abnormality was the most positive in his discussion of medical professionals. He remembers:

> I think with the exception that I mentioned, the medical system was a positive experience, I mean the pediatrician was close to retirement, he was revered and is

> still revered… but I think there was a bit of a feeling that maybe he wasn't pulling out all the stops, maybe he wasn't exploring things carefully enough, but in general the system was good.

Tom's experience appeared to be a bit different from the other parents because as discussed above, Grace was hospitalized promptly after her delivery and most of her care was centralized to one medical team at one central location. Although the process "dragged on for quite while," he felt that "some of the greatest pediatric minds in the world" were taking care of his daughter.

Summary

From interviews with families of children with disabilities, it thus appears that the diagnostic process is a difficult one. Many families face intensive testing, perceive rigid thought processes in medical professionals, feel that doctors are pessimistic about disabilities, and are dissatisfied with the support and intervention information provided. It is also important to note that some parents discussed the different medical professionals they encountered, and how some were more helpful than others. It is unfortunate that this process was so negative for the parents I interviewed because families are now facing a lifelong relationship with medical professionals. The final section of this paper will discuss suggestions for practice and how this process could be improved for families.

Conclusion

This study found that the parents interviewed unfortunately faced negative encounters with medical professionals as they sought a differential diagnosis for their child. One of the most salient themes of the interviews with parents was that they did not feel doctors listened to their concerns about their child. This lead parents to doubt their

abilities as parents and initiated what I perceived as alienation between families and professionals. It has been suggested that clinicians hesitate to address families' initial concerns about their child's atypical development because they lack information about developmental disabilities (Goin-Kochel et al., 2006; Hasnat & Graves, 2000). There is an absence of epidemiological research on autism (Brogan & Knussen, 2003; Fombonne, 2001) and confusion for professionals surrounding the diagnostic criteria for Asperger syndrome and other autism spectrum disorders (Brogan & Knussen, 2003). In other disabilities such as Angelman syndrome, which is a genetic disability due to mutation, the diagnosis can be genetically confirmed in 80% of cases, but 20% of cases are diagnosed clinically and there is often uncertainty about the correctness of the diagnosis (Williams, Lossie, Driscoll, & the R.C. Phillips Unit, 2001). Williams et al. (2001) cite several mimicking conditions for Angelman syndrome, including Rett syndrome, PDD, and childhood autism. Such confusion obviously contributes to the multiple and incorrect diagnoses imparted to families.

It is important to stress that medical professionals are trying under pressure to do their best. Families are seeking a differential diagnosis and medical practitioners are perhaps feeling the pressure to come up with a name for the disability. Furthermore, given the confusion regarding autism spectrum disorders and the discovery of newer genetic disabilities, doctors must try to rule out similar diagnoses. Parents of children with autism spectrum disorders, in particular, reported an overreliance on diagnostic checklists, while parents of children with genetic disorders were concerned with the lack of up-to-date information about their child's differential diagnosis. These unique

concerns deserve further investigation, but also point to medical professionals' lack of knowledge about both kinds of diagnoses.

If medical professionals are uninformed about disabilities, perhaps they should use families as a source of information, many who reported having done their own research regarding differential diagnoses. From interviewing parents, I gleaned that parents would appreciate medical professionals conducting a brief preliminary evaluation as soon as parents express concern. The advantages of using parents' concerns in the diagnostic process are that they are easy to elicit and only take only a few minutes of professional time (Glascoe, 1999). Eliciting parental concern also takes less time than a formal screening instrument (Glascoe, 1999; Sices et al., 2004), plus provides a family-focused and collaborative approach to addressing behavioural and developmental problems.

Such a collaborative approach may improve relationships between families and medical professionals. Pediatricians report being upset when informing parents that their child has a disability; however, they also stress the importance of professional detachment because "if you get caught up in emotions you can't achieve much" (Hasnat & Graves, 2000, p. 30). Many clinicians admit they have little, if any, training on how to deliver diagnostic information in a sensitive manner (Hasnat & Graves; Ralston, 2000). Ralston discusses his medical school experience:

> What I was taught in medical school and in my training is that disability - no matter what its form - is a bad thing and to be avoided at all costs. Lectures or seminars on Down syndrome or other genetic syndromes were geared toward the description of the abnormalities and the efforts that can be made to prevent the

> problem in the first place; that children with congenital diseases may find their lives to be rich and valuable was hardly recognized, much less stressed. (p. 335)

Perhaps showing empathy and emotion would help parents, rather than providing detached, pessimistic information about disabilities. Patterson (1989) recommends that medical practitioners aid the family in appraising their strengths relative to the demands of the differential diagnosis. Encouraging and directing the family in "developing and acquiring additional resources (such as support groups) would be an additional kind of intervention..." (pp. 113-114). She further discusses how community resources such as funding bodies and intervention programs can become both a source of strain and support, as evidenced in this study. Listening to families as disability advocates may further serve to ameliorate physician perceptions about disabilities and how to talk about them. Such communication may also help to coordinate services in order to provide families with information regarding funding and intervention support. As Francine, the mother of a daughter with Angelman Syndrome states,

> I think it would be great if the doctors' offices, once they had given you the diagnosis, did line some things up, at least initially for you and put some things in place, and then once you get yourself together then you could say, OK.

It is important to note that finding out that your child has a developmental disability and being concerned for your child will never be completely positive, but the process may not have to be as difficult as these parents reported. Considering family members as part of the diagnostic team and responding quickly to parental concerns will increase family satisfaction with the diagnostic process and would also expedite the process by eliciting initial information from families. Asking family members what they

would like out of a differential diagnosis would also be helpful, so that medical professionals know what kind of information would aid families during a potentially stressful time.

Speaking to medical professionals about their experiences of the diagnostic process would also be beneficial. I interviewed parents, who provided their perceptions of what practitioners said, but having both sides of the story would be beneficial. Telling a family that their child has a developmental disability and providing potentially frightening prognostic information is undoubtedly a difficult experience. Understanding professionals' perspectives would help to tailor their training about how to make this process better for everyone involved.

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Why do Parents Seek a Differential Diagnosis?

A Search for Meaning and Support

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Abstract

This basic interpretive study addressed the reasons why parents seek a differential diagnosis for their child who has a developmental disability. Fourteen parents were interviewed about why they sought a label for the disabilities of their child. Participants included six parents of children with identified genetic conditions, three parents of children with diagnosed autism spectrum disorders, and five parents of children with unidentified developmental disabilities. Parents described searching for a diagnosis because of the importance of "knowing," understanding the cause for the disability, gaining knowledge about future expectations and appropriate interventions, and gaining access to funding or specific services. An application to Patterson's (1988, 1989) Family Adjustment and Adaptation Response model (FAAR) is presented.

Why do Parents Seek a Differential Diagnosis?

A Search for Meaning and Support

The value of labels has been debated in the literature (Gillman, Heyman, & Swain, 2000; Lauchlan & Boyle, 2007; McDermott, Goldman, & Varènne, 2006) and many theorists disagree about the value of labelling a disability. However labels can serve an important function in providing information regarding strengths and weaknesses (Griffiths & Watson, 2004), as well as in accessing services and funding. Families often embark on a major quest for a differential diagnosis for their child, but the American Association on Mental Retardation (AAMR; 2002) reports that 30 – 40 % of individuals with developmental disabilities do not have a specific diagnosis. Consequently, many families may search for a differential diagnosis for a prolonged period of time or never receive a specific label for the disability of their child.

This study is an investigation of the family experience of the diagnostic process and asks what meaning a differential diagnosis provides to families. More specifically, it seeks to understand why families search for a differential diagnosis for their child. For the purposes of this study, the term *differential diagnosis* will refer to a specific diagnosis, such as Angelman syndrome or autism, contrasted with a nonspecific diagnosis such as global developmental delay. The present study extends the literature on family adaptation to diagnosis of a disability by using an interpretivist perspective (Merriam, 2002) to examine the ways in which families adapt to their child's differential diagnosis or lack of differential diagnosis. I interviewed both families of children with a differential diagnosis as well as families of children who are still looking for a specific

diagnosis. Following the interviews, I analyzed the data using thematic analysis (Merriam, 1998).

Literature Review

The following literature review begins with a brief description of the theoretical approaches typically applied to research on families of children with developmental disabilities and then addresses research conducted specific to differential diagnosis. The Double ABCX model of family functioning (McCubbin & Patterson, 1983, 1987) will be discussed, followed by the Family Adjustment and Adaptation Response (FAAR; Patterson, 1988, 1989) model. This theoretical discussion will be followed by the limited literature on differential diagnosis and families.

Research on the experience of parenting a child with a disability often applies the Double ABCX Model of family functioning (McCubbin & Patterson, 1983, 1987). Several variations of this model have been proposed, but the basic model describes how families adjust and adapt to crisis situations. The model proposes that familial adaptation (*XX*) to a crisis is shaped by the following factors: severity of the stressor (e.g., severity of the child's disability) and pile-up of demands or additional life stressors (*aA*); the family's resources, capabilities, and strengths (e.g., social support) (*bB*); and the family's subjective definition of the stressor and its effect on the family (*cC*). Each of these factors build up over time, explaining the double *a*, *b*, *c*, and *x* factors. This subjective meaning reflects the family's values and previous experience in dealing with change and crisis (McCubbin & Patterson, 1987). Hodapp, Dykens, and Masino (1997) state that although this model has proven useful, many aspects remain vague, specifically, the individual characteristics of the child, which influence the *aA* and *bB* factors. Such a critique is

relevant to the present study, which focuses on differential diagnoses and the concerns of families with specific kinds of developmental disabilities.

The FAAR model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) is similar to the Double ABCX model (McCubbin & Patterson, 1983, 1987) and may be considered an elaboration, amplification, or reconsideration of the model. While the Double ABCX Model considers how various factors interact to produce a degree of adaptation at any given time, the FAAR model is focused on the balance of these factors and how the resulting state of adaptation develops over time. An important component of the model is called *appraisal,* which plays an essential role in moderating child and environmental factors. Appraisal is how an event is perceived by an individual, how one evaluates a situation in terms of its relevance to oneself, such as one's goals or well-being (Lazarus, 1966). From an interpretivist perspective, appraisal might be seen as a lens through which parents view their child and other environmental factors. The presence of appraisal as a factor in the model implies that "objective" measures of child factors such as age, intelligence, diagnosis, and behaviour may be less significant in determining parental responses than the parents' interpretation of these characteristics. One parent may view the same differential diagnosis and the same behaviour as catastrophic, while another may view it as a minor concern. While parental predisposition and parental choice may be important factors in parental appraisal, professionals, social contacts, and the world in general may play a major role in influencing parental appraisals of their children's conditions. Therefore, research that looks at the effects of specific diagnoses on parental responses may reflect unique child characteristics but might equally reflect the socially-constructed and shared appraisals that influence parental responses.

