

The Face of Autism



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Early childhood autism was first described by Dr. Leo Kanner in 1943, yet there are many reports of the existence of this disorder well before it was formally recognized (Olley, 1999; Frith, 2003). As it is defined today, autism is estimated to affect 13 in 1000 children (Fombonne, 2005). According to the Diagnostic and Statistical Manual of Mental Disorders IV (DSM-IV) and the World Health Organization (WHO), autism is classified as the most severe type of pervasive developmental disorder (PDD). It is characterized by a triad of behavioural abnormalities, namely, difficulty in social development and communication, as well as repetitive or obsessive interests (APA, 1994; WHO, 1993; Baron-Cohen, 2004). All three deficits must be present for a positive diagnosis of autism, while less severe or fewer deficits may result in a positive diagnosis of another PDD. Here we will explore the early life of a child affected by autism, including difficulties faced by the child and his or her family, and what can be done to help them. Furthermore, we will investigate some of the uncertainties and issues that demand further research.

Since its official discovery, the understanding of autism has changed dramatically. Kanner originally reported no association of autism with other disorders, although more recent research has shown a link between autism and disorders such as Fragile X Syndrome, epilepsy, mental retardation, tuberous sclerosis and neurofibromatosis (Rutter, Bailey, Bolton & LeCouteur,

1994; Smalley, 1998; Williams & Hersh, 1998). Once thought to be caused by neglectful or abusive parental behaviour, it is now widely accepted that autism is a neurobiological disorder with a strong genetic basis (Bettelheim, 1967; Volkmar, Chawarska & Klin, 2005; Baron-Cohen, 2004). As of yet, there has been no single environmental risk factor that has been shown to increase the risk of autism (Fombonne, 2003). As its onset typically occurs before 3 years of age, autism and other PDDs can be diagnosed by 24 months. Unfortunately, early diagnosis does not always occur (Volkmar & Pauls, 2003). It is often years after parents have noticed signs and have sought professional help that their child gets a diagnosis. Factors such as the lack of availability and accessibility of appropriate tools and awareness of health care professionals contribute to delays in intervention. This is problematic, as intervention for autism is most beneficial when managed at an earlier stage (McGahan, 2001; Erba 2000).

AUTISM AND DIAGNOSIS

The American Academy of Neurology recommends screening for autism in children that fail to meet developmental milestones. Validated screens include The Checklist for Autism in Toddlers, to be used for infants and toddlers (up to 18 months); The Screening Tool for Autism in Two-Year-Olds; and the Autism Screening Questionnaire, for children older than four years of age (Stone, Coonrod, Turner & Pozdol, 2004; Filipek et al., 2000). Autism is diagnosed by experienced clinicians using standardized diagnostic tools that focus on the symptoms of autism through observation of the child's behaviour rather than by examination of etiological factors. Instruments for diagnostic observation include the Childhood Autism Rating Scale, and the Autism Diagnostic Observation Schedule-Generic (ADOS-G) (Lord et al., 2000). Also useful are diagnostic parental interviews, which include the Gilliam Autism Rating Scale, The Parent Interview for Autism, The Pervasive Developmental Disorders Screening Test and the Autism Diagnostic Interview- Revised (Filipek, Accardo, Ashwal, Baranek, Cook, Dawson, et al., 2000). These systematic interview and observation schedules for diagnosis seek to find typical autistic signs and symptoms in a combination that would synergistically point to a PDD.

Due to the symptoms exhibited by children with autism, a lack of social interest may be observed during their first year of life. Impairment in social interaction includes minimal eye contact, a lack of social reciprocity, and reluctance to form peer relationships (Volkmar & Pauls, 2003). Failure to respond to verbalizations or to one's name, as well as a tendency to be more interested in objects



Figure 1: Interactive social behaviour, as seen here, may be absent in children with autism (Courtesy of Dr. Lonnie Zwaigenbaum).

than in people may also be observed (Volkmar, Chawarska & Klin, 2005). Other behaviours in these young children include atypical eye contact and visual tracking, lack of visual attention, imitation, and reactivity (Zwaigenbaum et al, 2005). It has also been shown that impaired sensory-motor functioning, in the form of abnormal movement patterns, object manipulations, and postural adjustments, also have the potential to serve as an early marker of autism in infancy; however, signs of impairment usually become more evident as the autistic child grows older (Baranek, 1999).

