

Understanding Autism Spectrum Disorders (ASDs): An Introduction

What are autism spectrum disorders (ASDs)?

Autism spectrum disorders (ASDs) are a group of related brain-based disorders that affect a child's behavior, communication, and social skills. These disorders include autistic disorder, Asperger syndrome, and pervasive developmental disorder—not otherwise specified (PDD-NOS). They are defined by the number and severity of the symptoms.

Because most children with ASDs will master early motor skills such as sitting, crawling, and walking on time, parents may not initially notice delays in social and communication skills. Looking back, many parents can recall early differences in interaction and communication.

ASDs are developmental disorders whose symptoms may change with maturation and intervention. While infrequent, some children improve so much that they no longer can be considered to have an ASD. Most of these children will have other developmental, learning, language, or behavioral diagnoses.

The sooner an ASD is identified, the sooner an intervention program directed at core symptoms of autism can start. Each child with autism has different needs. The intervention that helps one child may not be as helpful for another. Research shows that starting an intervention program as soon as possible can improve outcomes for many children with ASDs, so children can and should be referred for diagnosis and early intervention (EI) as soon as the ASD symptoms are noted.

The following are excerpts from the American Academy of Pediatrics' (AAP) booklet *Understanding Autism Spectrum Disorders (ASDs)*.

How common are ASDs?

ASDs affect an estimated 1 out of every 88 children. The number of children reported to have an ASD has increased since the early 1990s for unclear reasons. The increase could be caused by many factors. Many families are more aware of ASDs. Pediatricians are doing more screening for ASDs. Also, there have been changes in how ASDs have been defined and diagnosed. In the past, only children with the most severe ASD symptoms were diagnosed. Now children with milder symptoms are being identified and referred to intervention and educational programs. Boys with ASDs typically outnumber girls by about 4 to 1.

What are the symptoms of ASDs?

No 2 children with an ASD have exactly the same symptoms. There are standard criteria for the diagnosis of ASDs. However, the number and severity of symptoms can vary greatly. The following are examples of how a child with an ASD may act:

Social differences

- Doesn't keep eye contact or makes very little eye contact
- Doesn't respond to a parent's smile or other facial expressions
- Doesn't look at objects or events a parent is looking at or pointing to
- Doesn't point to objects or events to get a parent to look at them
- Doesn't bring objects of personal interest to show to a parent
- Doesn't often have appropriate facial expressions
- Unable to perceive what others might be thinking or feeling by looking at their facial expressions

- Doesn't show concern (empathy) for others
- Unable to make friends or uninterested in making friends

Communication differences

- Doesn't point at things to indicate needs or share things with others
- Doesn't say single words by 16 months
- Repeats exactly what others say without understanding the meaning (often called *parroting* or *echoing*)
- Doesn't respond to name being called but does respond to other sounds (like a car horn or a cat's meow)
- Refers to self as "you" and others as "I," and may mix up pronouns
- Often doesn't seem to want to communicate
- Doesn't start or can't continue a conversation
- Doesn't use toys or other objects to represent people or real life in pretend play
- May have a good rote memory, especially for numbers, letters, songs, TV jingles, or a specific topic
- May lose language or other social milestones, usually between the ages of 15 and 24 months (often called *regression*)

Behavioral differences (repetitive and obsessive behaviors)

- Rocks, spins, sways, twirls fingers, walks on toes for a long time, or flaps hands (stereotypic behavior)
- Likes routines, order, and rituals; has difficulty with change
- Obsessed with a few or unusual activities, doing them repeatedly during the day
- Plays with parts of toys instead of the whole toy (for example, spinning the wheels of a toy truck)
- Doesn't seem to feel pain
- May be very sensitive or not sensitive at all to smells, sounds, lights, textures, and touch
- Unusual use of vision or gaze—looks at objects from unusual angles

What are the different types of ASDs?