The FAAR singles out two important factors in appraisal more precisely, situational meanings and global meanings (Patterson, 1988, 1989). Situational meanings refer to specific appraisals of challenges and resources in a specific circumstance (e.g., getting help for the child at school, responding to in-law criticism of parenting) while global meanings refer to a more general set of cognitive beliefs about family, community, and life in general (Patterson). Patterson and Garwick (1994) more recently proposed that global meanings can be further divided into family identity and family worldview. Family identity addresses how the family views itself, including relationships, roles, and norms. Family worldview addresses how the family interprets reality, their core assumptions about their environment, and their existential beliefs, such as their purpose and place in life. Situational meanings and global meanings continuously interact with each other. Global meanings act as templates that help determine how an individual will respond to specific situations. Successes or difficulties in applying situational meanings produce frequent refinements and occasional reformulations of global meanings.

Both situational and global meanings are significant for the present study because they can add to or reduce stressors or strains that may occur. Specific to the diagnostic process, Patterson (1989) states that for a diagnosis to be accepted, it must somehow fit with the family's existing set of beliefs, which are integral to their worldview. Therefore, if a family is not comfortable with a diagnosis or does not understand the diagnosis, this will upset both their global and situational meanings, which may lead to disruption in functioning, called maladaptation.

Addressing meaning is important for both parents and for interpretive research. In a study of parents of children with Asperger syndrome, Pakenham, Sofronoff, and

Samios (2004) found that parents' ability to ascribe meaning to their child's disability and their own parenting experiences had a significant influence on their experiences. Parents in this study adapted better when they were able to make meaning of the disability. The emphasis on meanings in the FAAR model is therefore imperative for studying family experience and critical in an interpretivist study, since the study endeavours to understand the meaning of differential diagnosis to the family. The following section will outline the limited research that has been conducted on families and their search for differential diagnosis.

Diagnosis Research

Most people seek explanations for events and experiences that they regard as illness or disability for several reasons. Griffiths and Watson (2004) discuss the perceived benefits of differential diagnosis, including increased support for families, caregivers, and the individual; knowledge regarding the individual's learning challenges and strengths; as well as knowledge regarding medical or mental health risks and resiliencies. Clinically, it can be important to know about genetic diagnoses because the underlying genetic mechanisms are related to phenotypic differences among individuals with different etiologies for a genetic syndrome or any other condition (Summers & Pittman, 2004) and many disorders are associated with specific medical conditions that can affect quality of life. It is essential to recognize that these benefits are apparent, whether the differential diagnosis is of genetic origin or not. Differential diagnoses provide information and may allow families to make more informed decisions about their child's health and supports.

In Gillman et al.'s (2000) study of individuals with developmental disabilities, their families, and professionals, it was found that parents of children with disabilities

sought a differential diagnosis in order to relieve the stress or ambiguity of the unknown and in the belief or hope that identification and classification of the symptoms would result in treatment, intervention, and social support. Families also believed that a differential diagnosis would finally lead to an improved quality of life for the family and the individual. Gillman et al. also talk about a hierarchy of disabilities, discussing how some labels are more stigmatizing than others. These authors provide the example of a hearing or visual impairment, which is often perceived as less stigmatizing than a learning difficulty. They further maintain that families may search for an alternative label that is regarded by the person or society as less disgraceful. It is also critical to recognize that some labels might be more stigmatizing to the child and some labels, such as Fetal Alcohol Spectrum Disorder, may be more stigmatizing to the parent.

Burden (1991) postulates that not having an understandable cause for a disability is one of the reasons parents may experience guilt in reaction to having a child with a disability. Burden suggests that when the etiology of the disability can be satisfactorily explained, there will be a reduction in guilty feeling, but as long as the etiology "remains shrouded in mystery, the likelihood of self-blame is greater" (p. 333). This is supported in Burden's study of psychosocial transitions in the lives of parents of children with disabilities. Burden studied three groups of mothers in London, England. One group of mothers were recipients of service after the differential diagnosis of the child's disability, a comparison group consisted of mothers whose children had been similarly diagnosed but did not have professional support until the child was 2 years of age, and a third group of mothers whose children's disabilities had not been formally diagnosed participated in the study. Burden found that mothers of children without a differential diagnosis had

more difficulty working through their feelings of anger and guilt than mothers in the other two groups. Mothers in this group also reported higher levels of family crisis and were less successful than other groups in working through these crises.

Families have also cited benefits to receiving a differential diagnosis for their child. In their questionnaire study of 254 families of children with Fragile X syndrome, Carmichael et al. (1999) found that most families considered having a diagnosis a benefit rather than a disadvantage. Families have reported benefits of receiving a differential diagnosis, including appropriate intervention (Carmichael et al.; Poelmann, Clements, Abbeduto, & Farasad, 2005), contact and support from other families (Carmichael et al.; Wilcox, 1991), and help explaining to their friends (Carmichael et al.; Gillman et al., 2000). Being informed also plays a part in parents' sense of control, which has been found to contribute to parents' positive adaptation to having a child with a disability (Knox, Parmenter, Atkinson, & Yazbeck, 2000). When parents do not know what is unique about their child, they feel a lack of control over the situation because they do not have the pertinent information to help their child (Knox et al.). Patterson (1989) highlights the importance of control when looking at the resources of a family. Resources include intelligence, knowledge and skills, time, health, and other factors, but also a sense of mastery, which Patterson describes as the belief that one has some power over the circumstances of one's life. If one has limited resources, then this can lead to maladaptation. Maladaptation is also the result when families have difficulty attributing meaning to an experience.

The meaning a differential diagnosis provides to the family can contribute to coping processes and social support (Pakenham et al., 2004). Even when the formal

information is "unpalatable" (p. 12), Leonard (1999) has found that parents prefer "knowing" to the frustration and anxiety associated with the uncertainty of not knowing what is wrong with their child. Although there have been a few studies on the benefits of receiving a differential diagnosis, little is known about the effects on the family of nonspecific developmental delays or disabilities of unknown etiology (Keogh, Garnier, Behnheimer, & Gallimore, 2000).

Trute (2005) believes that the greatest stressor for parents is "not knowing" and this contributes to the fear and anxiety experienced by parents. Not knowing may take several forms; this could include not knowing how or why the child has a disability, not knowing what to expect in the future, or not knowing if there is something useful that would help the child. Not knowing is also a concern for parents of children with diagnoses such as autism or pervasive developmental disorder (PDD) because these diagnoses are made on strictly behavioural grounds (Lord & Rissi, 2000), leaving many parents wondering whether the diagnosis of autism is "correct." In other disabilities such as Angelman syndrome, the diagnosis can be genetically confirmed in 80% of cases, but 20% of cases are diagnosed clinically and there is often uncertainty about the correctness of the diagnosis (Williams, Lossie, Driscoll, & the R.C. Phillips Unit, 2001). Rett syndrome, PDD, and childhood autism have been cited as mimicking conditions for Angelman syndrome (Williams et al.).

It is vital to recognize, however, that seeking a differential diagnosis is rarely a positive experience for most families. Several researchers describe the negative perceptions of parents in dealing with professionals while seeking a diagnosis for their children (Leonard, 1999; Pianta et al., 1996; Poelmann et al., 2005; Watson, manuscript

in preparation; Woolfe & Bartlett, 1996). Leonard also describes parents who discover the differential diagnosis by accident or parents whose doctors have withheld the diagnosis in trying to protect the family, thus resulting in a negative experience.

Although many researchers have theorized about the perceived benefits of receiving a differential diagnosis, few studies have actually asked families why they want a diagnosis and what meaning a diagnosis would provide. Moreover, the limited studies that have been conducted are from the United States and the United Kingdom. Since meaning is socially constructed (Crotty, 1998) and the diagnostic process differs internationally, a Canadian perspective is relevant. Finally, little is known about the experience of families who do not receive a differential diagnosis and an exploration of this experience is significant. The following study addresses the meaning of a differential diagnosis and why families seek such a diagnosis for their child.

Methodology and Methods

Methodology

A basic interpretive approach, drawing from phenomenology and symbolic interactionism, was chosen to address the research questions because the overall purpose of this methodology is to understand how people make sense of their lives and their experiences (Merriam, 2002). Such an approach allows researchers to uncover what meaning individuals attribute to their experiences, how people interpret their experiences, and how they construct their worlds (Merriam).

Constructionism underlies this approach to research and recognizes that individuals construct reality in interaction with their social worlds (Merriam, 2002). As discussed by Crotty (1998), "meaning is not discovered but constructed. Meaning does

not inhere in the object, merely waiting for someone to come upon it... Meanings are constructed by human beings as they engage with the world they are interpreting" (pp. 42-43).

Procedure

I recruited family members through respondent-driven or snowball sampling, a technique often used in hidden populations that are difficult for researchers to access (Salganik & Heckathorn, 2004). Families were contacted through disability support organizations such as the Association for Community Living and the Canadian Angelman Syndrome Foundation. I also used word of mouth, communicating with professionals I knew who might know families interested in participating in the study.

Participants. Fourteen parents participated in the study, representing 13 families of children with developmental disabilities. A family was defined as anyone the parents considered to be a member of their family and all family members were invited to participate. Although extended family or other individuals were open to partake in the interviews, parents, specifically mothers, made up most of the participants. Twelve participants were mothers and two were fathers. Unfortunately busy time schedules prevented meeting with two additional fathers who wanted to participate.

Families had children with a range of developmental disabilities. Six parents representing five families participated who had children with specific genetic disabilities: Shannon and Gillian have children with have Angelman Syndrome; Stuart and Francine are also the parents of a daughter with Angelman syndrome; Tom and Kathy both have daughters with rare chromosomal abnormalities. Three parents, Ingrid, Collette, and Farah, participated who had children with diagnosed autism spectrum disorders, ranging

from PDD to Asperger syndrome, with two of these families having two children with disabilities. Finally, five parents, Nina, Judy, Deborah, Theresa, and Phoebe, participated who had children with unidentified disabilities or nonspecific developmental delay. Differentiating between the types of disabilities became more complicated than I had anticipated; I originally thought it would be clear about how to identify families, but the distinction between groups was a blurred continuum. Nina, for example, disagreed with the differential diagnosis of their child, while Phoebe and Deborah were certain of an autism spectrum disorder diagnosis, but an official label has never been given. The above categorization represents the manner in which I thought it best to classify families and this grouping corresponded as close as possible with parents' own beliefs about the label for their child.

