

## Families and Differential Diagnosis of Developmental Disability

### Abstract

*This literature review presents the issues around families and differential diagnosis of developmental disability. The review begins with a brief overview of research on families of children with disabilities, followed by the arguments for and against labelling developmental disabilities. The limited information known about the diagnostic process from families 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.*

### In-Press

#### Author

Shelley L. Watson  
Laurentian University  
Ontario Canada

#### Correspondence

swatson@laurentian.ca

#### Key Words

labelling  
differential diagnosis  
families

Differential diagnosis of developmental disability 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 useful (e.g., Lauchlan & Boyle, 2007; McDermott et al, 2006), while others believe that differential diagnosis is essential for tailoring supports to 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 bring about intervention and social support, eventually resulting in an improved quality of life for the family and the individual (Gillman et al., 2000). 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. It is important to note that much of the research cited does not employ the term "differential diagnosis" specifically, but it is the author's belief that "differential diagnosis" can be readily substituted for terms such as labelling or classification.

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 concept of stigmatization, which is characterized by a negative evaluation of someone who possesses an attribute that is devalued; individuals who are stigmatized have a social identity that does not meet society's belief about what attributes the individual should possess (Crocker, Major, & Steele, 1998; Goffman, 1963; Kurzban & Leary, 2001). Link and Phelan (2001) define stigma as the "co-occurrence of its components - labeling, stereotyping, separation, status loss, and discrimination" (p. 363).

Scheff (1966) was the first to apply labelling 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. An individual's label then separates them from society's normative expectations, leading to stigmatization.

However, labels can also be helpful and this literature review presents the arguments for and against labelling developmental disabilities. The review begins with a brief overview of research on families of children with disabilities 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 interpretive research on family adaptation to diagnoses.

## **Families of Children with Developmental Disabilities**

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 became 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 utility 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 Lauchlan and Boyle's arguments will be highlighted in turn.

### **Access to treatment and resources**

The first and most significant reason in favour of differential diagnosis is that a label facilitates treatment and access to resources (Dykens & Hodapp, 2001; Dykens, Hodapp, & Finucane, 2000; Griffiths & Watson, 2004; Hodapp et al., 1998; Lauchlan & Boyle, 2007). Differential diagnosis can provide admittance 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 receive instructional emphasis on contextual learning and visual integration rather than focusing on auditory short-term memory and that 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 anomalies 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 differential diagnosis is often given, but once the label is in place, there is little concern for the actual type of intervention. They affirm that it is purely a matter of "label equals more money" or "label equals placement at special school" (p. 36), without thought for how the extra resources are helping the individual's challenges or specific needs. Differential diagnosis can be helpful if it leads to a targeted strategy that facilitates learning or adaptation, 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 specific challenging behaviour or learning 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. Students were taught about the impairments connected with autism and were allowed to ask questions about autism spectrum disorders. This open discussion led to an increased acceptance of the peer with autism and improved the classroom environment, as measured by a questionnaire completed by the students and discussion with the teacher. 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 facilitate psychological and educational discussion because they lower uncertainties regarding behavioural challenges and uniqueness (Griffiths & Watson, 2004; Dykens & Hodapp, 2001; Lauchlan & Boyle, 2007). Research and practice have 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 rarely agreement among professionals about how differential diagnoses are assigned and labels can lead to a generalization of children's problems. 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. Assignment of labels is also associated with cultural differences. In the United Kingdom, the term "learning disability" is used 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). When assessed by a psychologist, for example, an individual in Canada with a learning disability would have an IQ within the normal range, but would score below on an achievement measure. 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 families of children with disabilities by "explaining" their challenges (Lauchlan & Boyle, 2007; 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 lowered expectations. 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, are typically reported to have social and musical strengths (Finucane, 2004) and individuals with Prader-Willi syndrome to often excel at jigsaw puzzles (Dykens & Kasari, 1997). Each of these assets can be useful when tailoring interventions and supports for individuals with these differential diagnoses.

### **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 (Lauchlan & Boyle, 2007). 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 self-esteem issues and problems with 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. 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 associated with 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, 1991; Gillman et al., 2000; Watson, manuscript in preparation- b); access to appropriate intervention (Carmichael, Pembrey, Turner, & Barnicoat, 1999; Gillman et al., 2000; Poelmann et al., 2005; Watson, in preparation- b); 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., 2000). 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., 2005; Skotko, 2005; Watson, manuscript in preparation- a; 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, 2003), see fewer professionals throughout the diagnostic process (Goin-Kochel et al., 2006), and if professionals accept parents' first suspicions of disability (Brogan & Knussen, 2003).