The symptoms that define ASDs are described in a book called the *Diagnostic and Statistical Manual of Mental Disorders (DSM)*. This manual lists agreed-on definitions for behavioral, developmental, and psychiatric disorders. Shortly after publication of this booklet, a new version of the DSM will be published. It will eliminate the subcategories of autistic disorder, Asperger syndrome, and PDD-NOS that were defined by the fourth edition, text revision of *DSM (DSM-IV-TR)* and consider all diagnosed individuals to have ASDs. The *International Classification of Diseases, 10th Revision (ICD-10)* will continue to use the *DSM-IV-TR* categories within ASDs that are described in this booklet. In the future, children diagnosed with Asperger syndrome might be considered to have high-functioning autism or ASDs with typical intellectual abilities.

Autistic disorder

Children with autistic disorder have a pattern of core problems with language and communication; problems relating to others socially; and unusual or repetitive

behaviors. While social symptoms are usually present in the first year of life (although they may be subtle), language problems are more obvious in the second year, and repetitive behaviors may appear even later.

Many of these children will have intellectual deficits; others might *appear* to have deficits when, in fact, scores on intelligence tests are low because of lack of cooperation. Others may have normal scores on intelligence tests, yet they may have trouble with abstract and real-life reasoning. Children with autism are considered *high functioning* when their intelligence is in the normal range.

Asperger syndrome

Asperger syndrome (also called Asperger disorder) is usually not diagnosed until preschool age or later. This is because early speech development, especially language and sentence structure, is relatively normal. Sometimes children with Asperger syndrome speak in an odd way. Some children may speak in only one tone of voice (monotone) without raising or decreasing the pitch of their voice. Other children may speak in language above what would be expected for their age like “little professors.” They may make little eye contact while talking and may have trouble maintaining a back-and-forth conversation. They may focus on 1 or 2 topics and will talk about these topics whether or not the listener is interested. Children with Asperger syndrome often interpret language literally and may have particular trouble with humor, teasing, and figures of speech. Many may also have problems with motor coordination. Intelligence is normal. Some experts do not consider this a separate disorder from high-functioning autism.

Pervasive developmental disorder—not otherwise specified

Children with PDD-NOS (sometimes described as *atypical autism*) show some signs of autism or other PDD but do not have sufficient number or intensity of symptoms to be diagnosed with autistic disorder or Asperger syndrome. Though children with PDD-NOS may have fewer overall symptoms, the symptoms can still be challenging.

What causes ASDs?

Many factors may lead to symptoms of ASDs. There is a genetic predisposition to ASDs. This was initially determined by twin and other family studies. Although many chromosomal and gene abnormalities have been associated with symptoms of ASDs, none of these are present in all affected children. If a family already has a child diagnosed with an ASD, the chances that siblings might also have some form of ASD are at least 5 to 10 times higher than in the general population. Environmental factors are likely to play a secondary role in some children with ASDs, but what these agents are and when they affect development is not yet known.

Studies have shown that the relatives of children with autism are more likely to have some symptoms similar to those seen in children with ASDs. These difficulties may include language delays, learning disabilities, anxiety, attention-deficit/hyperactivity disorder (ADHD), or moodiness.

ASDs may occur more often in children with certain medical conditions, such as fragile X syndrome, tuberous sclerosis, Down syndrome, and other genetic disorders. Most children with ASDs do not have these or other specific genetic conditions. Symptoms of ASDs may be more common in babies born prematurely. Considering whether a child with an ASD has another genetic condition is an important part of the initial evaluation.

What are the early signs of ASDs?

Many children with ASDs may show developmental differences throughout their infancy, especially in social and language skills. Because they usually sit, crawl, and walk on time, more subtle differences in the development of gesture, pretend play,

Is there a link between the MMR vaccine and ASDs?

There has been a great deal of attention in the media around vaccines as a cause of autism. Current scientific evidence does not support a link between the measles-mumps-rubella (MMR) vaccine or any combination of vaccines and autism spectrum disorders (ASDs). In fact, the original research article that suggested a link between the MMR vaccine and autism has been officially retracted (that is, removed permanently) because of serious flaws in how the research was conducted. There also is no scientific proof to support a link between thimerosal (a mercury containing preservative) and ASDs. In any event, almost all vaccines in the recommended immunization schedules administered to children in the United States no longer contain mercury. Vaccines used for immunization of large numbers of people, like influenza vaccine, may still contain very small amounts of thimerosal. Families who remain concerned about this should discuss their concerns with their pediatrician.

and social language often go unnoticed by families and doctors. In addition to delays in spoken language, families may notice differences in interaction and gesture.