Interviews. I invited parents to participate in individual semistructured interviews. Stuart and Francine were married and they were interviewed separately in order to allow each parent a chance to speak. Trute (1995; 2005) has found gender differences in mothers' and fathers' reactions to disability and recommends interviewing parents separately to facilitate a more in-depth analysis of each parent's meanings of the experience. Parent interviews took place at the convenience of participants and in locations agreed upon by both the parent and myself, such as coffee shops, family homes, and participants' places of employment.

The interview process began with an orienting interview, where the participant and I went over the study description and consent was discussed. This orienting interview typically took place over the phone. Following this initial orienting interview, a semistructured interview was conducted, as described below, lasting approximately one

hour. Follow-up questions were asked of the participant, using e-mail and telephone conversations.

The semistructured interviews blended more and less structured questions (Merriam, 2002) and each interview involved three segments. I first asked families to describe their son or daughter and to tell a favourite family experience. The goal of this first segment was to establish rapport with the parent while allowing the participant to discuss either strengths or weaknesses of their child and how these characteristics have affected the family. The second part of the interview focused on the process of learning the child's diagnosis; I asked the parent to chronicle the diagnostic process, including their initial concerns about their child's development; recalling any emotional experiences related to receiving or failing to receive a differential diagnosis; and asking parents to assess any changes in their thoughts or feelings between the initial suspicion of a developmental disability and a differential diagnosis. For families who had failed to receive a differential diagnosis for their child, questions were adapted to reflect this. Before concluding the interview, I asked parents to discuss their child's developmental progress since the diagnosis or lack of diagnosis, including any experiences that have helped or hindered themselves and/or the family. This question provided an opportunity for families to make recommendations for making the diagnostic process better for families.

Consent and Ethical Issues

I received informed consent from all participants and I also revisited consent throughout the research process. I employed "process consenting" (Munhall, 1989), which allows the researcher and participant to assess consent throughout the study. This

provides more protection and freedom of choice for participants since unforeseen issues inevitably arise and risk can never fully be anticipated. This is especially true when dealing with sensitive issues. Such an approach to consent allows the participant the freedom to withdraw from the interview or study at any time and ensures that the participant has a say in confidentiality throughout the research process.

Absolute confidentiality cannot be guaranteed, but I took reasonable measures of ensuring privacy throughout the study. I changed participant names and any other identifying information for data presentation, plus interview transcripts and recordings were kept in a locked filing cabinet to ensure safety of the information. I also invited participants to read the transcripts to ensure accuracy. Since parents were consulted throughout the study, they were also able to make changes as needed to ensure confidentiality to the best extent possible.

Analysis of Interview Data

I digitally recorded all interviews and transcribed the contents verbatim. I reviewed digital recordings and transcripts several times to familiarize myself with the content of the interviews, plus listened to the recordings carefully in order to enhance the written word by the nuance and tone conveyed on the recordings (Heneghan, Mercer, & Deleone, 2004). Because I was both the researcher and interviewer, I had more insight and in-context knowledge and was thus able to establish a variety of important links between the research questions and the data gathered (Litosseliti, 2003).

After careful transcription and thought, I conducted a thematic analysis on the interview transcripts (Merriam, 1998) by reviewing transcripts, making notes and comments throughout. A codebook was kept, including a detailed description of the code,

inclusion and exclusion criteria, and exemplars of real text for each theme (MacQueen, McLellan, Kay, & Milstein, 1998).

I followed Merriam's levels of analysis, beginning with a descriptive account, followed by category construction, then making inferences. A descriptive account is a narrative that conveys the meaning the researcher has derived from studying the topic of interest. The second level of analysis is the construction of categories or themes. However, before themes may be created, codes must be organized as a way to organize the data. According to Merriam, "category construction *is* data analysis" (p. 180), supporting Miles and Huberman's (1994) statement that "coding is analysis" (p. 56). Ryan and Bernard (2003) recommend looking for repetitions in the data, as well as looking for transitions and linguistic connectors. In speech, pauses and changes in voice tone may indicate a transition and connectors may indicate a new theme. I also compared and contrasted participants' responses in order to uncover themes. DeSantis and Ugarizza (2000) make the important distinction between the terms categories, factors, variables, and themes, critiquing much qualitative research that confuses these terms, thus compromising methodological rigor. DeSantis and Ugarizza define a theme as "an abstract entity that brings meaning and identity to a recurrent experience... A theme captures and unifies the nature or basis of the experience into a meaningful whole" (p. 362). Once themes emerged from the data, inferences were made and I summarized the data. A summary of the data now follows.

Findings

Parents sought a diagnosis for many reasons and spoke about the meaning a differential had for them. Five themes emerged from the interviews. These themes

included the importance of "knowing" and having a name; knowing the cause for the disability; understanding future expectations; obtaining knowledge regarding appropriate interventions; and gaining access to funding or specific services. Each of these themes will be discussed in turn.

"It was something you have to do"- The Importance of "Knowing" and Having a Name

Many families could not articulate why they needed to "know," yet they felt the pressing desire to have a differential diagnosis for their child; the search was described as simply something you "have to do." Parents discussed the need to receive a diagnosis because their child was ill, they knew something was wrong, or they felt that having a name would make it easier to explain their child's behaviour to others.

Eleven of the parents reported knowing that something was "going on" with their child and were concerned because their son or daughter was missing developmental milestones. Some parents discussed simply having a sense that something wasn't "quite right." As Tom the father of a child with a rare genetic disorder recalled, "she just wasn't growing and I remember [my wife] was so concerned about… When she missed a certain number of milestones, our concern grew and grew and grew."

Theresa, the mother a son with an unidentified developmental delay remembers suspecting for a long time that her son had some presenting issues, but "it wasn't until he was probably 2 or more that we knew what it was. As a mother, I think I always knew something was wrong, for some reason right off the bat, I just sort of knew, I just sort of had a feeling." Many parents shared this sentiment and almost all families discussed the suspicions they had regarding their child's development. Shannon, the mother of a son with Angelman syndrome, remembers:

> I just knew there was something wrong because he wasn't progressing like other children. At 5 months old he still was unable to hold his head, he could not focus his eyes, he cried a lot, and he also had feeding issues.

Although parents suspected there was something going on with their child, doctors told seven of the participants that nothing was wrong or they were advised to be patient, that their child would catch up on their milestones. They felt this type of advice lead them to doubt their abilities as parents or to question their instincts. Consequently, some parents felt that a diagnosis would validate their concerns. As Judy, the mother of a daughter with unidentified developmental delay recounted, "So, I quickly got busy and thought this is stupid, I'm not wrong, it's not me! I know this. So then I finally got myself a pediatrician and we started the saga."

When doctors finally began to listen or started to become concerned themselves, resulting in a differential diagnosis, five parents reported relief because it substantiated their anxiety. A diagnosis also allowed them to explain to family and friends why their child acted differently than other children. As two mothers of children with autism spectrum disorders remember, "Her siblings kept saying, "What's wrong with her?!" (Farah); "a diagnosis meant justification that his odd behaviour was for a reason... It just made it easier to explain his behaviour" (Collette). Having a name also made it easier for parents to explain their child's medical vulnerabilities when they had to go the Emergency room, which was quite a common occurrence for many families. Kathy reported being able to say, "this is what is the matter," which facilitated getting the proper medical care for her child in a more expedient fashion. Being able to provide a

name, parents felt, would also enable understanding regarding the etiology or cause of the disability, which is the next theme.

"Of course you think, 'It's all my fault'"- Knowing a Cause for the Disability

One of the main reasons families sought a diagnosis was to know what caused their children's behavioural difficulties or delays. Recognizing a cause or etiology was important for parents because of the risk to future children and because without knowing the reason for the disability, nine of the 14 parents wondered if they had done something to cause their child's delays or unique conduct. Thus, receiving a diagnosis helped to reduce feelings of guilt or parental blame.

The risk to future children was important for many parents if they were planning on having another child and two parents sought a differential diagnosis when they found out they were pregnant again. Judy, the mother of a daughter with an unidentified delay remembers, "[the doctor] really had no answers apart from the fact that no, it's not genetic, which was good because I was pregnant again… that was the only reason for the relief." Theresa, another mother of a son with unidentified developmental delay recounts:

> because we were thinking at that time of having another child and we were seriously thinking of not having another child if there was a chance that the child would be mentally challenged like Curtis is. And we went to [the doctor] and he said "no, nothing congenital, just a freaky thing that happened."

It is important to note that none of the families who participated in the study had children whose disabilities were described as congenital, so there were no feelings of guilt or resentment toward family members of spouses. The term, "congenital" will be discussed in the conclusion section of this paper. Parents also talked about the fear that

they had done something during pregnancy or after the child was born to cause the disability and no family reported such parental treatment factors to be the cause of the disability.

Knowing the cause therefore alleviated a lot of blame for families, being informed that their child's disabilities were not due to something the mother had done during pregnancy or a failure of the family to provide sufficient or appropriate stimulation after birth. Deborah was concerned about her son's delays and was worried that it was because he had fallen off his change table when he was an infant. Phoebe, Shannon, and Theresa reported being concerned because they had had colds or other types of infections during pregnancy and worried if that was what instigated their child's behaviours. Mothers were reassured to find out from medical professionals that this was not the case. Tom's wife, Diane, had cancer and worried that this was what caused her daughter's disabilities, but when it was revealed that the disability was a chromosomal abnormality, the family was relieved. Diane died shortly after her daughter received a differential diagnosis, but Tom recalls:

> She, we wondered whether the cancer was implicated in this, the doctors, well most doctors, said it had nothing to do with it....she really took it on herself that she had done something. I mean she didn't - she never had a drink, she didn't smoke or anything like that. It was also important for her to know if her cancer was implicated and it wasn't- that was what Diane wanted to hear.

A differential diagnosis also provided information for families about characteristic behaviours associated with the disability, which alleviated blame because some parents felt that it was perhaps the home environment that had caused their child's

behavioural differences. As Tom, the father of a daughter with a chromosomal abnormality recounts:

> She was extremely sociable with adults. I naturally assumed that this was because I was older when she was born so that my friends didn't have little friends running around. It was my second marriage, so she didn't have brothers and sisters. So I assumed this was because she just didn't have experience with children her age. And that turned out to be one of the characteristics of some children with Williams syndrome, which blew me away- you wouldn't think that that would be genetic in any way. It actually took a lot of guilt from me because I was blaming myself for this problem.

Although Tom's daughter does not have Williams syndrome but has a deletion on the same chromosome, doctors told him that his daughter's features were very similar. Knowing that individuals with Williams syndrome are often sociable with adults alleviated his feelings of responsibility for her disability due to his advanced age as a father. This benefit to differential diagnosis brings us to the next theme, which is the information provided regarding future expectations.

