

The Diagnostic Process of Children with Autism Spectrum Disorder: Implications for Early Identification and Intervention

Abstract

Obtaining a diagnosis of autism spectrum disorder (ASD) often includes long delays, multiple diagnoses, and several visits to healthcare professionals. Consequently, parents often report dissatisfaction and frustration with the diagnostic process and their child's diagnosis. One hundred and sixty-six parents/caregivers of children in Canada with various ASD diagnoses completed a self-report questionnaire related to their experiences with the diagnostic process. Parents expressed high levels of satisfaction with their child's current diagnosis, despite delays and concerns related to additional psychological issues identified, particularly anxiety. Difficulties in obtaining a diagnosis may not only have a significant impact on the timing of intervention, it can also increase family stress and caregiver burden. As such, it is essential to understand families' experiences in order to help improve the diagnostic process.

The degree of heterogeneity of symptom presentation of individuals with autism spectrum disorder (ASD), coupled with potential co-morbid mental health concerns, can complicate the process of obtaining an accurate diagnosis. As a result, individuals with ASD may be undiagnosed, misdiagnosed, or receive numerous diagnoses (Siklos & Kerns, 2007; White, Oswald, Ollendick, & Scahill, 2009). Delays in early identification have detrimental effects on the child receiving early and appropriate intervention. Consequently, parents are often dissatisfied with the process in obtaining an ASD diagnosis, which can contribute to the stress and caregiver burden that these families often feel (Howlin & Moore, 1997; White et al., 2009). It is therefore important to understand more about how the diagnostic process affects caregivers, and how co-morbid mental health issues may complicate the diagnostic process.

Autism Spectrum Disorder

ASD encompasses a variety of functionally impairing symptoms ranging in severity from mild to severe, and emerges during early childhood. The Diagnostic and Statistical Manual of Mental Disorders, 4th edition (DSM-IV-TR; American Psychiatric Association [APA], 2000), differentiated several disorders under the diagnostic category pervasive developmental disorders (PDD), including three that will be a focus for the present study: autism, pervasive developmental disorder-Not Otherwise Specified (PDD-NOS), and Asperger syndrome (AS). All are characterized by delays or abnormalities in social and communication skills, such as significant qualitative

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impairments in social interactions and restricted or stereotyped patterns of behaviour, interests and activities. Language delays, delays in cognitive development, self-help skills, and adaptive behaviour are also common, though generally milder among individuals with AS. The recently-released Diagnostic and Statistical Manual of Mental Disorders, 5th Edition (DSM-5; APA, 2013), uses the single term “autism spectrum disorder” to encompass these disorders. Persons with this diagnosis vary in the degree of expression of the core symptoms, along with other associated characteristics, such as degree of cognitive impairment. The individuals who are the focus of the present study were diagnosed according to the DSM-IV-TR criteria; its terminology will be used throughout.

Difficulties in Obtaining a Diagnosis

Children with more severe ASD symptoms, such as severe speech and language deficits, and overt behavioural impairments, such as hand flapping and toe walking, often receive a diagnosis earlier compared to children with less obvious impairments (Mandell, Maytali, Novak, & Zubritsky, 2005; Siklos & Kerns, 2007). Symptoms associated with higher functioning ASD such as social and behavioural impairments are often less observable, and therefore more difficult to diagnose (Siklos & Kerns, 2007). Delays and difficulties in obtaining a diagnosis are also associated with families making multiple visits to a variety of health-care professionals. In a recent Canadian study, Siklos and Kerns (2007) found that parents visited an average of 4.5 health professionals before obtaining a diagnosis, and received that diagnosis an average of 2.8 years after their first visit to a professional (Siklos & Kerns, 2007). Similar experiences have been reported elsewhere in Canada (Oullette-Kuntz et al., 2009), the United States (Wiggins, Baio, & Rice, 2006), and the United Kingdom (Howlin & Moore, 1997).

Further, it has been found that longer delays between the initial expression of concerns and diagnosis were associated with less parental satisfaction with the diagnostic process (Howlin & Asgharian, 1999). Howlin and Asgharian (1999) also found that parents of children diagnosed with AS expressed initial concerns at a later age than parents of children with autism; they also experienced significantly longer delays in

receiving a diagnosis, and more frustration with the diagnostic process. These authors suggest that this longer delay may be due to the fact that children with AS often present as developmentally advanced due to their mature vocabulary and extensive knowledge of particular topics.

