

CHAPTER 1

What is an Autism Spectrum Disorder?

“

On April 23, 1995, I came back from a diagnostic appointment at the university with Coty in tow. I just found out that my baby boy, my sweet, precious little one, has autism. I am supposed to get the diagnosis confirmed by a pediatric neurologist. I am devastated, confused and uncertain of "our" future.

”

What Is an Autism Spectrum Disorder?

Autism Spectrum Disorder refers to a developmental disability with a neurological basis that affects an individual's verbal and nonverbal communication and social interaction. Each individual with ASD has unique abilities, symptoms and challenges.

ASD is a spectrum disorder and the symptoms and characteristics of ASD can range from mild to severe. Individuals may exhibit any combination of characteristics in any degree of severity. This means that two children with the same diagnosis can act very differently from one another and has varying skills and needs. Children with a diagnosis of ASD can be either nonverbal or verbal.

Every person with ASD has a unique personality and combination of characteristics. Some individuals with mild ASD exhibit only slight delays in language, but greater challenges with social interactions. For example, they may have difficulty initiating and/or maintaining a conversation. Individuals with ASD are often described as talking at others instead of with others (e.g., they may monologue on a favorite subject despite attempts by others to interject comments).

You can review the entire DSM 5 Diagnostic criteria required for a clinical diagnosis of ASD in Appendix A.

There is also an educational definition of ASD. The educational definition was designed to identify children eligible for services under the Individuals with Disabilities Education Act (IDEA). This identification is made by the child's evaluation team which is convened by the school district. Parents are included as members of this team. This evaluation takes into consideration whether the child's symptoms adversely affect her educational performance. (See Chapter 5). A clinical diagnosis is not required for an educational identification of ASD, nor does it automatically guarantee identification under IDEA.

ASD does not discriminate according to social, economic status, or race. There is no proven cause of ASD.

Possible Signs of an ASD

*“If you’ve met one child with autism, you’ve met one child with autism.”
(Stephen Shore, adult with ASD)*

In general, ASD impacts communication and social interaction, as well as an individual’s ability to develop and maintain relationships and adjust behavior to a variety of social situations. Behaviors and function can vary widely within and across individuals with ASD.

Because of the complexity of ASD, the clinical diagnosis has several components. In order for a child to receive a clinical diagnosis of ASD, two core symptoms must be present:

- (1) Persistent deficits in social communication and social interaction across multiple contexts, and
- (2) Restricted, repetitive patterns of behavior, interests or activities.

Each of these symptoms must be qualified with a rating of their level of severity, either:

- (1) Requiring support,
- (2) Requiring substantial support, or
- (3) Requiring very substantial support.

Further, the identified symptoms must:

- Be present in the early developmental period (although they may not become fully manifest until social demands exceed the capacity of the individual),
- Cause clinically significant impairment in social occupational or other important areas of current functioning,
- Cannot be better explained by an intellectual disability or a global developmental delay

And finally, because ASD can often co-occur with an intellectual disability and language impairments can occur separate from ASD, the clinical diagnosis must also specify if the ASD is:

- With or without accompanying intellectual impairment,
- With or without accompanying language impairment, and
- With or without a known medical genetic condition or environmental factor.

(To review the complete DSM-5 Diagnostic Criteria, see Appendix A.)

Here are descriptions of two individuals with ASD:

John is a 7-year-old boy who received a clinical diagnosis of ASD when he was 3 years old. He does not speak, but uses gestures to make his needs known. When he is not understood, he shows frustration by squealing, throwing himself on the floor and crying. In school, he receives full-day instruction in a classroom for children with ASD. He can complete simple puzzles and match blocks by color when asked and supervised directly. John does not interact with his peers. He prefers playing alone and does not play with toys in the way they were intended.

Gracie is an 8-year-old girl who was identified by her school evaluation team under the category of autism. After her identification at school, her parents took her to a children's hospital for evaluation where she was diagnosed with a high functioning form of ASD. Gracie is very verbal and attends a regular second grade classroom. While she can read words at a sixth grade level, her comprehension skills are at a first-grade level. Her teachers report that Gracie has difficulty interacting with her classmates. She loves to talk about spiders and bugs and has begun her own bug collection. She continually tries to dominate conversations with her peers around the topic of bugs. Gracie does not realize that her peers are not interested when they walk away while she is talking.