"What you might expect down the road"- Obtaining Information Regarding Future Expectations

Parents also reported seeking a differential diagnosis in order to receive information about what to expect for their child. Stuart, Tom, Theresa, Kathy, Gillian, Francine, Shannon, Nina, Judy, and Deborah all wanted information about life expectancy, medical vulnerabilities, and behaviours typical of a specific disability. Kathy,

the mother of a daughter with a rare chromosomal abnormality summarizes this meaning nicely:

> To find out what syndrome she has would give you your viewpoint on their life, like, what you have to look forward to, like this they could have, or that they could have, so you know what's gonna come ahead of you, what you have to deal with. It's the unknown that's very difficult because you're going day by day, like, you don't know if um… if you're Down syndrome, this can happen, that can happen. If you're this syndrome, this can happen, this happen. Without a definite knowledge of what she has, you're flying day by day. And you don't know the outcome, you don't know, like, her life expectancy. You don't know that, well, by the time she's teenager she's, you know, not going to be able to do this or that, or whatever. So, it's a total unknown.

Medical vulnerabilities were a major concern for parents. Many differential diagnoses, especially genetic syndromes, are associated with specific health issues and knowing these susceptibilities allows families to be prepared. As Stuart, the father of a daughter with Angelman syndrome discussed, "But on the upside, you know Down syndrome life expectancy is lowered because of cardiac problems, and Michaela will have a normal life expectancy. Here are the things you can expect- you can expect seizures, severe cognitive delays, and she won't talk." When Michaela was first diagnosed, she had not had any seizures but the family knew that this was quite common. Because of this advance knowledge, when she had her first seizure at 3 years of age, the family was prepared.

A differential diagnosis also allowed families to organize for the future. As Stuart remembers, "That part of it was the hardest part of it. Trying to figure out what her future was going to be." Kathy, Francine, Stuart, Deborah, Theresa, Tom, and Shannon were all concerned about what will happen to their child when they are gone, such as who will take care of them and if the child would require out of home care. However, a differential diagnosis gave families information about life expectancy and level of functioning so that they could arrange for such events. Theresa and Deborah were not successful in receiving a differential diagnosis, but one of the reasons for seeking such a diagnosis was this expectancy information.

It is significant to point out that this is where some parents differed in their desires. For Theresa, Collette, Ingrid, Gillian, and Deborah, there was some comfort in knowing about future expectations. However, Shannon, Kathy, and Francine wanted the differential diagnosis but did not want to know specifics about the future. As Kathy, the mother of a daughter with a rare chromosomal abnormality recounts, "not knowing is kind of nice because then you don't have this time date stuck in you saying, 'OK, by this time they deteriorate.'" Francine, Shannon, Stuart, and Gillian discussed not wanting to become involved in parent-professional groups because they did not want to associate their child with "that population" and were fearful to see older children with the same label. Francine, the mother of a daughter with Angelman syndrome remembers, "For a long time I didn't even look up anything about Angelman syndrome because I didn't want to be associated with that group of people." Having information about future prognosis was quite frightening for many families and not knowing was a bit of a relief.

Although Shannon, Kathy, and Francine were content with not knowing predictive information, the rest of the parents sought this kind of knowledge. Prognostic information allows families to know about future expectations and thus what kinds of supports or interventions are going to be required:

> There are some people who never know what's wrong with their child and I think that knowing what's wrong at least give us a hint that hey OK, here are some typical problems that Angelman children run into. We should be aware of that so that we can be proactive in dealing with those problems. I think that's really important and I am glad we got the diagnosis for that reason. (Stuart, father of daughter with Angelman syndrome)

Such information regarding future expectations also allowed families to be proactive in accessing early intervention programs or targeted treatments to maximize their child's learning, which is discussed in the next theme.

"How do I teach her?"- Obtaining Information Regarding Interventions

One of the main reasons parents wanted a differential diagnosis was to find out what types of interventions would be appropriate for their child. A differential diagnosis also made remediation more urgent and would provide pertinent information regarding the specific strengths and weaknesses of the child, so as to select the most appropriate form of remediation. Parents ultimately wanted to do whatever they could to help their child function in society and learn important skills. As Francine remembers, "We were fortunate in a way because we got the diagnosis early because we knew we couldn't push her too much, but at that same time, we knew we needed to do certain things to make sure she was active and cognitively engaged." Obtaining a differential diagnosis was thus

helpful in knowing the child's possible capabilities and limitations so as to tailor the supports appropriate to the child's skill level and potential.

A differential diagnosis could also provide parents with information on how to implement a behavioural program at home so that they could support their child and alleviate some of the developmental gaps and delays that might have been apparent. Judy, the mother a daughter with unidentified developmental delay remembers,

> I kept thinking of how is she going to manage, because that speaks to function as opposed to this diagnosis. The label didn't mean that much to me. Because this diagnosis, I mean developmental delay, what does that mean? I was searching more for that functional data, you know, how do I teach her to function?

At the beginning, Judy was seeking a diagnosis for her child, but after several years of testing which never resulted in a differential diagnosis, all she wanted was information about how to help her daughter. She went on to say:

> Finally I said, that's enough you know, none of this running back and forth because we had the [occupational therapist], physio[therapist], psychologist, doctors, speech therapist, whole gamut. Finally I thought about this and I thought, "You know, never mind what it is what she's got, what it's called, or whatever, we just have to get on with raising this child!" … you have to get on with this, forget about trying to fix her, how do we teach her?

This last story brings us to the final theme regarding why parents sought a differential diagnosis. Specific diagnoses are often associated with specialized intervention programs and differential diagnoses are required in order to access services and funding.

Consequently, many families sought a differential diagnosis in order to be eligible for money or restricted programs.

"The label was much more important for funding purposes than it was for anything else" - Gaining Access to Funding or Specific Services

One of the most significant reasons parents initiated the diagnostic process was the perceived need for a label in order to receive funding dollars. With funding comes access to services and many families searched for a differential diagnosis in order to obtain government money, as well as admission to specific programs, or eligibility for supports in the school system.

For Phoebe, Collette, Shannon, Farah, Ingrid, and Deborah, a differential diagnosis was sought in order to be eligible for provincial funding. Collette, the mother of two sons with autism spectrum disorders, asserts, "It just meant continued funding for schooling to us." Kathy, who has a daughter with a rare chromosomal abnormality, echoed Collette's reasons, "Yeah, you know for her [*government assistance*], and she is also supported with [*provincial funding body*] so she needed [a diagnosis] for that. Also for the school system, she needed [a diagnosis] for her aide, without that, there wouldn't have been any funding."

Farah, the mother of a daughter with an autism spectrum disorder recalls seeking a differential diagnosis for financial support and to receive help in the school system:

> My efforts finally landed my daughter funding for 2 years and with the extra assistance, [she] began to develop and use pragmatic communication at school. At this time, I think the extra individual interaction helped her become more aware of who she was and what was going on around her. Having a definite

> diagnosis worked in our favour at this time because I was able to then access a psychiatrist who came into the school to educate the teachers and in turn, the teachers made the students aware of my daughter's autism.

Thus, a differential diagnosis was important in order to gain access to services in the school system. A label was also imperative for eligibility for specialized intervention programs as discussed by Judy: "when I started, in order to qualify for the [*specific intervention program*], we needed to have that diagnosis, medical model stuff, so you have to have a diagnosis."

Eligibility for specialized programs is where parents' experiences became very interesting and almost all parents had different stories regarding the need for a differential diagnosis to access services, or if a nonspecific diagnosis was sufficient. Stuart and Francine, parents of Michaela who has Angelman syndrome, felt the need for an additional autism diagnosis, even though their daughter already had a differential diagnosis. Stuart recalls, "everyone kept saying, try to get her diagnosed as having autism because she will get wonderful therapy that way."

Deborah, the mother of a son with an undiagnosed autism spectrum disorder, recalls that funding bodies "wanted a label" in order to qualify for government dollars and admission to certain assistance programs. Her son meets almost all of the DSM-IV criteria for autism, but her son's official diagnosis is a severe communication deficit. Deborah discussed how the difference between a diagnosis of autism and one of severe communication deficit affected access to services and she has "used the label [of autism] because 'it's good enough for them.'" For Deborah and her son, a formal diagnosis of autism was not required in order receive the supports they desired, but Shannon and

Theresa reported being unsuccessful in qualifying their child for restricted intervention programs due to their child's differential diagnosis or lack thereof. Theresa and Deborah reported having to lie or really fight to access these services.

Parents thus seek a differential diagnosis for many reasons. Receiving a formal diagnosis for the disabilities of one's child reduces ambiguity regarding cause and future expectations, provides information regarding appropriate interventions, and allows access to funding or disability services. These themes will be elaborated below, with reference to Patterson's (1988, 1989) FAAR model. The article concludes with recommendations for future practice and research.

Conclusion

Medical practitioners are key to providing information regarding the etiology, prognosis, and recommended interventions for the differential diagnosis. Such information allows the family members to reach a "shared definition" (Patterson, 1989, p. 113), an unambiguous understanding of the disability, which in turn may accelerate their ability to adapt to the diagnosis. On the basis of the FAAR, Patterson predicts that when a family faces the stress of a major illness or disability in one of its members, the family moves into a crisis. From the perspective of the FAAR model, the family's recovery from crisis could be facilitated by the way medical professionals relate to the family. Patterson asserts that there are several things that might be helpful, but most relevant to the present study, she states that medical practitioners can help families by reducing the ambiguity about what is happening, resulting in less family strain.

Reaching a "shared definition" appears to be what families are seeking when trying to find a name for the disability of their child, which crosses several of the themes

discussed by parents. A shared definition means that the diagnosis is free from uncertainty and there is information provided regarding the condition, such as etiology, treatment, and prognosis. Having a name for the differential diagnosis provided the parents in this study with information regarding future expectations, a finding supported by Burden (1991) and Poelmann et al. (2005). There is always the hope that diagnosis will yield a finding pointing toward something useful, such as how best to provide supports for their children and how to access specialized services. Knowledge regarding the future appears to give parents some sense of control (Knox et al., 2000) and parents need to feel like they are doing everything possible to help their child (Burden). Patterson (1989) asserts that a sense of control is crucial for family adaptation. Even though the information provided may be incorrect, even the illusion of having some information can make planning easier and give parents a sense of greater control.

It is important to note that the meaning behind a differential diagnosis was the same for families with and without a specific diagnosis for their child. Regardless of their level of success with receiving a diagnosis, parents' reasons behind the search were similar; however, there were some interesting differences between the different types of disabilities. Parents with autism spectrum disorders or suspected autism spectrum disorders seemed to seek access to interventions and funding support, while parents of children with more severe disabilities seemed to focus more on the future expectations and medical issues.