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., 2000). Poelmann et al. (2005) cite one mother's reaction to receiving a diagnosis of fragile X syndrome 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 1991, 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 be to 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; Watson, manuscript in preparation- a; 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, 2005; Woolfe & Bartlett, 1996); doctors not listening to parents' concerns (Baird, McConachie, & Scrutton, 2000; Harrington, Patrick, Edwards, & Brand, 2006; Leonard, 1999; Watson, manuscript in preparation- a; Woolfe & Bartlett, 1996); negative information regarding disabilities (Hedov, Wikblad, & Annerén, 2002; Poelmann et al., 2005; Skotko, 2005); and the lack of information regarding disabilities and interventions (Hedov et al. 2002; Skotko, 2005; Sloper & Turner, 1993; Watson, in preparation- a).

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, 1999; Sloper & Turner, 1991; Woolfe & Bartlett, 1996). 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 syndrome or autism since these disorders are not typically recognized at birth or early infancy, but rather are diagnosed 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., 2006). In their interpretive study of 21 mothers of children with fragile X syndrome or 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 led parents to doubt the quality of the medical care their child was receiving (Harrington et al., 2006; Watson, manuscript in preparation- a; Woolfe & Bartlett, 1996). Consequently, many families direct frustration toward medical practitioners.

## Medical Professional Issues

Some researchers have postulated that the delays in receiving a differential 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., 2005) 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. (2005) 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 or 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 research on families of children with developmental disabilities 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 have employed a qualitative approach (e.g., Gillman et al., 2000; Leonard, 1999; Poelmann et al., 2005; Woolfe & Bartlett, 1996). Given that labelling often is 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, 2005; 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, 2005; Sloper & Turner, 1993), 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, 2005) 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 research with families of children with disabilities 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, 1999; 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 learning and social potentials of individuals with disabilities (Skotko, 2005).

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.

## Acknowledgements

This literature review was part of Shelley Watson's doctoral dissertation at the University of Alberta.

## References

- Abbeduto, L., Mailick Seltzer, M., Shattuck, P., Wyngaarden Krauss, M., Orsmond, G., & Murphy, M. M. (2004). Psychological well-being and coping in mothers of youths with autism, Down syndrome, or fragile x syndrome. *American Journal on*

- Mental Retardation*, 109(3), 237-254.
- American Association on Mental Retardation (2002). *Mental retardation: definition, classification, and systems of supports*. Washington, DC: Author.
- Baird, G., McConachie, H., & Scrutton, D. (2000). Parents' perceptions of disclosure of the diagnosis of cerebral palsy. *Archives of Disease in Childhood*, 83, 475-480.
- Becker, H. S. (1963). *Outsiders: Studies in the sociology of deviance*. New York: Free Press.
- Brogan, C. A. (2001). *The pathway to care for children with autism spectrum disorders aged 0 to 12 years*. Glasgow: National Autistic Society Scotland.
- Brogan, C. A., & Knussen, C. (2003). The disclosure of a diagnosis of an autistic spectrum disorder: Determinants of satisfaction in a sample of Scottish parents. *Autism*, 7(1), 31-26.
- Burden, R. (1991). Psycho-social transitions in the lives of parents of children with handicapping conditions. *Counselling Psychology Quarterly*, 4(4), 331-344.
- Carmichael, B., Pembrey, M., Turner, G., & Barnicoat, A. (1999). Diagnosis of fragile-X syndrome: The experiences of parents. *Journal of Intellectual Disability Research*, 43(1), 47-53.
- Crnic, K. A., Friedrich, W. N., & Greenberg, M. T. (1983). Adaptation of families with mentally retarded children: A model of stress, coping, and family ecology. *American Journal of Mental Deficiency*, 99(2), 125-138.
- Crocker, J., Major, B., & Steele, C. (1998). Social stigma. In D. T. Gilbert & S. T. Fiske (Eds.), *The handbook of social psychology* (pp. 504-553). Boston: McGraw-Hill.
- Dykens, E. M., & Hodapp, R. M. (2001). Research in mental retardation: Toward an etiologic approach. *Journal of Child Psychology and Psychiatry*, 42(1), 49-71.
- Dykens, E. M., Hodapp, R. M., & Finucane, B. M. (2000). *Genetics and mental retardation syndromes: A new look at behavior and interventions*. Baltimore, MD: Paul H. Brookes.
- Dykens, E. M., & Kasari, C. (1997). Maladaptive behavior in children with Prader-Willi Syndrome, Down Syndrome, and nonspecific mental retardation. *American Journal on Mental Retardation*, 102, 228-237.
- Ferguson, P. M. (2002). A place in the family: A historical interpretation of research on parental reactions to having a child with a disability. *Journal of Special Education*, 36, 124-135.
- Filipek, P. A., Accardo, P. J., Ashwal, S., Baranek, G. T., Cook, E. H., & Dawson, G., Gordon, B., Gravel, J. S., Johnson, C. P., Kallen, R. J., Llevy, S. E., Minshew, N. J., Ozonoff, J., Prizant, B., Rapin, I., Rogers, S. J., Stone, W. L., Teplen, S. W., Tuchman, R. F., & Volkmar, F. R. (2000). Practice parameter: Screening and diagnosis of autism: Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Child Neurology Society. *Neurology*, 55, 468-479.
- Finucane, B. (2004). Williams syndrome. In D. Griffiths & R. King (Eds.), *Demystifying syndromes: Clinical and educational implications of common syndromes associated with persons with intellectual disabilities* (pp. 75-88). Kingston, NY: NADD Press.
- Fotheringham, J. B., Skelton, M., & Hoddinott, B. A. (1972). The effects on the family of the presence of a mentally retarded child. *Canadian Psychiatric Association Journal*, 17, 283-289.
- Gersten, R., Irvin, L., & Keating, T. (2002). Critical issues in research on families: Introduction to the special issue. *Journal of Special Education*, 36, 122-123.
- Gillman, M., Heyman, B., & Swain, J. (2000). What's in a name? The implications of diagnosis for people with learning difficulties and their family carers. *Disability and Society*, 15(3), 389-409.
- Glascoe, F. P. (1999). Using parents' concerns to detect and address developmental and behavioral problems. *Journal of the Society of Pediatric Nurses*, 4(1), 24-35.
- Goffman, E. (1963). *Stigma: Notes on the management of spoiled identity*. Englewood Cliffs, NJ: Prentice Hall.
- Goin-Kochel, R., Mackintosh, V. H., & Myers, B. J. (2006). How many doctors does it take to make an autism spectrum diagnosis?