Problems associated with communication involve a lack of language with no attempt to compensate for communication, or, if language is present, echolalia and persistent pragmatic difficulties (Volkmar & Pauls, 2003). This would include an absence of gestures and facial expressions. A child with autism is unlike a child who is hearing impaired, as those children will typically find alternate means of communicating their needs. Other behaviours typically seen in young autistic children are abnormal preoccupations, interests or activities; difficulties with a change in routine; and stereotyped mannerisms (Volkmar & Pauls, 2003). One example might be an unusual interest in a certain object, like beads on a string or spinning the wheels of a toy truck. A child with autism may have elaborate rituals or routines that are followed meticulously, and may even possess obsessive tendencies. Autistic stereotypes include odd finger movements or finger flicking, hand flapping, and body rocking or unusual posturing.

A "sensory phenomena" is often reported in children with autism. Although not necessary for a diagnosis, many autistic children seem to exhibit either a hyper or hyposensitivity to certain stimuli. This may result in either a heightened tolerance or sensitivity to pain, sound, smell, taste, or sight (Frith, 2003).

INTERVENTION & TREATMENT

Despite early claims of a cure (for example, Lovaas, 1987), autism is a lifelong disability (Volkmar & Pauls, 2003). Because of links between autism and various neurological signs, it is widely accepted that autism is a neurological disorder, despite the fact that its etiology is still to be established. In the absence of a cure targeted at the basic etiology, treatment for autism is based on behavioural, developmental, and educational approaches. However, in the absence of randomized controlled trials that utilize appropriate methodology to test the effects of these interventions, many questions still remain unanswered (Howlin, 2005). It has become evident that no one treatment for autism will be effective for all autistic children, thus treatments need to be adapted for every individual's needs (Howlin, 1998). Early detection and intervention is of high priority for the best outcome from any treatment (Bryson, Rogers & Fombonne, 2003). Several models for treatment are available and implemented, some of which are described here.

As stated above, every intervention needs to be individualized and well planned to suit each child. However, in general, a therapy that is consistently applied, and targets language and other areas of development will significantly aid in the child's development, language and cooperative skills (Bryson, Rogers, & Fombonne, 2003).

INTENSIVE APPLIED BEHAVIOURAL ANALYSIS

Intensive Applied Behavioural Analysis (ABA) – widely used in Ontario – is based on a method developed by I.O. Lovaas. Children are subjected to the reinforcement-based, intensive, one-on-one treatment for 20-40 hours per week over a span of two years (Lovaas, 1987). ABA is clearly beneficial to many children with autism, and although there is evidence for its efficacy, controversy still exists with regards to its true benefit. Lovaas' study (1987) is methodologically stronger than many studies looking for similar outcomes, despite some major problems such as lack of randomization and limited outcome measures (Bryson, Rogers & Fombonne, 2003). Lovaas lacks the power to make such strong claims such as the achievement of normal functioning, making many researchers hesitant to accept his results (Basset, Green & Kazanjian, 2000). In the face of these limitations, several problems arise. The challenge becomes providing effective intervention, while simultaneously obtaining useful information regarding the facets of treatment and program intensity (Miller & Zwaigenbaum, 2001). Sheinkopf and Siegal (1998) performed a study that observed the development of children and the impact of intensive behavioural treatment in a home environment. The researchers found higher post-treatment IQ scores as well as decreased symptoms in those children who had received the treatment. However, all patients continued to meet criteria for autism and PDD (Sheinkopf & Siegal, 1998). From this evidence, it is clear that ABA is effective and beneficial, but cannot provide a complete solution.

TEACCH

The Treatment and Education of Autistic and Related Communication Handicapped Children Program (TEACCH) is a program that focuses on developing cognitive, academic, and prevocational skills in school-aged children (Ozonoff & Cathcart, 1998). Developed in 1966 at the University of North Carolina, the TEACCH program provides a structuralized environment for skill acquisition. It aims to gradually help children gain independence and hone in on a child's strengths in areas such as visual-spatial understanding and object manipulation (Schopler & Reichler, 1971; Dawson & Osterling, 1997; Bryson, Rogers & Fombonne 2003). It utilizes parents as co-therapists to increase intervention time. In a study that compared children who received the home-based treatment to children that did not, the former demonstrated significantly greater improvement in areas such as imitation, fine motor, gross motor, and cognitive performance, and to a lesser extent, greater improvement in perception, and cognitive verbal skills (Ozonoff & Cathcart, 1998). Despite limited generalizations of the results due to design of such studies, these results outline the effectiveness of the TEACCH treatment program for improving cognitive and developmental skills.

