



Sven Bölte · Joachim Hallmayer (Editors)

# Autism Spectrum Conditions

**FAQs on Autism, Asperger Syndrome,  
and Atypical Autism Answered by International Experts**



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**Sven Bölte  
Joachim Hallmayer  
(Editors)**



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# Preface

Autism spectrum conditions (ASC) are characterized by limited social reciprocity and thus constitute a substantial public health problem in a world of high social demands. It is now recognized that about 1% of the general population is functionally affected by an ASC. Thus, ASC are not rare conditions and they pose challenges to society, experts, families, and people with ASC themselves. Awareness of and knowledge about ASC has increased tremendously in recent years. Nevertheless, the roots of ASC remain enigmatic, effective intervention options are limited, and outcome as well as quality of life are still low for many people on the spectrum. In addition, many myths and misconceptions about ASC survive.

Countless books have been published on ASC. This volume is novel and unique in bringing together several qualities. It combines 78 contributions from 66 internationally leading clinical and research experts and autism organizations from North America and Europe, thus providing a broad, state-of-the-art view on research and practice in dealing with ASC. The book has a FAQ format that communicates current knowledge on ASC in a well-structured and concise way. It has been written with the clear aim of making it a readable and informative resource for laypersons as well as experts. Organized in 10 sections, this book includes everything essential you need to know about the symptoms, diagnoses, frequency, possible causes, treatments, education issues, and outcomes in ASC.

Our sincerest thanks to all who made this book possible. And, as always, our best wishes to all people with ASC and their families.

*Sven Bölte*, Karolinska Institutet, Stockholm, Sweden  
*Joachim Hallmayer*, Stanford University, CA, USA



# Section 1

# Characteristics, Identification, and Diagnosis



# Q1 What Are ASC?

Peter Szatmari

The ASC are a group of neurodevelopmental disorders characterized by impairments across the so-called “autistic triad.” These include difficulties with reciprocal social interaction and with verbal and nonverbal communication as well as a preference for repetitive, stereotyped activities, patterns of behaviors, and interests. The age of onset is always prior to 36 months, which is an important characteristic that differentiates ASC from other neuropsychiatric or developmental disorders. The ASC are more common than previously thought and have a prevalence of between 0.5 and 1% (Autism and Developmental Disabilities Monitoring Network, 2009). ASC are treatable using strategies that are intensive, based on behavioral principles, and focus on improving IQ, language, social communication, and play (National Research Council, 2001).

Leo Kanner, a child psychiatrist at Johns Hopkins University, first described 11 children with what he called “infantile autism” in 1944 (Kanner, 1968). These children had difficulties with social communication and presented with patterns of restricted interests and insistence on sameness. However, over the ensuing years, the meaning of the term “autism” became somewhat diluted until more precision was introduced in 1980 with the publication of DSM-III (APA, 1980). In that classification system, the term “pervasive developmental disorders (PDD)” was introduced to describe a group of conditions, all of which are characterized by the autistic triad. PDD included autism, as well as other subtypes. In DSM-IV, there are five subtypes of PDD: autistic disorder, Asperger disorder, PDD not otherwise specified (PDD-NOS), childhood disintegrative disorder, and Rett disorder (APA, 1994).

Asperger disorder is diagnosed in children with ASC who do not present with clinically significant cognitive or language delay. This is usually operationalized as IQ in the typical range and speaking in phrases (spontaneously and functionally) by 3 years of age. In addition, there is a hierarchical relationship between autism and Asperger disorder in that, if a child meets criteria for the former, he cannot meet criteria for the latter (APA, 1994). Children with Asperger disorder, therefore, are usually higher functioning and verbally fluent but with poor conversation skills, and have poor social skills as well as a circumscribed range of interests that are pursued to an unusual degree of intensity and usually alone (rather than as part of a social group) (see Szatmari, 2000; Frith, 2004).