- Autism*, 10(5), 439-451.
- Griffiths, D. M., & Watson, S. L. (2004). Demystifying syndromes associated with developmental disabilities. In D.M. Griffiths & R. King (Eds.), *Demystifying syndromes: Clinical and educational implications of common syndromes associated with persons with intellectual disabilities* (pp. 1-30). Kingston, NY: NADD.
- Gus, L. (2000). Autism: Promoting peer understanding. *Educational Psychology in Practice*, 16(4), 461-468.
- Harrington, J. W., Patrick, P. A., Edwards, K. S., & Brand, D. A. (2006). Parental beliefs about autism: Implications for the treating physician. *Autism*, 10(5), 452-262.
- Hasnat, M. J., & Graves, P. (2000). Disclosure of developmental disability: A study of paediatricians' practices. *Journal of Paediatrics and Child Health*, 36 (1), 27-31.
- Hedov, G., Wikblad, K., & Annerén, G. (2002). First information and support provided to parents of children with Down syndrome in Sweden: Clinical goals and parental experiences. *Acta Paediatrica*, 91, 1344-1349.
- Hodapp, R. M., Fidler, D. J., & Smith, A. C. (1998). Stress and coping in families of children with Smith-Magenis Syndrome. *Journal of Intellectual Disability Research*, 42(5), 331-340.
- Keogh, B. K., Garnier, H. E., Bernheimer, L. P., & Gallimore, R. (2000). Models of child-family interactions for children with developmental delays: Child-driven or transactional? *American Journal on Mental Retardation*, 105(1), 32-46.
- Knox, M., Parmenter, T. R., Atkinson, N., & Yazbeck, M. (2000). Family control: The views of families who have a child with an intellectual disability. *Journal of Applied Research in Intellectual Disabilities*, 13, 17-28.
- Kurzban, R., & Leary, M. R. (2001). Evolutionary origins of stigmatization: The functions of social exclusion. *Psychological Bulletin*, 127(2), 187-208.
- Lauchlan, F. & Boyle, C. (2007). Is the use of labels in special education helpful? *Support for Learning*, 22, 36-42.
- Learning Disabilities Association of Canada. (2005). *Official definition of learning disabilities*. Retrieved July 9<sup>th</sup>, 2007, from [http://www.ldac-taac.ca/Defined/defined\\_new-e.asp](http://www.ldac-taac.ca/Defined/defined_new-e.asp)
- Lee, T. H., Blasey, C. M., Dyer-Friedman, J., Glaser, B., Reiss, A. L., & Eliez, S. (2005). From research to practice: Teacher and pediatrician awareness of phenotypic traits in neurogenetic syndromes. *American Journal on Mental Retardation*, 10 (2), 100-106.
- Leonard, A. (1999). *Right from the start: Looking at diagnosis and disclosure: Parents describe how they found out about their child's disability*. London, UK: Scope.
- Link, B. G., & Phelan, J. C. (2001). Conceptualizing stigma. *Annual Review of Sociology*, 27, 363-385.
- Lord, C., & Risi, S. (2000). Diagnosis of autism spectrum disorder in young children. In A. M. Wetherby & B.M. Prizant (Eds.), *Autism spectrum disorders: A transactional developmental perspective* (pp. 11-30). Baltimore, MD: Brookes.
- McCubbin, H. I., & Patterson, A. I. (1987). The T-Double ABCX Model of Family Adjustment. In H.I. McCubbin & A.I. Thompson (Eds.), *Family assessment inventories for research and practice* (pp. 3-32). Madison, WI: University of Wisconsin-Madison.
- McDermott, R., Goldman, S., & Varènne, H. (2006). The cultural work of learning disabilities. *Educational Researcher*, 35(6), 12-17.
- Ogilvy, C. M. (1994). What is the diagnostic significance of specific learning difficulties? *School Psychology International*, 15, 55-68.
- Patterson, J. M. (1989). A family stress model: The family adjustment and adaptation response. In C.N. Ramsay Jr. (Ed.), *Family systems in medicine* (pp. 95-118). New York: Guilford Press.
- Poelmann, J., Clements, M., Abbeduto, L., & Farasad, V. (2005). Family experiences associated with a child's diagnosis of fragile X or Down syndrome: Evidence for disruption and resilience. *Mental Retardation*, 43(4), 255-267.
- Ralston, S. J. (2000). Reflections from the