A lack of differential diagnosis can aggravate or complicate other problems. For example, the parent may feel that there is something wrong with medicating or treating their child in a particular way, but it is harder to argue effectively when the diagnosis is

unclear. The treatment issue needs to be resolved, but it is hard to determine until the diagnostic foundation can be trusted. Families who did not have a differential diagnosis, Theresa, Deborah, Barb, Judy, and Phoebe, felt frustrated with not knowing the "best" way to provide intervention for their child.

The need for a differential diagnosis also became very important for accessing specific services and parents sought a label for their child for this reason, some with greater success than others. Shannon, for example, has a son with Angelman syndrome, but there are no specialized treatments for this differential diagnosis; she sought access to autism services, but without an official autism spectrum disorder, her son did not qualify for the programs. Theresa has a son with a nonspecific disability and this mother discussed the difficulties in gaining access to intervention because her son did not meet the specific program criteria. Certain diagnoses are associated with differential funding or intervention programs and it makes sense to have specialized programs tailored to specific developmental needs, but when someone has similar presenting issues, it seems unfair to restrict someone from service because medical professionals are unable to identify a specific gene that causes their disability or apply a formal label. The same is true for individuals who have a differential diagnosis but the diagnosis is so rare that there are no specific interventions, as described by Shannon above. As this study demonstrated, parents are seeking a diagnosis in order to access the best treatment for their child, but many were unable to do so because of how funding bodies are set up (Burden, 1991; Gillman et al., 2000).

One theme deserves significant mention and that is the issue of a differential diagnosis relieving guilt. Several families reported the relief they felt when they

determined the specific diagnosis because this often lead to knowledge regarding the etiology and cause of the disability. These findings are similar to Burden's (1991) study, where mothers of children without a differential diagnosis had more difficulty working through their feelings of anger and guilt than mothers of children with a specific diagnosis. However, as with Burden's study, all parents interviewed described causes that were not congenital or the result of something they had done. There appears to be some confusion about the word, "congenital" and the causes of disabilities. Six parents from five families participated in the study whose children had genetic disorders, and there is the possibility that the disabilities were inherited from one of the parents. Angelman syndrome, for example, is commonly caused by a deletion on the maternal copy of the 15^{th} chromosome, but can be due to four other causes. Parents did not report if medical professionals went over this information or it was deemed relevant. Furthermore, Theresa and Deborah reported that their medical practitioners assured them that their child's disabilities were not genetic, a concern of the parents because they were planning for another child. However, the children of both these parents have unknown disabilities and there is always the possibility that an unknown genetic condition caused the child's disabilities. Parents' experiences would of course be different if medical practitioners had informed them that their child's disabilities were due to maternal drinking, was inherited from one of the parents, or was the result of some type of family environmental cause. Future research should look at the experiences of families of children with Fetal Alcohol Spectrum disorder or inherited disabilities such as Fragile x syndrome, to see if meanings are the same. Furthermore, interviewing the medical practitioners would be beneficial to see what information regarding genetics is actually given to parents.

The present study addressed the meaning behind a differential diagnosis and investigated the experiences of parents of children with and without specific labels. Only 14 parents were interviewed and future research needs interview more parents and to look more in depth at the reasons why parents seek a differential diagnosis. Future research should also look further into what the meanings are for different types of diagnoses, such as autism spectrum disorders versus more severe disabilities associated with more medical vulnerabilities. Further, Keogh et al. (2000) state that we know little about the families of children with nonspecific disabilities, yet for 30 - 40% of individuals with developmental disabilities, the specific cause is unknown (AAMR, 2002). The current study is a start, but more information is required about these families. The word, "family" is also important to discuss. I attempted to include all family members when recruiting participants, but only parents participated in this study, and participants were mostly mothers. Interviewing all family members, including the child with the developmental disability, would be important to see if all family members have the same reasons for seeking a diagnosis.

It appears that parents are seeking a differential diagnosis for their child for several reasons. Parents express the need to "know" in order to alleviate guilt and what to expect about their child's future, they want to know what caused the disability, they desire knowledge regarding appropriate interventions, and they seek a label in order to access funding or specific services. It is important for medical practitioners to know what parents desire when beginning the diagnostic process so that they can provide relevant information to families regarding the disabilities. As reported elsewhere (Poelmann et al., 2005; Watson, manuscript in preparation), parents feel medical professionals provide too

much pessimistic information about disabilities and not enough information about intervention. By providing the desired information, this will facilitate greater family adaptation to the disability and the best supports for the child.

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CHAPTER 5: CONCLUSION

This was a study of the parental experience of the diagnostic process when a child has a developmental disability. This chapter presents a summary of each of the papers of the dissertation, then applies the Family Adjustment and Adaptation Response (FAAR; Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) model to the present study. Strengths of the study are then discussed, followed by recommendations for future research and practice. Finally a conclusion brings theory and findings together.

Summary of Research Findings

This study addressed the parental experience of the diagnostic process when a child has a developmental disability. Findings were presented in one literature review and two research papers. Each of these papers will be reviewed below.

This dissertation began with a literature review outlining the main issues around differential diagnosis and the diagnostic process. A brief overview of family research in general discussed the history of research on families of children with disabilities and how most studies have not specified the differential diagnoses of the children, nor considered the diagnostic process as contributing to family adaptation. The debates for and against differential diagnosis and labels were then presented, concluding that labels can be beneficial, but have the potential to do harm as well. The debated benefits include providing access to treatment and resources, raised awareness, reducing ambiguities, providing comfort to families, and creating a sense of identity, but labels can also contribute to exclusion from society, lowered expectations, and even when a label is given, it may not give details regarding how to provide intervention. The limited research on the diagnostic process was then cited, highlighting parents' dissatisfaction with the

process and medical professionals' lack of knowledge regarding many differential diagnoses and their minimal training in providing information regarding disabilities. The literature review concluded with a critique of the literature, highlighting the need for more qualitative approaches to family research and a more in-depth look at family experience of the diagnostic process. It is hoped that the two research papers that followed addressed these critiques.

The first research paper, "Parents' Experiences Dealing with Professionals when Seeking a Differential Diagnosis" uncovered six main themes. The first theme was *tolerating intensive testing*, and parents detailed the invasive procedures and number of medical professionals encountered throughout the diagnostic process. When parents talked about the second theme, *sensing rigidity in thinking by professionals*, they recalled medical professionals not listening to their initial concerns about their child's development and parents felt they had to fight to initiate the diagnostic process. The third theme, *perceiving pessimistic prognostic information*, involved parents' discussion of the information they received when and if a differential diagnosis was given. Parents reported receiving pessimistic information about their child's future functioning and prognostic information that for many parents was not deemed relevant at the time. The fourth theme was *receiving multiple diagnoses*, and parents detailed an "emotional roller coaster", being given a tentative diagnosis or speculative diagnosis, only to receive a different diagnosis at the following medical visit. Parents also discussed *feeling dissatisfaction with the information provided*, which was the fifth theme. Here, mothers and fathers expressed disappointment with the amount of knowledge medical professionals had about the differential diagnoses and about appropriate interventions,

when this was the type of information they were seeking. The first five themes were thus quite negative, but the sixth theme, entitled *encountering a range of professionals*, spoke to some of the positive experiences parents had with medical professionals. Parents recalled medical professionals trying to be helpful when providing prognostic information and one father, Tom, commented on the quality of the medical team and their "medical minds."

The second research paper, " Why do Parents Seek a Differential Diagnosis? A Search for Meaning and Support," discussed the reasons behind the quest for a diagnosis. Five themes were discussed by parents including the importance of "knowing" and having a name for the disability; knowing the cause for the disability; understanding future expectations; obtaining knowledge regarding appropriate interventions; and gaining access to funding or specific services. The *importance of "knowing*" theme included parents' need for a name in order to explain their child's behaviour to friends and family, as well as feeling relief that their concerns about their child's development were validated. In the second theme, *knowing the cause for the disability*, parents discussed the fear of having another child with a disability and the alleviation of guilt that they did not do something to cause their child's developmental delays. *Understanding future expectations* was the third theme, and parents detailed their desires for prognostic information, such as medical vulnerabilities, so that they could plan for the future. Preparing for the future was also important in the fourth theme, *obtaining knowledge regarding appropriate interventions*, and here parents discussed seeking information about the best education and remediation for their child's developmental needs. In the final theme, *gaining access to funding or specific services*, mothers and fathers talked

about the need for a differential diagnosis in order qualify for specialized intervention programs and to receive funding dollars.

A Return to the FAAR Model

> The stories families tell incorporate cognitive factors that go beyond the definition the family gives to the stressor (that is, the onset of the disability) as families search for meaning in a life that, in many ways, has been shattered by the presence of added demands, multiple losses, changed routines, roles, and expectations. (Patterson & Garwick, 1998, p. 73)

Developmental disability and family functioning interact continually in a pattern over time. Families "go through repeated cycles of adjustment-crisis-adaptation" (Patterson & Garwick, 1994, p. 132), seeking to achieve balance, a state that is influenced by the meanings families attribute to their situation, as well as to their own identity as a family and to their view of the world (Patterson & Garwick, 1994). The family attempts to maintain balanced functioning by using its resources to meets its demands, including stressors and strains. According to Patterson and Garwick (1994), "the meanings the family ascribes to what is happening to them (demands) and to what they have for dealing with it (capabilities) are critical factors in achieving balanced functioning" (p. 132). Such is the premise behind the FAAR (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) model, which will be reviewed below and applied to the present study on family experience of seeking a differential diagnosis.

The FAAR model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) addresses how families deal with potentially stressful situations. Several variations of the model have been developed, but the original model evolved from the ABCX framework

(McCubbin & Patterson, 1987) in an effort to describe the process by which families achieve adjustment and adaptation (Patterson, 1988). According to the FAAR, the family is a social system and thus attempts to maintain a balance by using its capabilities to meet its demands. There are two phases in the FAAR, adjustment and adaptation, which are separated by a crisis. During the adjustment phase, only minor changes are possible as the family attempts to meet its demands with existing capabilities. However, a crisis emerges, where the demands exceed the family's capabilities and the imbalance persists (Patterson, 1988). After the crisis or several crises, families enter the adaptation phase, where they may acquire new resources, deduce the demands they must deal with, and/or change the way they view a situation (Patterson, 1988). The FAAR highlights three important contributing factors to adjustment and adaptation: Demands, Meanings, and Resources. Reaction to a potentially stressful situation follows one of two spirals, and adaptation is seen along a continuum from bonadaptation to maladaptation. Figure 1 presents an overview of the FAAR model.