Co-occurring mental health disorders can further complicate diagnosis of ASD. In particular, symptoms of anxiety are prevalent among children and youth with ASD (Bellini, 2004; White et al., 2009). Although anxiety is not considered a phenomenological feature of ASD, it may exacerbate social difficulties associated with ASD, particularly family relationships and social interactions (Reavan, et al., 2009). Clinicians often attribute shared features of anxiety and ASD to the core symptoms of ASD, instead of diagnosing the co-morbid anxiety (Grondhuis & Aman, 2012). This often increases the time to receive a diagnosis and subsequent treatment for anxiety, if it is addressed at all (Grondhuis & Aman, 2012).

Implications for Delays in the Diagnostic Process

Delays in the ASD diagnostic process can adversely impact children and their families in a variety of ways. For example, delays in accessing needed services and treatment can have a negative impact on prognosis as early intervention is important (Siklos & Kerns, 2007). Delays can also affect the quality of life for family members, for example, the presence of problem behaviours (Estes et al., 2009; Konstantareas & Homatidis, 1989; Lecavalier, Leone, & Wiltz, 2006; Wiggins, et al., 2006), caregiving challenges (Abbeduto et al., 2004), financial burden and limited social activities (Lecavalier et al., 2006), as well as increased risk of mental health problems among parents (Weiss, Cappadocia, MacMullin, Vecili, & Lunskey, 2012).

The Present Study

The present study aimed to examine various aspects of the diagnostic process of children with an ASD in a Canadian sample, with four key objectives.

- 1) To monitor the stability of the diagnosis received. The initial diagnosis may have

remained constant, or the primary diagnosis may have changed a number of times, such as if individuals were previously diagnosed with other mental health issues (e.g., anxiety) or other childhood disorders (e.g., attention deficit hyperactivity disorder; ADHD). Consistent with previous research, we hypothesized that less severely affected individuals would have the most unstable diagnosis.

- 2) To examine how long families waited to obtain an ASD diagnosis; specifically, the lengths of time between when families were initially concerned, when they received their first diagnosis, and when they received their current diagnosis. We expected that children with less severe presentations of ASD (i.e., AS, PDD-NOS, etc.) would have experienced a number of changes in their primary diagnoses, and that time to diagnosis would be longer compared to children with more complex expressions (i.e., autism, ASD).
- 3) To examine the number of families who expressed concerns about the presence of other mental health difficulties, and how associated that was with caregivers' satisfaction with their child's current diagnosis. We expected that anxiety would be a significant concern for parents of children and adolescents with ASD, based on the findings of White and colleagues (2009).
- 4) To better understand families' satisfaction with their child's current diagnosis. Consistent with the work of Howlin and Moore (1997) and Howlin and Asgharian (1999), we expected that parents of children with a current diagnosis of AS would be the least satisfied with their child's current diagnosis in comparison to other ASD diagnoses.

Method

Participants

The current study is part of a larger research project examining the diagnostic process with Canadian families of individuals with ASD. Families were recruited through a participant registry created and maintained by the ASD Canadian-American Research Consortium

(ASD-CARC, Queen's University), through local agencies and community newsletters, and through the Children's Learning Projects lab, York University. Ethics approval was obtained through the Research Ethics Boards of York University and Queen's University.

The data from 166 caregivers of children with various ASD diagnoses (i.e., autism, ASD, AS, PDD, and PDD-NOS) living in Ontario, Canada were used. The individuals who are the focus of the present study were diagnosed according to the DSM-IV-TR (American Psychiatric Association, 2000) or earlier editions, by which individuals may have been diagnosed with AS or PDD-NOS, as well as autism. Due to the small sample sizes in each diagnostic sub-group, diagnoses were clustered into three diagnostic groupings: (1) ASD and PDD; (2) autism and PDD-NOS; and (3) AS. Given the similarity in the presentation and severity of the symptoms of specific subgroups (e.g., ASD and PDD), these groupings seemed to be the most appropriate.