- trenches: One doctor's encounter with disability rights arguments. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights* (pp. 334-3369). Washington: Georgetown University Press.
- Scheff, T. (1966). *Being mentally ill: A sociology theory*. Chicago: Aldine.
- Shah, K. (2001). Research in brief: What do medical students know about autism. *Autism, 15*, 127-133.
- Shapiro, J. (1983). Family reactions and coping strategies in response to the physically ill or handicapped child. *Social Science and Medicine, 17*, 913-931.
- Shapiro, J., Blacher, J., & Lopez, S. R. (1998). Maternal reactions to children with mental retardation. In J.A. Burack, R.M. Hodapp, & E. Zigler (Eds.), *Handbook of mental retardation and development* (pp. 606-636). New York: Cambridge University Press.
- Sices, L, Feudtner, C., McLaughlin, J., Drotar, D., & Williams, M. (2004). How do primary care physicians manage children with possible developmental delays? A national survey with an experimental design. *Pediatrics, 113*(2), 274-282.
- Simon, E. (2004). Smith-Magenis syndrome. In D. Griffiths & R. King (Eds.), *Demystifying syndromes: Clinical and educational implications of common syndromes associated with persons with intellectual disabilities* (pp. 89-105). Kingston, NY: NADD Press.
- Singer, G. H. S. (2002). Suggestions for a pragmatic program of research on families and disability. *Journal of Special Education, 36*(3), 148-156.
- Skotko, B. (2005). Mothers of children with Down syndrome reflect on their postnatal support. *Pediatrics, 115*, 64-77.
- Sloper, P., & Turner, S. (1991). Parental and professional views of the needs of families with a child with severe physical disability. *Counselling Psychology Quarterly, 4*(4), 323-331.
- Summers, J. & Pittman, D. (2004). Angelman syndrome. In D. Griffiths & R. King (Eds.), *Demystifying syndromes: Clinical and educational implications of common syndromes associated with persons with intellectual disabilities* (pp. 161-188). Kingston, NY: NADD.
- Sutcliffe, J., & Simons, K. (1993). *Self advocacy and people with learning difficulties*. Leicester, UK: NIACE.
- Trute, B. (2005). *Family-centred care in childhood disability services: Understanding the steps in the dance*. Paper presented at the Family Supports for Children with Disabilities Research Symposium, Edmonton, AB.
- Turnbull, A. P., Friesen, B. J., & Ramirez, C. (1998). Participatory action research as a model for conducting family research. *Journal of the Association for Persons with Severe Handicaps, 23*(3), 178-188.
- Turnbull, A. P., & Turnbull, H. R. (1997). *Families, professionals, and exceptionality: A special partnership*. Upper Saddle River, NJ: Merrill.
- Watson, S. L. (manuscript in preparation- a). *Parents' experiences dealing with professionals when seeking a differential diagnosis*.
- Watson, S. L. (manuscript in preparation- b). *Why do parents seek a differential diagnosis? A search for meaning and support*.
- Wilcox, D. (1991). Heather's story: The long road for a family in search of a diagnosis. *Exceptional Parent, 21*. 92-94.
- Williams, C. A., Lossie, A., Driscoll, D., & the R.C. Philips Unit. (2001). Angelman syndrome: Mimicking conditions and phenotypes. *American Journal of Medical Genetics, 101*, 59-64.
- Wing, L. (1999). Notes from Autism '99 Internet conference. Retrieved May 15<sup>th</sup>, 2006, from [www.autism99.org](http://www.autism99.org)
- Woolfe, R., & Bartlett, S. (1996). One bad experience is one too many: Parents' experiences of the diagnosis of disability in their infants. *Counselling Psychology Review, 11*(2), 7-11.