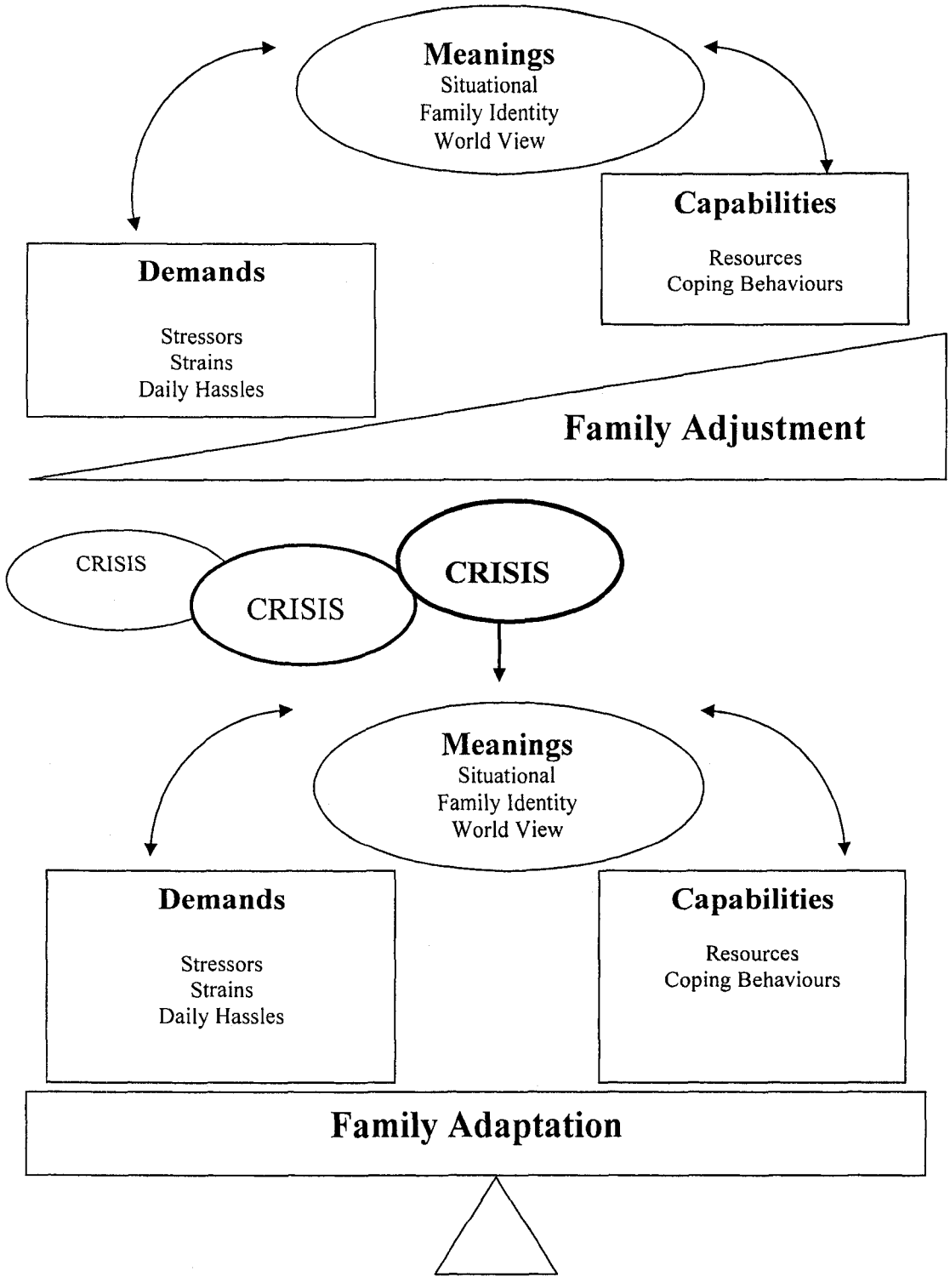

Figure 1: On overview of the Family Adjustment and Adaptation Response (FAAR) model.

Demands. According to the FAAR (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998), demands are conditions that call for a change in the family system. Three kinds of demands exist, including stressors, strains, and daily hassles (Patterson & Garwick, 1994, 1998). Stressors are life events that occur at a discrete period of time and that produce change. Strains, on the other hand, are conditions of felt tension associated with a need or desire to change something. Daily hassles are minor irritants that we encounter throughout our day-to-day functioning (Lazarus & Folkman, 1984), such as waiting in traffic. Parents of children with disabilities face all three kinds of demands, but strains and stresses appear to be the most relevant demands to the diagnostic process. Furthermore, according to the FAAR, demands can come from three sources, including individual family members, the family itself, and the larger community.

Applied to the present study on seeking a differential diagnosis, stressors might be receipt of the actual differential diagnosis or the first suspicion of a disability. Strains would be the pile-up of demands, such as dealing with multiple medical professionals and receiving incorrect diagnoses, whose source is the larger community. Another strain would be the perceived need for a differential diagnosis, which is influenced by the individual family members, but also by the larger community, since the perceived need for a differential diagnosis often comes from a need for a label for funding or access to services.

Disability itself places a chronic set of extra demands on the family system (Patterson & Garwick, 1994). The exact nature of these demands varies depending on the condition. Such variances would be due to specific sensory, motor, or cognitive effects of the specific disability; the level of visibility of the condition; the prognosis or life

expectancy associated with the diagnosis; whether the course of the illness is constant or progressive; the amount of home treatment and the expertise needed to provide it; and the amount of pain or other symptoms associated with the condition (Patterson & Garwick, 1994). The FAAR (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) highlights different sources of stresses and strains and as discussed above, the differential diagnosis itself can contribute to strains, such as medical vulnerabilities, medication issues, challenging behaviours, and prognosis, but the focus of this study is on the diagnostic process. The largest sources of strain reported by parents were the associated strains of waiting for differential diagnosis and the desire/need for diagnosis.

Parents who were successful in attaining a differential diagnosis reported less demands than families who were less successful, since this process lasted less time. Families of children with differential diagnoses faced less uncertainty and the visits to medical practitioners decreased after a differential diagnosis was received; however, for families who were unable to receive a diagnosis, demands were unresolved and thus pile-up occurred. Patterson and Garwick (1994) stress that the nonoccurrence of an event can be more stressful than if the event occurred. Therefore, if parents do not receive a differential diagnosis for their child, this can be more taxing, a finding emphasized by parents in my study. When there is considerable uncertainty about the prognosis, this increases stress for the family because they are less sure about how to plan for the future (Burden, 1991; Patterson & Garwick).

Furthermore, parents of children with autism spectrum disorders appeared to experience different kinds of demands than children with identified genetic disorders. When an autism spectrum disorder or behavioural disorder was suspected, parents

discussed the overreliance on checklists and felt that medical professionals were not performing a thorough observation of the child. Parents of children with genetic disorders, however, were more concerned with the invasive medical procedures. Such differences indicate that there are unique experiences that accompany the differential diagnosis, and these variations must be considered when looking at family supports.

Situational and global meanings. The FAAR also singles out two important factors in appraisal, situational meanings and global meanings (Patterson, 1989) and each of these meaning levels are interrelated (Patterson, 1993). Situational meanings refer to specific appraisals of challenges and resources while global meanings refer to a more general set of cognitive beliefs about family, community, and life in general (Patterson, 1989). Global meanings can be further divided into family identity and family worldview (Patterson & Garwick, 1994, 1998). Family identity addresses how the family views itself, including relationships, roles, and norms, while family worldview integrates Antonovsky's (1979, 1987) concept of coherence and addresses how the family interprets reality, their core assumptions about their environment, and their existential beliefs.

Applied to the present study, situational meanings would deal with ambiguous situations, such as desiring a differential diagnosis in the hopes of knowing how to proceed with intervention or remediation, or the need to know a cause for the disability. Another example would be trying to resolve calling the doctor when first suspecting a developmental disability or trying to decide to take their child to the hospital for more diagnostic tests. This is often referred to as "social ambiguity" (Mechanic, 1974) and if families do not know what to do, they will turn to someone in community for

interpretation and meaning. Families in my study turned to medical practitioners because of their perceived expertise in diagnosing disabilities (Patterson, 1989).

The second level of meaning, family identity, deals with ambiguities regarding social placement and boundaries. Here, families may deal with boundary ambiguity, where the physical and psychological presence of family members needs to be considered. If a child is hospitalized for an extended period of time, for example, are they still considered a part of the family? Furthermore, if one parent is devoting an extended period of time at medical appointments, roles and boundaries can become blurred. Dealing with multiple medical professionals for extended periods of time may also affect the family identity, as professionals may appear to almost become part of the family.

The final level of meaning, worldview (Patterson & Garwick, 1998) deals with global meanings and it more difficult to articulate. The family worldview addresses how the family members interpret reality, their core assumptions about the world, and their existential beliefs (Patterson & Garwick). All meanings are created and maintained through social interaction (Berger & Luckmann, 1966) and a family's worldview is affected by the community in which it resides. Applied to the present study, a family's worldview would include their perceptions about disability in general and their beliefs about labelling or differential diagnosis. In our society, there is quite a strong belief that differential diagnosis means access to support and information (Gillman, Heyman, & Swain, 2000). However, some family members may not value this type of information and may wish to stay uninformed. Shannon, Kathy, and Francine, for example, desired a differential diagnosis, but did not want to know potentially frightening information about the future, such as life expectancy.

One's views on disability would also be a component of the worldview. If disability is thought of as something that should be prevented, then parents may seek a differential diagnosis as a means for remediation or prevention of disability in future offspring. However, if one has a different conception of disability, then differential diagnosis may be sought in order to gain greater understanding of the child's strengths and how best to teach the child. Although two parents discussed wanting to know about the risk to future children, each of the parents in the study sought a diagnosis in order to access information regarding how best to tailor supports for their child.

The worldview speaks to the social construction of disability. Judy makes this point nicely. When asked what meaning a differential diagnosis would provide, she responded,

> Nothing. That is such medical model stuff, when you are dealing with disability, I mean given my background, you know, of course every mother is different, but for me, given my background with disability… at the time, it didn't really mean that much to me… I kept thinking of how is she going to manage, because that speaks to function as opposed to this diagnosis. Because this diagnosis, I mean developmental disability, what does that mean?

Parents thus appear to be seeking a differential diagnosis for what it provides, not for the label itself. A comment on labels will be provided in the conclusion of this chapter.

A differential diagnosis for Tom also changed his worldview and perspective on life. When he read an article about the few cases of the chromosomal deletion his daughter, Grace, had, he was surprised at the findings and these altered his thoughts toward his daughter:

> Anyways, the article. They had again about six or seven more cases and in probably half the cases, the child was stillborn and they diagnosed the problem with an autopsy. So what that did for me, among other things, was think, "Wow, how fortunate that this little gaffer even made it into this world and sustained herself."