Children with PDD-NOS present with ASC symptoms but do not meet the criteria for either autism or Asperger disorder. It was intended to be used as a leftover category and applied to only a small number of children. This has not turned out to be the case as community studies indicate that a substantial proportion of children with ASC are diagnosed with PDD-NOS, not autism or Asperger disorder (Chakrabarti & Fombonne 2001). Children with PDD-NOS often receive that diagnosis because they are too young to demonstrate many repetitive behaviors, too low functioning to be able to assess communication impairments in excess of developmental level, or too high functioning (or older) and so not demonstrating many impairments in social communication (Walker et al., 2004). Nevertheless, the ability of clinicians to distinguish between PDD-NOS and autism is quite poor (Mahoney et al., 1998) and often results from a lack of information available to the clinician, either from the parent, from direct observation, or from a third source, such as a teacher.

The term PDD has slowly been supplanted by the more commonly used term “autism spectrum disorder” or “autism spectrum condition.” This arises from the observation that it is very difficult to differentiate the different subtypes of PDD and it appears, based on current evidence, that the distinctions among these subtypes are not only difficult to make but carry little useful clinical information (for a review see Macintosh & Dissanayake, 2004; Van Lang et al., 2006). The American Psychiatric Association Neurodevelopmental Disorders Work Group developing DSM-V proposes a single term, autism spectrum disorder, to cover all 5 current PDD subtypes (see [www.dsm5.org](http://www.dsm5.org)).

It has been clear since the 1970s that autism is caused by biological factors affecting brain development. The first indication of this was the observation that there are approximately 4 boys to every girl with the disorder and that a substantial minority of children with autism also have neurological problems, such as epilepsy, or other medical disorders (Spence & Schneider, 2009). More recently, it has become apparent that genetic factors play an important role in ASC (Weiss, 2009). Approximately 10% of children with ASC have a comorbid single gene disorder, such as fragile X syndrome or tuberous sclerosis. Perhaps as many as another 10% have chromosomal abnormalities (i.e., large duplications, translocations, insertions, and/or deletions) that can be seen under the microscope with routine karyotyping. More subtle genomic or chromosomal abnormalities (known as “copy number variants”) are currently being discovered and may apply to a further proportion of individuals with ASC (Merikangas, Corvin, & Gallagher, 2009). Whether more common genetic factors cause ASC is currently not known; however, this is an active area of investigation.

Although there has been quite a bit of research into possible environmental factors that may cause ASC, this has not led to many positive findings (Newschaffer et al., 2002). It is unlikely that environmental factors cause autism in the absence of genetic susceptibility. Perhaps the strongest evidence for environmental etiology comes from maternal use of anticonvulsants (i.e., a mother taking valproic acid during pregnancy), which is associated with an increase in births with ASC (Ornoy, 2009). Extreme prematurity (Limperopoulos, 2009), *in vitro* fertilization (Hvidjtorn et al., 2009), and advanced parental age (King, Fountain, Dakhlallah, & Bearman, 2009), are also factors associated with envi-



ronmental variables that may be associated with ASC, but these data need to be replicated and more strongly confirmed.

It is also quite clear that a number of biological factors that have been promoted in the media and actively investigated are not involved in the etiology of ASC. These include vaccinations, unusual diet, infections of various sorts, and the so-called “leaky gut.”

While it is well known that the majority of individuals with ASC have a poor outcome (Howlin Goode, Hutton, & Rutter, 2004), this picture may shift as the diagnostic criteria broaden to include more higher-functioning individuals, and as universally accessible, early intensive behavioral interventions become available. It is true that recovery can occur in a small number of cases, perhaps as many as 5% (Helt et al., 2008). Most children with ASC show slow but steady improvement in autistic behaviors and in social and communication skills (Bennett et al., 2008). It must be emphasized, though, that outcome is extremely variable, and some individuals with ASC need an extraordinary amount of support, not only in adolescence but also in adulthood (Billstedt, Gillberg, & Gillberg, 2007; Hofvander et al., 2009). The strongest predictors of a positive outcome are early language and average cognitive skills (Szatmari, Bryson, Boyle, Streiner, & Duku, 2003). However, even individuals with these positive prognostic factors may have a poor outcome.