Delay or lack of joint attention

One of the most important developmental differences between children with ASDs and other children is a delay or lack of *joint attention*. Joint attention is looking back and forth between an object or event and another person and connecting with that person. It is a building block for later social and communication skills. Engaging in many back-and-forth social interactions, such as exchanging a lot of emotional expressions, sounds, and other gestures, is called *reciprocal social interaction*. Delays in joint attention skills are found in most children with ASDs and rarely seen in children with other types of developmental problems. Thus, joint attention deficits are thought to be among the most characteristic deficits of ASDs. There are several stages of joint attention. Children with ASDs usually show delays or absent skills at every stage.

Subtle milestones in use and understanding of gestures occur at the following times:

- At about 10 to 12 months of age, most typically developing children will immediately look in the direction of an object to which a parent is pointing. They will then look back at the parent and mimic the parent’s expression, usually a smile. Children with ASDs will often ignore the parent. This often causes parents to worry about their child’s hearing.
- Most children are able to point to out-of-reach objects that they want by 12 to 14 months. The child with an ASD may instead take a parent’s hand and lead the parent to the object without making eye contact. Sometimes the child may even place the parent’s hand on the object itself.
- By 14 to 16 months of age, most children point at objects they find interesting. Children will look back and forth between an object and a parent to make sure that the parent is tuned in to what they are looking at. The child with an ASD only will point to an object because he wants the parent to get it for him, not because he wants the parent to enjoy looking at an object together.

Language delays

Almost all children with ASDs show delays in nonverbal communication and spoken language. They may have words that they use to label things but never request things. They may have unusual words for their overall language level, such as saying letters or numbers when they do not yet have names they use for family members. Most young children go through a phase where they repeat what they hear. Children with ASDs may repeat for a longer period and repeat

movies or conversations with the tone of voice in which they heard them. Those children later diagnosed with Asperger syndrome will seem to have met language milestones during the toddler years, but use of language may be abnormal or overly sophisticated, or mature (little professors).

Regression in developmental milestones

About 25% of children will seem to have normal or near-normal development until about 18 months of age, after which they will gradually or suddenly stop using words they once had and become more withdrawn. Some families will recall subtle differences that might have been present prior to the regression, such as the child not turning to her name.

How is the diagnosis made?

Diagnosis of ASDs can be complicated for a number of reasons. There are no specific medical laboratory tests to diagnose an ASD, so doctors must rely on information from parents and on what can be observed during well-child checkups. The condition is complex, and symptoms are different for each child. This is why the AAP recommends that there be screening for ASDs at specific well-child checkups as well as ongoing surveillance in the course of well-child care.

Evaluation

When an ASD is suspected as a cause of language and social delays, the child should be referred for *both* EI services and a full evaluation to determine if an ASD is the proper diagnosis. The evaluation may be done by a doctor or psychologist who has expertise in the diagnosis of ASDs or, preferably, by a team of specialists that may include developmental pediatricians, child neurologists, child psychiatrists, psychologists, speech or language pathologists, occupational or physical therapists, educators, and social workers. Testing of developmental domains may occur through EI programs or the school system.

Typically, an evaluation will include the following:

- Careful observation of play and child-caregiver interactions.
- Detailed history and physical examination.
- Developmental assessment of all skills (motor, language, social, self-help, cognitive). ASDs are suspected when the child's social and language functioning are significantly more impaired than the overall level of motor, adaptive, and cognitive skills.
- Hearing test. All children with any speech delays or those suspected of having ASDs should have their hearing formally tested.
- Language evaluation that provides standardized scores of expressive language (including speech) and receptive language, as well as an evaluation of pragmatic language (social use of language) and articulation (pronunciation).

Diagnosis of an ASD is made by applying DSM criteria using all the information collected by history, observation, and testing. (See "What are the different types of ASDs?")

Medical tests

ASDs may be associated with a known syndrome or medical condition. Newer, more sensitive tests have determined an underlying cause of an ASD in many more children than was previously thought. Laboratory tests may be indicated to rule out other possible medical conditions that could cause autism symptoms based on the child's history and physical examination. If indicated, the child may be referred to other specialists, such as a geneticist or a pediatric neurologist, to help diagnose medical conditions that might cause or be associated with symptoms of ASDs.