Overall, 41% ($n = 68$) of caregivers had children who were currently diagnosed with ASD or PDD, 28% ($n = 47$) had a diagnosis of AS, and 30% ($n = 51$) had a primary diagnosis of autism or PDD-NOS. The children were predominantly male, ranging in age from approximately 2 years to 53 years ($M = 16.6$ years, $SD = 9.32$). However, 45 parents did not indicate their children's age, and over half ($n = 108$) of the sample failed to indicate the child's gender. Demographic information was not obtained from the caregivers/parents who completed the questionnaire.

Measure

A 19-item questionnaire was designed to examine parents' experiences in obtaining an ASD diagnosis. Questions were grouped into four main focal areas. In the *Stability of Diagnosis* area, families answered a multiple-choice question in which they identified their child's current primary ASD diagnosis. Additionally, families answered yes/no if their child had received a different diagnosis previously, and if so which diagnosis(es) they had received. In the *Delays in Diagnosis* area, parents were asked to answer open-ended questions pertaining to the ages at which their child had received their current and previous diagnoses. Therefore, the

number of items caregivers completed in this section was dependent on how many previous diagnoses their child had received. In the *Additional Psychological Concerns* section (two items), caregivers indicated if they had any additional psychological concerns about their child, and if so, they specified which from a list of common psychological concerns. Caregivers were able to note multiple concerns. Lastly, the section related to *Parent's Satisfaction with Current Diagnosis* involved one question in which caregivers were required to indicate if they were satisfied with their child's current diagnosis (yes/no), and to provide a qualitative explanation as to why or why not. The questionnaire was administered on-line, or, if preferred, by paper copy.

Statistical Analysis

Frequency analyses, chi-square analyses, and one-way analysis of variance (ANOVA) were conducted to examine the quantitative data of the present study.

Results

Stability of Diagnosis

Of our sample of 166 families, forty-three percent ($n = 72$) reported that the diagnoses had remained stable over time, meaning that the child's current primary diagnosis was the same as the initial diagnosis. Of the respondents who indicated their child had a different diagnosis, 21.7% ($n = 36$) had previously received a diagnosis within the ASD classification. For example, the child was initially diagnosed with autism, but had a current diagnosis of AS. The remaining 31.3% ($n = 52$) of caregivers reported an initial diagnosis outside of ASD. Most commonly, the initial diagnosis was related to a learning-related disability, medical disability (e.g., Cerebral palsy, spina bifida), behavioural difficulty (e.g., oppositional defiant disorder), mental health disorder (e.g., anxiety), or another child disorder (e.g., attention deficit hyperactivity disorder). Of note, six families failed to indicate if their child had received a previous diagnosis.

There were significant differences between three diagnostic groups and the stability of

children's current diagnosis [$\chi^2_{(4, 160)} = 19.64, p < .001$]. In particular, individuals with AS were more likely to have experienced instability in their diagnosis, where 52% had a non-ASD diagnosis initially. In contrast, the ASD/PDD (43.5%) and autism/PDD-NOS (63.83%) groups were more likely to have maintained the same diagnosis.

Delays in Diagnosis

Overall, there was no significant difference between the diagnostic subgroups in terms of the age at which caregivers first became concerned with their child's development, [$F_{(1, 159)} = 2.23, p > .05$]. Data on the child's age at which families first consulted a health professional was only available for 59 participants; there was no difference between diagnostic subgroups, [$F_{(2, 57)} = 2.77, p > .05$]. However, caregivers of children with AS ($M = 41.80$; $SD = 38.01$) reported waiting significantly longer to receive their initial diagnosis than children with ASD/PDD ($M = 18.81$; $SD = 16.57$) or autism/PDD-NOS ($M = 19.77$; $SD = 26.77$).

Of those children who had experienced a change in their diagnosis ($n = 86$), the average length of time between their initial diagnosis and their current diagnosis was 28.16 months ($SD = 38.13$), with 52.3% ($n = 46$) of children experiencing a wait of 12 months or longer (ranging between less than one month to 164 months). Wait times between receiving current and initial diagnosis, also significant differed by diagnostic group. Families with a child with a current AS diagnosis ($M = 45.28$; $SD = 47.81$) had waited significantly longer between their initial and current diagnoses compared to those with a current diagnosis of ASD/PDD ($M = 18.59$; $SD = 26.25$) or autism/PDD-NOS ($M = 10.00$; $SD = 11.22$), $F_{(2, 85)} = 7.51, p < .001$.