LEARNING EXPERIENCES...AN ALTERNATIVE PROGRAM FOR PRESCHOOLERS AND PARENTS (LEAP)

The LEAP program was developed in 1984 by Hoyson, Jamieson and Strain, and is administered for about 15 hours per week (Dawson & Osterling, 1997). It incorporates a combination of several learning theories, with the primary focus being social

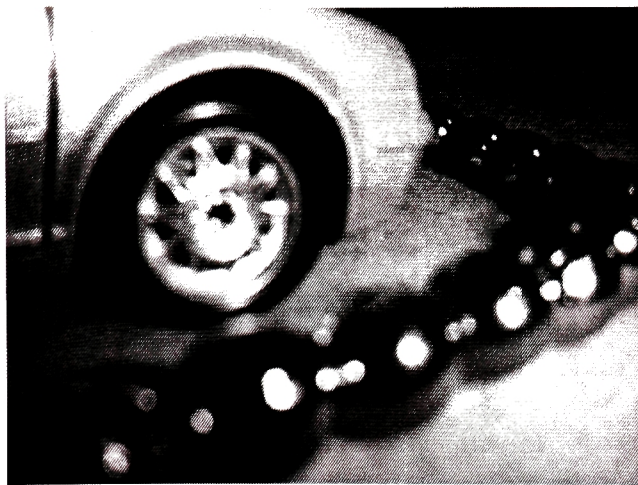



Figure 2: Autistic children sometimes display unusual interest in an object, like beads on a string or the spinning wheels of a toy car (Taken by Sophie Kuziora).

development. The program uses both reinforcement and stimulus techniques, while creating an integrated and consistent learning environment at home and at school along with peer-based learning. This program is highly individualized, taking into account individual strengths, interests and needs as well as cultural and social backgrounds (Erba, 2000). Reported outcomes for the LEAP program include increases in language, cognitive and motor skills; however, few studies have been performed to validate such claims (Hoyson, Jamieson, & Strain, 1984).

THE ROLE OF PRIMARY HEALTH CARE PROVIDERS

Parents of autistic children frequently report that they felt something was wrong with their child, with difficulties centered on abnormal social development by about 18 months of age (Howlin & Moore, 1997; Howlin & Asgharian, 1999). Diagnosis rarely takes place this early, despite evidence that there are often clear symptoms of autism before one year of age (Werner, Dawson, Osterling, & Dinno, 2000; Baranek, 1999). Health care professionals that fail to make a diagnosis risk limiting the child's access to early intervention. However, there is also a hesitancy to provide a diagnosis due to the perceived risk of accusation or exaggeration (Dossetor, 2005). Dr. Kennedy and colleagues (2004) addressed the issue of differences between the knowledge of health care professionals and subsequent behaviour as it is related to diagnosis of autism. Some family medicine residents attribute their hesitation of giving a positive diagnosis or referral to the stigmas attached to the word "autism", as well as their own lack of knowledge about the disorder (Kennedy, Regehr, Rosenfield, Roberts, & Lingard, 2004). Even when patient practice guidelines are available, they are not always followed due to barriers that include lack of awareness, familiarity, agreement with the guidelines, and outcome expectancy (Cabana, Rand, Powe, Wu, Wilson, Abboud, et al, 1999). Despite the difficulties in reliably diagnosing autism in early childhood, experts agree that early intervention is an important predictor of outcome. Early detection provides an opportunity for intervention to minimize or prevent the symptoms of autism (Sigman, Dijamco, Gratier, & Rozga, 2004).

Family physicians and primary health care providers need to become increasingly aware of the early warning signs of autism in order to provide optimal care for children who are at risk. Physicians, teachers, and other health professionals who suspect the presence of typical autistic tendencies should refer the child to a specialized assessment, with clinicians trained in the diagnosis of autism. It is through early diagnosis that the opportunity for intervention, a better outlook for the child, and ultimately, a happier family are made possible. 

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