Although this example does not speak to the reasons for seeking a diagnosis, once the differential diagnosis was received, along with the resultant prognostic information, this altered Tom's worldview and subsequent adaptation to the disability of his child.

Both situational and global meanings are significant for the present study because they can add to or reduce stressors or strains that may occur. Specific to the diagnostic process, for a diagnosis to be effective, it must somehow fit with the family's existing set of beliefs, which is integral to their worldview (Patterson, 1989). Therefore, if a family is not comfortable with a diagnosis or does not understand the diagnosis, this will upset both their global and situational meanings, which may lead to disruption in functioning, called maladaptation. Importantly, all family members must reach a shared definition and maladaptation can occur if family members differ in their meanings. According to Patterson and Garwick (1998), "shared meanings reduce ambiguity and uncertainty about a complex array of stimuli and make coordination of response among group members possible" (p. 81). Thus, medical professionals need to become aware of why families are seeking a differential diagnosis in order to provide relevant information to parents. Communication between parents and medical practitioners is thus essential to maximize family adaptation to disability.

Resources. In the FAAR model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998), resources are competencies or traits of the family system, its members, or the larger community. These resources can be used to manage stressors and strains. Self-esteem and a sense of mastery have been named as important personal resources that facilitate adaptation. Parental control dealing with medical professionals would thus be a relevant competency applied to this study. Family resources are the quality of the marital relationship, communication skills, and shared goals, so if parents both agree with the value of a differential diagnosis, this would lead to greater family adaptation, but if there are disagreements about this process, maladaptation may occur. Finally, community resources are one of the strongest protective factors associated with family resilience (Patterson & Garwick, 1998). These resources include the quality of the relationships families have with medical professionals, as well as access to funding bodies and intervention services.

Access to specific intervention programs was a theme that many parents discussed. Francine discusses below the process by which they were successful in gaining access to an autism specific program for their daughter, Michaela, who has Angelman syndrome:

> Basically we wanted her to get into this program at [specific location]. So we basically did research and we found that some people think there is a genetic link between autism that might be related to the Angelman site on the chromosome, and we also found out that a lot of people with Angelman have a lot of similar traits to autism and there is a big crossover between the diagnoses. So we kind of watched Michaela and found a lot of things that she has done - hand waving, poor

> attention span - a lot of features of autism we could see in her. So we got her into the [hospital] to be assessed for autism. And they agree that she had some features, but they couldn't be sure because her cognitive functioning wasn't far enough along to go through the testing, but the doctor there agreed that [intensive behavioural intervention] would be the best therapy for her regardless. And so that's how we were able to get into that program. And that has been a lot of help.

When prompted about the autism diagnosis, Francine went on to say:

> That's the only thing that gets you into that program. Yeah, that used to be because of [funding body] rules, that they wouldn't fund that therapy for just anybody, but then they changed it and said that it wasn't dependent on diagnosis, but dependent on whether the child would benefit. As soon as they said that, the [funding body] decided that they only wanted to be an autism organization and fortunately we were already in the program at that point so they just said you can continue on kind of, but [a friend] was interested in getting her son into it, but she couldn't because they had already decided that.

An autism diagnosis was therefore important to Francine in order to qualify her daughter for a specific intervention program. Francine and her family thus had access to resources, a factor Patterson (1989) stresses is crucial for family adaptation. If these resources were put into place, then parents were able to adapt better, perhaps because they felt that they were doing what was best for their child. However, if these resources were unavailable, such as for Shannon and Theresa, whose children did not qualify for specific intervention programs due to their child's diagnosis or lack of differential diagnosis, more stress and possible maladaptation could occur.

Bringing it all together. The FAAR model (Patterson, 1988, 1989; Patterson & Garwick, 1994, 1998) provides a framework for how families deal with potentially stressful situations, considering the family values, their search for meaning, and their perception of stressors and strains. If resources are high, then families adapt well, but if stressors and strains overwhelm family resources, then maladaptation may occur. Using FAAR terminology applied to the present study, figure 2 provides possible outcomes for families enduring the diagnostic process. The figure demonstrates how maladaptation can occur when stressors or strains overwhelm resources; when parents are seeking a differential diagnosis, are dealing with multiple professionals, and are enduring intensive testing, these strains are greater than their resources, which would be having a differential diagnosis, gaining access to intervention information and a sense that their concerns about their child were being heard. The figure also illustrates how adaptation is possible when a family receives a differential diagnosis. Such a label becomes a resource and this is accompanied by access to information and services. These resources are greater than the strains of the diagnostic process and the family fares better.

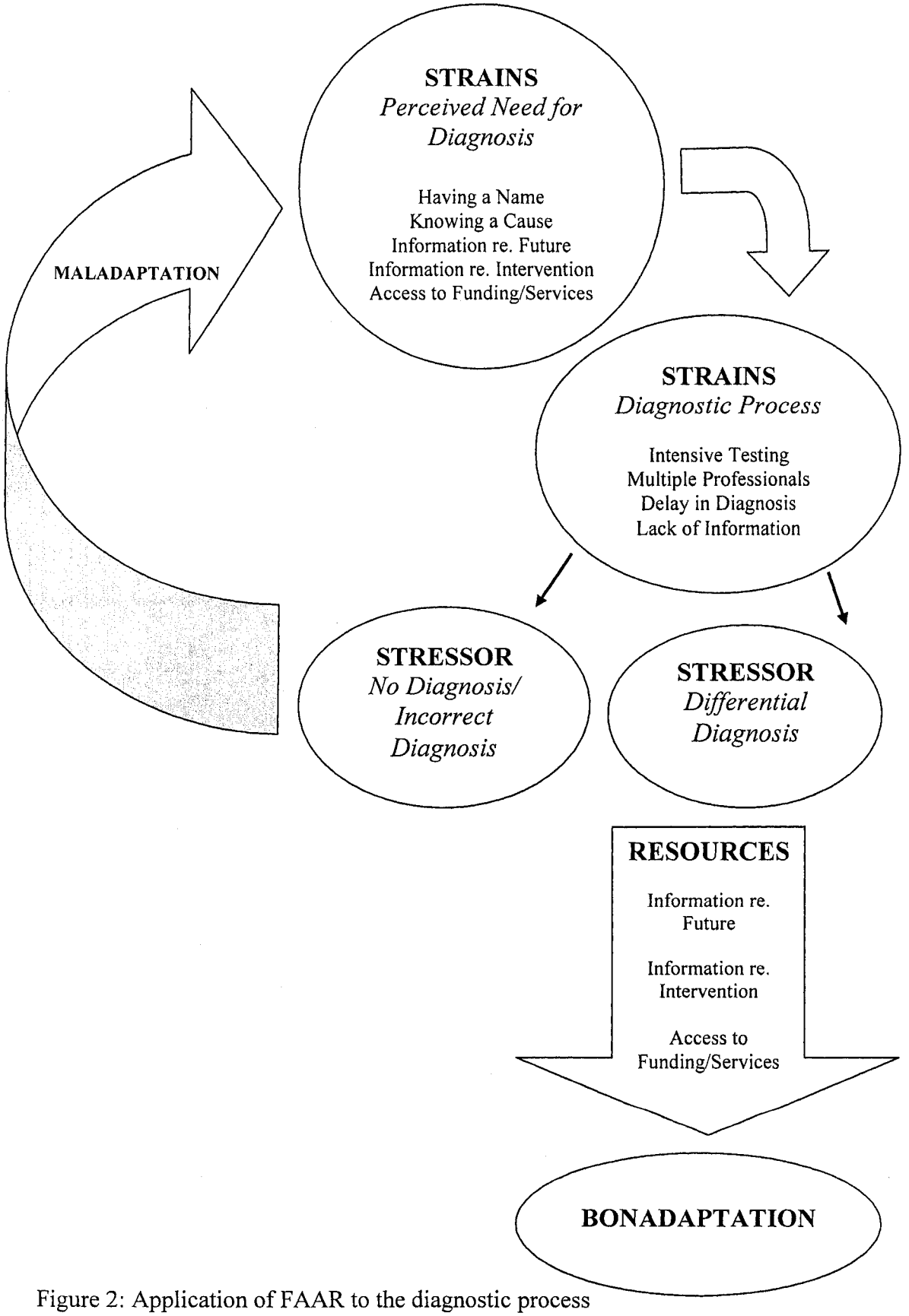

Figure 2: Application of FAAR to the diagnostic process

It is important to note that "over time, families go through repeated cycles of adjustment-crisis-adaptation" (Patterson & Garwick, 1994, p. 132), a statement that was certainly articulated by the participants in my study and is demonstrated in the above figure. Parents described an "emotional roller coaster" of hopes and desires, multiple or incorrect diagnoses, and frustrations with not feeling heard by medical professionals. They may thus continue the cycle of perceiving a need for a diagnosis, felt strains of the diagnostic process, and receiving incorrect diagnoses or failing to receive a differential diagnosis. As Francine, the mother of Michaela, who has Angelman syndrome, recalls, "It was an emotional roller coaster… some tests would rule out something bad and we would be happy, then someone else would bring up another concern they had about her and we would be upset again." Therefore, families may go through times where there is adaptation, but if they do not have sufficient resources, such as a sense of competency and control, or do not feel heard by medical practitioners, this increases their strains and can lead to maladaptation. Shannon remembers:

> I was tired of being told there was nothing wrong with him, he was colic [sic]. When you are a mother of two you know when something is wrong with your child. I just kept coming back until they agreed to do some testing.

Strengths of the Study

A number of writers (e.g., Guba & Lincoln, 1989; Lincoln, 1995; Lincoln & Guba, 1985; Merriam, 1995, 2002; Mertens & McLaughlin, 2004; Patton, 1990) have outlined criteria for judging the quality of interpretive research. Researchers also disagree on how strict criteria should be (Barbour, 2001; Woolcott, 1994), and Lincoln (1995) prefers to refer to this dialogue as "emerging criteria". However, all theorists agree that

rigor is needed in all kinds of research to insure that findings are to be trusted and believed and trustworthiness has been defined as how well a particular study does what it is designed to do (Merriam, 1995). In other words, how can a researcher persuade his or her audience that the findings of a study are "worth paying attention to?" (Lincoln & Guba, 1985, p. 290).

Patton (1990) stresses the need to utilize rigorous techniques and methods of data collection. This task may be accomplished through prolonged engagement, member checks, and progressive subjectivity.

Prolonged engagement is the investment of sufficient time in order to build trust, test for misinformation from participants, and learn about the experience in question. There are no rules to govern how long a researcher should continue to interview participants, but it is indicated that the researcher must have confidence that if the themes and examples are simply repeating and not extending, then it is time to finish interviewing (Mertens & McLaughlin, 2004). I continued the interview process with each participant until I felt all avenues had been explored and answers to questions and stories began to repeat themselves.