Additional Psychological Concerns

Fifty-eight percent of respondents ($n = 94$) expressed concern about additional psychological issues facing their children, despite reporting satisfaction with their child's current diagnosis. Of those respondents who indicated that they had additional psychological concerns, anxiety was the most common issue identified (68.1%; $n = 64$) followed

by aggression (29.8%; $n = 28$), and “other concerns” (13.8%, $n = 13$), including depression and future life outcomes. Multiple concerns could be reported, so that percentages do not add to 100%. Concerns about suicide were expressed the least often, by 7.4% ($n = 7$) of respondents.

Parental Satisfaction with Current Diagnosis

A frequency analysis revealed that the majority (86.7%) of parents were satisfied that the current diagnosis captured their child’s characteristics and issues. A Pearson chi-square revealed that there were no significant differences in parental satisfaction across diagnostic categories, $\chi^2_{(2, 165)} = 1.03, p > 0.05$.

Discussion

This study examined aspects of the diagnostic process of parents of children with ASD in a Canadian sample, in particular the stability of the diagnosis received, the length of time to obtain a diagnosis, the presence of additional mental health concerns, and families’ satisfaction with their child’s current diagnosis.

As expected and consistent with previous research, over half of our respondents indicated instability in their diagnosis over time. Due to the range and severity of symptoms presented by children with AS, parents often report misdiagnosis, change in diagnosis, and/or diagnostic substitution (King & Bearman, 2009). In our sample, significant changes over time in diagnosis were noted most frequently by caregivers of children with AS. This finding is not surprising considering the previous research on AS (e.g., Howlin & Asgharian, 1999). As Siklos and Kerns (2007) noted, symptoms that are typically associated with higher functioning individuals with ASD are often less identifiable, and therefore more difficult to diagnose. In our sample, children with AS were more likely to have been previously diagnosed with other medical, childhood, and mental health disorders, prior to receiving their current AS diagnosis.

Families who received multiple diagnoses or changes in their diagnosis also experienced

delays and long wait times between when their children received their initial and their current diagnoses, with families of individuals with AS experiencing the longest delays. In particular, families of children with AS not only waited longer to receive their initial diagnosis, but also had longer wait times between receiving their initial diagnosis and the current AS diagnosis. Delays for children with AS could be attributed to a number of factors, including later recognition of “atypical” development, waiting longer to consult a health professional, receiving multiple diagnoses prior to their current AS diagnosis, and delays due to the complex and less severe presentation of symptoms. However, our results showed that families of children with AS became concerned with their child’s development around the same age as did other families, and they did not wait longer to consult a health professional. Additionally, post-hoc analyses showed children with AS did not receive a greater number of diagnoses prior to their current diagnosis, compared to the other ASD diagnostic subgroups; this indicates that the length of time to obtain a diagnosis is not likely due to multiple diagnoses. As such, delays in diagnosis may be best explained by difficulties among health professionals in diagnosing their child; that is, taking significantly longer to identify the symptoms of AS. It would be beneficial for future research to examine this hypothesis further.

In terms of other psychological concerns, the findings are consistent with previous work (Bellini, 2004; White et al., 2009), insofar as several parents had concerns about their child’s anxiety in addition to their ASD diagnosis. This finding emphasizes the need to further investigate to co-occurrence of anxiety and ASD, and possibly to develop ASD interventions that address issues associated with anxiety within this population. Overall, approximately half of the parents who participated in the study expressed concern about their child’s additional psychological issues. This high level of parental concern is surprising in light of parents’ high levels of satisfaction with their children’s ASD diagnoses.

The present study also found that most parents reported satisfaction with their child’s current ASD diagnosis, despite long wait times and diagnostic instability.