- **Genetic tests.** It is recommended that families be offered genetic testing, such as cytogenetic microarray testing. At present, up to 10% to 20% of

children with ASDs have abnormalities of their chromosomes identified using cytogenetic microarray testing. Fragile X syndrome may be present in up to 2% of boys with ASDs, so fragile X testing should also be considered. Testing girls with ASD symptoms for Rett syndrome may be discussed depending on the child's history and physical examination. Genetic testing should be strongly considered if a child has atypical physical features or developmental delays, or if there is a family history of fragile X syndrome or intellectual disability (formerly known as mental retardation) of unknown cause. Other genetic tests may be needed in certain cases. Recommendations for genetic testing may change as new tests are developed.

- **Lead test.** Lead screening is an important component of primary care. A lead level should be performed when a child lives in a high-risk environment, such as older buildings, or continues to put things in his mouth.
- **Other tests.** Based on the child's medical history and physical examination, an electroencephalogram (EEG), a magnetic resonance imaging (MRI) scan, or tests for metabolic disorders may be ordered. Children with ASDs may be picky eaters, so your child's pediatrician may recommend looking for evidence of iron or vitamin deficiencies (especially vitamin D).

Note: There is not enough clinical evidence to recommend any of the following tests specifically for autism: hair analysis, routine measurement of multiple vitamin or nutrient levels, intestinal permeability studies, stool analysis, urinary peptides, or measurement of mercury or other heavy metals.

Living with ASDs

There are many different strategies and techniques to help children with ASDs learn to interact with others and acquire new skills that may help them talk, play, participate in school, and care for their needs.

According to an expert panel writing for the National Academy of Sciences, effective educational programs designed for children with ASDs from birth to 8 years of age should

- **Offer choices.** The program should offer a variety of behavioral, language, social, play, and cognitive strategies that are individualized to the child. If possible, the child should also receive direct speech, occupational, and physical therapies according to individual need.
- **Have clear goals.** An individualized plan should include specific, observable, and measurable goals and objectives in each developmental and behavioral area of intervention.
- **Be intense.** The program should be intense, with a goal of 20 to 25 hours of planned intervention or instruction per week. It should be given yearround. The majority of children benefit from a staffing ratio of 1:1 or 1:2 with an adult in initial interventions.
- **Encourage parents to be fully involved.** Siblings and peers should also be included in the program. Children often learn best by modeling typically developing children in inclusive settings. The family should have support from the therapy team so it can promote social skills, functional communication, and appropriate behavior at home.
- **Take place in everyday settings.** To promote generalization of newly acquired skills, interventions should take place in everyday settings. Playing and learning with children without ASDs may help children with ASDs learn social and language skills.
- **Address behavior problems.** A functional analysis of behavior should be done when there are behavior problems. Information gained should be used to design a behavior management plan. The family should be involved so it can work on the child's behavioral needs too.
- **Monitor progress often.** If goals and objectives are not being met in a reasonable amount of time, the program should be evaluated and revised as needed.

The types and quality of services may vary depending on where a family lives. Efforts are being made nationally to increase funding and training, so professionals can meet the needs of children with ASDs in medical and educational settings. While resources vary among communities, a combination of parent and professional interventions can improve the development of children with ASDs.

Children should be referred to an appropriate program as soon as a delay is suspected. Parents should not wait for a definitive diagnosis of an ASD because this may take quite some time. For example, speech therapy evaluation and treatment should be started as soon as a communication delay is identified. Once an ASD or another developmental disability is definitively diagnosed, the specific program can be changed to best meet the needs of the child and family. Keep in mind that diagnosis can be an ongoing process as additional signs and symptoms become noticeable or others improve.

Although *all* children with ASDs will need educational services and most will need therapy and behavioral interventions, only certain children may need medicine. Medicine may be used to help decrease behaviors that could interfere with learning or interaction with others, such as aggression, obsessions, or hyperactivity.

Parents are encouraged to learn as much as they can about all the different treatments available. Treatment should focus on supporting the child to succeed in the real world.