Member checks have also been identified as a strategy to increase credibility (Lincoln & Guba, 1985; Merriam, 2002; Mertens & McLaughlin, 2004). Member checks require the researcher to verify with the participants any constructions that are developing as a result of data collected and analyzed. In response to this recommendation, at the end of each interview, I summarized what has been said and asked the respondent if their position was accurately reflected and if they had anything to add or clarify. After the

transcripts were complete, each participant was given the opportunity to discuss the contents of the transcripts and the interpretations formed.

Progressive subjectivity has also been identified as a key strategy to increase credibility of research (Mertens & McLaughlin, 2004). This refers to the credibility of the researcher and monitoring the researcher's own developing construction (Guba & Lincoln, 1989). In interpretivist research, the researcher is the research instrument (Merriam, 1995, 2002; Patton, 1990). Nevertheless, the credibility of the researcher can be affected by bias or changes in perspective during the course of the study. To increase the credibility of the present study, information on my experiences, training, and perspective were revealed in the reflexivity section of the first chapter, including personal and professional information that may have affected data collection, analysis, or interpretation. I was also sensitive to change and recorded any thoughts and changes throughout the research process (Guba & Lincoln, 1989). Prior to engaging in any interviewing, I recorded what I expected to find in the study and continued to record my constructions throughout the research process. If I found only what I expected to find, then credibility suffers, but if my constructions change after speaking with participants, credibility is strengthened. As I discussed in my "experience with medical professionals paper", I was very surprised at the negative tone to the interviews and this finding was very unexpected for me.

Research should also provide enough description to contextualize the study such that readers will be able to determine the extent to which their situation matches the research context and hence, whether findings can be transferred (Lincoln & Guba, 1985; Merriam, 2002). To this end, thick description was provided, which is not simply a matter

of amassing detail (Schwandt, 2001). Rather, to thickly describe an experience or phenomenon is to actually begin to interpret it by recording the meanings, motivations, and circumstances that characterize a particular episode. Schwandt asserts that it is the interpretive quality of the portrayal, rather than the detail as such, that makes it thick.

In order to accurately convey the sentiments of my participants, I digitally recorded interviews to assist in the accurate recording of the conversations. If any further clarification was required, participants were asked to clarify wording or provide further explanation. I also kept an audit trail, which is a detailed account of the methods, procedures, and decision points carried out during the study (Merriam, 2002). I reported the details of data collection and analysis procedures in a notebook, which described how themes were derived, and demonstrated rival or competing themes (Patton, 1990). In this journal, I also kept any debates I had with regard to theme names, inclusion, or exclusion criteria. I also saved all previous versions of data analysis and presentation, so that I could reflect back on my decision making process. Consultation with my dissertation supervisor with regard to the themes and his thoughts as to their accuracy also added to the credibility of the research findings.

Considerations and Recommendations for Future Research

Although this basic interpretive study provided an in-depth look at parents' experiences of the diagnostic process, only 14 parents were interviewed, representing 13 families. A larger sample size would have provided more information about this experience. Furthermore, only parents were interviewed, mostly mothers, so I cannot transfer this information to all family members. Future research should look at all family

members, including extended family, and should include more fathers, whose reasons for seeking a differential diagnosis may differ from mothers' hopes.

Parents in the study had children with a range of developmental disabilities, including genetic disorders, autism spectrum disorders, and nonspecific disabilities. Including only one specific differential diagnosis would have provided a more in-depth look at the experiences of parents of children with these disorders, rather than having a range of experiences. Such a diversity of disabilities allowed for a look at commonalities between the types of disabilities, but a deeper look at one specific differential diagnosis is an important consideration for future research.

These issues deal with the matter of accessing families for research; parents of children with disabilities are very busy and it was difficult to find parents to participate in the research study. I tried to be purposeful in finding participants at the beginning of the study, but when dealing with rather rare disorders, the availability of families is quite low. Furthermore, being able to schedule time with one parent was often difficult and involved rescheduling several times, or being creative with staying in contact with parents, such as conducting some interviews over the phone, and following up over e-mail. To coordinate with both parents or extended family members would have been even more difficult.

Addressing the above considerations, future research should look at the experiences of all family members and further focus on families of children with one specific disability. As previously discussed, particular diagnoses are associated with strengths and weaknesses, as well as unique developmental patterns (Dykens & Hodapp, 2001), which ultimately influence parents and other family members. This study found

some differences between the experiences of parents of children with autism spectrum disorders and those of children with more specific medical conditions. Future research should look more in-depth at this issue and should also concentrate on the experiences of all family members, not just parents.

This study provided an in-depth look at parental experiences, but much of what parents discussed was their interactions with medical professionals. Interviewing medical practitioners about their experiences of the diagnostic process and providing differential diagnoses to families would be beneficial to see what they are experiencing. Telling a parent that a child has a developmental disability with some potentially frightening information is no doubt a stressful experience. Furthermore, this study only represented parents' views of their interactions with medical professionals and their memories may be clouded by emotions or subsequent meetings with other medical practitioners. By interviewing both parties, a greater understanding of the interactions between parents and medical professionals would be possible.

Accessing families of children with disabilities for research can be difficult, especially when dealing with unique disorders. Research with unique populations also presents ethical challenges in protecting participants' identities and confidentiality. Future research should look at more creative ways of reaching families, in order to be more accommodating to families' busy schedules. Two parents in my study talked about their websites and blogs, where they connect with other parents and present their experiences. Looking at this new medium for social support would provide greater insight into family experiences and would be less invasive to families. This type of research would also provide access to larger numbers of participants.

Recommendations for Practice

Patterson and Garwick (1994) maintain, "crisis in a family is an optimal time for clinicians to intervene, since families are particularly receptive to information and recommendations for change at these turning points" (p. 133). Furthermore, medical professionals "can play a key role in preventing unnecessary crises by proactively providing anticipatory guidance about normal development and illness-relate changes" (p. 133). As this study found, parents are seeking information about disabilities and what they can expect for their child. However, parents reported discontent with the information provided, longing for more specific knowledge about the differential diagnosis, but being given general information or older articles not reflective of current practice. Skotko and Bedia (2005) found similar findings, citing parents' anxiety about not receiving specific medical information about their child and if they did, it was often out of date. Medical professionals thus need more up-to-date information on developmental disabilities in general, as well as information regarding newer differential diagnoses such as genetic disorders and the range of autism spectrum disorders.

Communication between parents and medical professionals is also crucial to make the diagnostic process better for both parties. It is important for medical professionals to understand the information that parents are seeking and why they desire a differential diagnosis. Such information would allow medical professionals to provide relevant information to families and may contribute to improved outcomes (Patterson & Garwick, 1994). Furthermore, if medical professionals listened to parents' initial concerns regarding their child's development, this would improve relationships between families and the medical team, plus would help to expedite the diagnostic process. Conducting a

brief preliminary evaluation as soon as parents express worry would lead to parents feeling like they were listened and would also provide information to medical professionals. Listening to parents' concerns requires only a few minutes of professional time (Glascoe, 1999) and takes less time than a formal screening instrument (Glascoe; Sices, Feudtner, McLaughlin, Drotar, & Williams, 2004). Such a strategy also provides a family-focused and collaborative approach to addressing developmental disability.

A final recommendation deals with access to intervention programs and the frustration that many of the parents in this study reported. Patterson and Garwick (1994) recommend a "family-focused systemic approach to intervention, whereby services are designed to fit the needs of the family instead of fitting families into pre-existing programs without regard to their particular needs" (p. 140). Specialized programs are important for providing high quality education and intervention, but when a child requires support and does not meet the criteria for any program, then nobody is being served. Intervention programs should look at their admission criteria or more programs should be designed to meet the needs of children with rarer diagnoses.

A Comment on Labelling

As discussed in the literature review component of this dissertation, there is a large debate about differential diagnosis and if labels are beneficial. The parents in this study discussed how important labels are in order to access services and how certain labels are required to qualify for specific programs. However, few parents talked about the social construction of labels and the actual words, "developmental disability." Stuart, the father of Michaela, who has Angelman syndrome, made a very interesting point about labels, though, and how the diagnostic process affected his perception of his daughter.

Because of the somewhat lengthy process that lead to Michaela's differential diagnosis, Stuart does not allow her label to affect his perception of his daughter:

> I don't think of my child as autistic or Angelman, I think of her as Michaela. And actually, to be honest with you, I think the reason for that is because it took almost a year to be diagnosed. If, I hate to say this, but I think that if she was born and the minute she was born someone handed her to me and said your child has a severe handicap, she has got these so-called defects and this is the label we are going to put on it, I think that automatically I would start looking at her through the lens of that label. But because I had such a long period of time when she was just my daughter and when they handed her to me, she was perfect. Then I never looked at her with that label… And I think that to the extent that those labels are useful, they are useful in the sense that they give you an idea in terms of what you might expect down the road, in terms of behaviour, what interventions might help, and it opens the doors for therapies, but I don't think any child can be viewed through that label, to be honest with you. I have met a lot of wonderful kids who have Down syndrome. They are as varied as the people who don't have Down syndrome. I think the aspect that tends to get lost is that we have diversity even among the normal population. I mean it would be weird to say that that person has all of her chromosomes intact and people like that are generally pretty friendly. I mean, we can't make generalizations like that and I think we can't for people who have special needs either… I am starting to learn that. Because I used to believe, Oh, we can stick a label on someone and we can know everything about them

based on their profile and what the disease will tell us, but the disease is only a small part of what they are and who they are.

I found Stuart's words so insightful and would like to finish the dissertation on this note. As this father points out, labels can be very useful for accessing programs and giving an indication about what one can expect, but in the end, a label is a way of categorizing a set of behaviours or characteristics and does not tell us everything there is to know about a person. Furthermore, parents talked about the length of the diagnostic process and how difficult that was, but perhaps the length of the process serves a function; to prepare the family for the prognostic information that comes with the diagnosis and to prolong the assignment of a label.

Conclusion

This study looked at parents' experiences of the diagnostic process when a child has a developmental disability. A large body of literature has looked at the stress levels of parents of children with disabilities, considering the disability as the most significant factor affecting stress levels. However, I would suggest that the diagnostic process, with the uncertainty and dealing with numerous medical professionals, is a considerable contributor to the stressful experience of parents. Dykens and Hodapp (2001) have discussed how different diagnoses are associated with specific behavioural and developmental outcomes, but each of these disabilities also affects families in unique ways. Furthermore, specific diagnoses are detected differently and the diagnostic process is significantly affected by the differential diagnosis of the child. If we can make the diagnostic process less difficult for parents, then this may facilitate greater overall family adaptation to developmental disability.

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