It is true that we have learned a great deal about ASC in the last 20 years. The profession used to think of autism as a psychotic disorder caused by poor maternal-child attachment patterns, with a very poor outcome, and as basically untreatable. This picture has changed dramatically, and it is anticipated that the next 20 years of research will bring even more positive advances. A key challenge will be bringing down the age of diagnosis so that children are able to access intervention earlier than they currently do. At present, the average age at which a child is diagnosed in North America is between 4 and 5 years of age (Mandell, Novak, & Zubritsky, 2005). However, parents first become concerned about the development of their children at around 18 months of age (Zwaigenbaum, 2010). This gap indicates that children are not receiving any services in the meantime and may not benefit from that window of opportunity. Another exciting development is the possibility that, as we gain a greater understanding of the genetic factors that contribute to ASC, more targeted biomedical treatments can be developed that may perhaps reduce some of the cognitive-behavioral changes. Such possibilities are currently being explored in clinical trials and in animal models of fragile X syndrome (Hagerman et al., 2009) and Rett syndrome (Tropea et al., 2009). It is not inconceivable that such advances may be applied to ASC as well.

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## What Are the Most Common Characteristics of ASC?

Peter Szatmari

Clinical experience has characterized ASC as a set of signs and symptoms that fall under three main categories: Impairments in reciprocal social interaction, impairments in verbal and nonverbal communication, and a pattern of repetitive, stereotyped behaviors, interests, and activities. While this has served as a very useful guidepost, more detailed measurement studies have somewhat changed this clinical picture (Snow, Lecavalier, & Houts, 2008; Lecavalier, Gadow, DeVincent, Houts, & Edwards, M., 2009). It is now generally thought that there are two main dimensions along which ASC children are placed (Georgiades et al., 2007). One is impairments in social communication, and the other is repetitive, stereotyped, behaviors. It has been shown, through measurement studies, that such children also have independent difficulties either with general cognitive skills or with more specific skills in one or two areas (Szatmari et al., 2002; Matson & Shoemaker, 2009; Fernell et al., 2010). Therefore, intellectual disability (ID) is seen in roughly 50% of individuals with autism and in 25% of those with ASC (Fombonne, 2005). Among those who do not have ID, some form of specific learning disability, either in reading, spelling, or mathematics or a nonverbal learning disability, is almost universally present (Jones et al., 2009).

Another universal characteristic of the disorder is that onset is prior to 36 months of age (American Psychiatric Association, 2000). Until recently, there has been much debate as to when children with ASC first begin to present with their difficulties. Retrospective reports, early videotapes, and parental reports have all suggested that, between 12 and 18 months of age, if not earlier, signs of difficulties in social communication, visual attention, and motor difficulties begin to present themselves (Rogers, 2009). Infant sibling studies (i.e., where the infant sibling of a child with autism is followed longitudinally from birth to 36 months of age) have provided much richer data on this topic (Zwaigenbaum et al., 2009). It now appears as though most children with ASC develop normally until 6 months of age, and there is then a slowing down in their social communication, as well as in cognitive and language skills over the following 12–18 months (Zwaigenbaum, 2010). Therefore, the most obvious time at which

difficulties begin to appear is between 12 and 18 months of age, whereas the full syndrome is not present until 24–36 months of age.

Difficulties in social communication are then an essential part of ASC. Such problems can be broken down into different subdomains, including social reciprocity, eye contact, smiling in response to another, directing attention, greeting, empathy, and offering and asking for comfort. Imitation, peer relationships, and social initiative with peers and adults are other key aspects of social interaction (Anderson, Oti, Lord, & Welch, 2009). Communication problems are seen in virtually all children with ASC (Fodstad, Matson, Hess, & Neal, 2009). These might include reduced babbling, speech delay (either of phrases or single words), and unusual speech patterns, such as echolalia, pronoun reversal, and making up words. There are significant difficulties in the pragmatics of communication, such as poor conversation skills, unusual inflection, and little expression of emotion in speech. In addition to these verbal communication difficulties, almost all children with ASD have nonverbal communication difficulties: They do not use gestures, will not point at objects close by or far away, and won't nod or shake their head.