The future

Children with ASDs are affected by many factors that will shape their future. Overall, the long-term outcomes of children with ASDs have been improving. In general, the sooner an ASD is identified, the sooner appropriate intervention programs can begin. While we believe that children make significant developmental gains with early and intense intervention, some children may make slow progress depending on their intelligence, the severity of their ASD symptoms, and whether they have associated medical problems such as seizures or significant behavioral disorders.

Children with intelligence in the typical range and milder symptoms of ASDs are more likely to complete academic programs at school and have greater success in community employment as adults.

The goal of all parents, whether their child is typical or has a disability, is to help their child reach his full potential with the help of all available resources.

Resources

Books

Attwood T. *The Complete Guide to Asperger's Syndrome*. London, England: Jessica Kingsley Publishers; 2007

Baker J. *Preparing for Life: The Complete Guide for Transitioning to Adulthood for those with Autism and Asperger's Syndrome*. Arlington, TX: Future Horizons; 2005

Bashe PR, Kirby BL. *The OASIS Guide to Asperger Syndrome: Advice, Support, Insight, and Inspiration*. Rev ed. New York, NY: Crown Publishers; 2005

Coplan J. *Making Sense of Autistic Spectrum Disorders: Create the Brightest Future for Your Child With the Best Treatment Options*. New York, NY: Bantam Books; 2010

Glasberg BA. *Functional Behavior Assessment for People with Autism: Making Sense of Seemingly Senseless Behavior*. Bethesda, MD: Woodbine House; 2006

Gray C. *The New Social Story Book*. Arlington, TX: Future Horizons; 2010

Harris SL, Weiss MJ. *Right from the Start: Behavioral Intervention for Young Children with Autism*. 2nd ed. Bethesda, MD: Woodbine House; 2007

Hodgdon LA. *Visual Strategies for Improving Communication: Practical Supports for School and Home*. Troy, MI: QuirkRoberts Publishing; 2000

Kluth P, Shouse J. *The Autism Checklist: A Practical Reference for Parents and Teachers*. San Francisco, CA: Jossey-Bass; 2009

McClannahan LE, Krantz PJ. *Activity Schedules for Children with Autism: Teaching Independent Behavior*. 2nd ed. Bethesda, MD: Woodbine House; 2010

Moor J. *Playing, Laughing and Learning with Children on the Autism Spectrum: A Practical Resource of Play Ideas for Parents and Carers*. 2nd ed. London, England: Jessica Kingsley Publishers; 2008

Notbohm E. *Ten Things Every Child With Autism Wishes You Knew*. Arlington, TX: Future Horizons; 2005

Notbohm E, Zysik V. 1001 *Great Ideas for Teaching & Raising Children with Autism or Asperger's*. Rev ed. Arlington, TX: Future Horizons; 2010

Simpson RL. *Autism Spectrum Disorders: Interventions and Treatments for Children and Youth*. Thousand Oaks, CA: Corwin Press; 2005

Thompson T. *Making Sense of Autism*. Baltimore, MD: Paul H. Brookes Publishing Co; 2007

Volkmar FR, Wiesner LA. *A Practical Guide to Autism: What Every Parent, Family Member, and Teacher Needs to Know*. Hoboken, NJ: John Wiley & Sons; 2009

Wiseman ND. *Could It Be Autism? A Parent's Guide to the First Signs and Next Steps*. New York, NY: Random House; 2006

Web sites

American Academy of Pediatrics

www.aap.org

www.aap.org/autism (Council on Children With Disabilities Autism Subcommittee)

www.HealthyChildren.org (official AAP Web site for parents)

www.medicalhomeinfo.org (National Center for Medical Home Implementation)

Autism Science Foundation

www.autismsciencefoundation.org

Autism Speaks

www.autismspeaks.org

Centers for Disease Control and Prevention

www.cdc.gov/autism

Easter Seals

www.easterseals.com/autism

National Institute of Mental Health

www.nimh.nih.gov

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The information contained in this publication should not be used as a substitute for the medical care and advice of your pediatrician. There may be variations in treatment that your pediatrician may recommend based on individual facts and circumstances.

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