Additionally our findings show little relationship between level of parental satisfaction and specific ASD diagnostic subgroups. This finding was contrary to Howlin and Moore (1997), who found that parents of children diagnosed with AS reported significantly less satisfaction with their children's diagnosis than parents of children with other forms of ASD. It is possible that the difference is due to greater awareness of AS due to increased media and government focus in recent years. This focus may have the dual effects of raising awareness of AS and other ASDs, and of educating the public, including affected families, leading to modified expectations and resulting in less dissatisfaction. Those families in the present study who did indicate that they were not satisfied with their children's current diagnoses indicated a number of reasons, such as they believed that another diagnosis fit their child's symptoms better (e.g., "Social problems may be due to NLD; but there is still a lack of understanding and support for NLD, so maybe better to be labelled Aspergers"), some difficulties are not being identified with the current diagnosis (e.g., "He probably still has some issues with ADD, but they ruled it out"), the diagnosis was too vague (e.g., "He seems to be straddling a fuzzy boundary between what one might call Autism and what one might call Asperger syndrome"), and that it was the intervention that mattered, not the label (e.g., "I don't care if the behaviours are given a specific diagnosis, rather I'm greatly concerned that they be addressed"). However, further analysis of this qualitative data is needed to better understand the reasons for family satisfaction and dissatisfaction.

The present study has a number of limitations that affect the generalizability of the findings. First, a number of families failed to answer individual questions, resulting in missing data, which consequently limits the generalizability of some of the present findings. Second, there may have been a response bias among participants. Specifically, parents who feel extremely satisfied or dissatisfied with their child's diagnosis may have been highly motivated to participate in our study in order to express their praise for the health system or to influence its change. Individuals with weaker opinions may have self-excluded from the sample. Second, the cross-sectional retrospective design of our study captures the experiences and attitudes

of respondents within a narrow time-frame. Attitudes towards the ASD diagnosis may change through the diagnostic process. Third, although it can be assumed that difficulties with diagnoses negatively impacted parent's level of stress, the present study did not directly investigate this. As such, it is difficult to draw firm conclusions regarding families' well-being and quality of life during the diagnostic process. The present study primarily relied on quantitative data; however, obtaining qualitative information regarding the diagnostic process would be particularly beneficial as it would provide rich information regarding the diagnostic process, and potentially what specific child, family, health professional, and system factors impact this process. Although the questionnaire asked if caregivers were currently satisfied with their child's diagnosis, caregivers were not asked directly about their satisfaction with the process of obtaining that diagnosis. As such, it will be important for future research to investigate this further in a Canadian sample.

To these authors' knowledge, this is one of the first studies in Canada to examine aspects of the diagnostic process among families of children with ASD, including the stability of diagnosis, delays in the process, and parents' overall satisfaction with their children's diagnoses. Despite the extensive research in the United States and the UK, the present study aimed to better understand this process for Canadians, and accessed families in the most populated province in the country. Unlike other studies, the present study investigated the incidence of other psychological issues in the children. Given the emergent evidence of the prevalence of mental health concerns in individuals with ASD (e.g., White et al., 2009), this study provides an indication that such other mental health difficulties are not uncommon. Lastly, this study is timely, as the new DSM-5 has been recently introduced, involving revised diagnostic criteria for ASD, which provides further urgency for improvement in diagnostic procedures, and the need for best practices, in order to ensure early identification of individuals with ASD.

Delays in obtaining a diagnosis and the stability of diagnoses over time have significant implications for effective and appropriate intervention. Early intensive intervention is critical when treating children with ASD. Delaying

families' access to early intensive treatment has cascading effects not only on the child, but also the family (e.g., caregiver burnout, etc.). Thus, continuing research investigating how professionals can detect and diagnosis ASDs earlier, effectively, and more efficiently is essential. Early detection of ASD may also help reduce long-term education, intervention, and public health costs, by enabling timely intervention for the child and family.

Key Messages From This Article

Persons with disabilities: Parents of children with Autism are satisfied with their child's current diagnosis even though their diagnosis changed a lot and families waited a long time.

Professionals: Several families of children with ASD reported instability in their diagnosis, along with long wait times and co-morbid mental health issues. The implementation of diagnostic guidelines, along with the consideration of co-morbid psychological issues, may improve the diagnostic process for families of children with ASD.

Policymakers: Changes in diagnosis, along with delays in obtaining a diagnosis may lead to years of inappropriate or non-existent intervention for the child, and may increase the stress for caregivers.

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