There are, finally, many examples of repetitive stereotyped behaviors (Szatmari et al., 2006; Lam, Bodfish, & Piven, 2008; Richler, Huerta, Bishop, & Lord, 2010). These include preoccupations with parts of objects (e.g., playing with a toy but not in the way in which it was meant to be played). Fascinations with concrete sensory experiences, such as smell, sight, touch, and taste, are also common. Older children with ASC often have extremely intense, circumscribed interests, such as specific cartoons and shows on TV such as the Weather Channel, and unusual hobbies and interests, such as listening to thunderstorms or reading science fiction. Repetitive motor movements, such as flicking one's fingers in front of the eyes, flapping one's hands, or running back and forth in a certain pattern, are also common, particularly among younger children with ASC. Rituals, such as an insistence on doing things in a particular way, are also not uncommon and should be differentiated from resistance to change, which signifies children not wanting to go from one activity to another, particularly if they are interested in that activity. This is a common problem in young children in general and is often confused with ASC.

A lot of research has gone into trying to understand how to characterize cognitive delays so often seen in children with ASC. Delays in expressive and receptive language are common, but children with Asperger syndrome do not show such language problems (Williams, Botting, & Boucher, 2008). Indeed, Asperger syndrome is characterized by an absence of clinically significant cognitive and language delay (Woodbury-Smith, Klin, & Volkmar, 2005). Perhaps a more parsimonious explanation of cognitive difficulty that virtually all ASC children share is a difficulty with abstract, metaphorical thinking, as opposed to more rote thinking or concrete understanding, which they are able to do well (Williams, Goldstein, Minshew, 2006). The more complex the cognitive operation, the more abstract, the more metaphorical, the more difficulty the ASC child will have with learning that concept, with understanding it, and with being able to perform certain roles.

Another active area of research has focused on trying to understand the fundamental deficit associated with ASC. Various theories have been proposed and



studies undertaken to test the extent to which difficulties in joint attention, theory of mind (ToM), executive dysfunction, weak central coherence, and shifting and disengaging attention might be a unifying paradigm to understand children with ASC (Happé & Frith, 2006; Martin & McDonald, 2003; Miller, 2006; Landry & Bryson, 2004)). It now seems apparent that there will not be a single explanation or a single cognitive model for the disorder, but rather that different components of the condition might be associated with different cognitive models (Happé & Ronald, 2008). For example, a lack of ToM may be a good explanation for some of the social difficulties that children with ASC experience but some other model would need to be proposed for the preference of repetitive, stereotyped behaviors, such as weak central coherence (Mandy & Skuse, 2008).

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## What Are the Differences (and Similarities) Between Autism, Asperger Syndrome, and PDD-NOS?

Fred R. Volkmar

This is an excellent question with a short and then a longer answer. The short answer is that, as currently defined in DSM-IV-TR (American Psychiatric Association, 2000), autism is defined on the basis of characteristic difficulties in the areas of social interaction, communication and play and restricted/unusual interests and behaviors. Asperger disorder differs from autism in that early language ability is relatively preserved, while pervasive developmental disorder not otherwise specified (PDD-NOS) is a category used for individuals with problems suggestive of autism but not meeting full criteria for that condition. All these conditions are currently recognized as members of the pervasive developmental disorder (PDD) class – a term that was coined in 1980 when autism was first included as an officially recognized diagnosis. In practical terms, PDD means the same thing as autism spectrum disorder (Volkmar & Klin, 2005).

Of these disorders, autism has been far and away the most frequently studied and, somewhat paradoxically, the least studied disorder, PDD-NOS, is the most frequent. This disparity in research reflects an understandable focus on the more “prototypic” disorder. However, interest in the broader spectrum of autistic-like conditions has increased dramatically in recent years, and it likely includes a number of subtypes, though none of these are, as yet, officially recognized (Towbin, 2005).

The condition termed “Asperger syndrome” was first described in 1944, the year after Kanner’s first description of autism. In the initial cases described by Hans Asperger, the children (all boys) had remarkable trouble in social interaction and weak motor skills, but good verbal abilities. They also exhibited circumscribed interest in various topics (e.g., trains, rocks, the weather, American gangsters), and this interest interfered with their abilities to learn other skills (McPartland & Klin, 2006). The issue of whether or not Asperger syndrome is truly different from autism or represents, essentially, more cognitively able