While both of these students have ASD, certainly, their characteristics in the areas of communication, behavior, and socialization vary greatly.

The following is a list of some common behaviors or characteristics you might observe in your child.

Language and Communication

- Difficulty in expressing needs (use of gestures or pointing instead of words)
- Delayed speech or no speech
- Immediate or delayed repetition of the words of another person (family member, peers, TB character, singer, etc.) instead of typical, responsive language
- Has difficulty processing language (may not understand and/or may take longer to respond)
- Literal interpretation of language
- Difficulty in understanding nonverbal cues, including facial expressions
- Not responsive to verbal cues (acting as if deaf although hearing tests in normal range)
- Does not use joint attention—showing or sharing something with another person: this is typically demonstrated by using eye gaze and gestures, particularly pointing, for social interaction

Social

- Lack of social interaction; may prefer to be alone
- Difficulty interacting with other children
- Difficulty initiating conversation or play with others
- Little or no eye contact
- Acts or speaks in socially inappropriate manner (such as spaking too loudly or for too long)
- Emotional responses that do not match situations (e.g., over- or under-reaction)

Unique Behaviors

- Insistence on sameness; resistance to change
- Tantrums/meltdowns
- Sustained odd play
- Special interests or inappropriate attachment to objects (e.g., spins or lines up objects)
- Limited food choices and/or textures
- Frequently walks on tiptoes (toe-walking)
- Stereotyped behaviors, including hand flapping, whole-body rocking, clapping, etc.
- Self-injurious behaviors

Emotional

- Lack of awareness of own and others' feelings
- No real fear of danger
- Anxious or easily stressed

Sensory and Motor Skills

- Over- or under-sensitivity related to one or more sensory processing systems, including touch, balance, body awareness, sight, hearing, taste, smell
- Impaired gross or fine-motor skills
- Odd posture or gait
- Noticeable physical over-activity or extreme under-activity

Causes of ASD

There is no known single cause of ASD, but it is generally accepted that it is caused by abnormalities in brain structure or function. For example, brain scans show differences in the shape and structure of the brains of children with ASD. Researchers are investigating a number of theories, including the links between heredity, genetics, and medical problems such as tuberous sclerosis, or epilepsy. In many families, there appears to be a pattern of ASD or related disabilities, further supporting a genetic basis to the disorder. It also appears that some children are born with a susceptibility to ASD, but researchers have not yet identified a single "trigger" that causes ASD to develop.

Occurrence of Autism Spectrum Disorder

In 2014, CDC's National Center on Birth Defects and Developmental Disabilities released a report confirming that the rate of ASD continues to rise. Research in 2010 targeting 8 year old children living in 11 communities indicated a rate of 1 in 68 children were currently diagnosed with ASD. This new estimate is 30% higher than the estimated 1 in 88 children in 2008, 60% higher than the estimates of 1 in 110 reported in 2006, and 120% higher than the estimates for 2002 and 2000 (1 in 150). ASD is more common than childhood cancer, Down Syndrome, and juvenile diabetes.

Genetics Research and Autism

As mentioned, there is currently no known cause of ASD. During the past decade, scientists have made significant breakthroughs in understanding the genetics of ASD. Researchers are now focusing on specific chromosomal regions that may contain autism-related genes. This has been accomplished by studying chromosomal abnormalities in individuals with ASD and by screening each chromosome for evidence of genes associated with ASD.

Current theory among autism genetics researchers supports the idea of "complex" inheritance. This means that multiple genetic factors are likely to be involved and may predispose an individual to develop autism. This theory also includes a role for environmental factors. That is, in addition to having a certain combination of autism-related genes, exposure to specific environmental factors may be necessary for autism to develop in some individuals.

For instance, if one version of a gene makes a person susceptible to a particular chemical, exposure to that chemical could trigger autism to develop. By focusing on the study of genetic factors and determining their underlying mechanisms, researchers may be better able to pinpoint environmental factors that contribute to autism.

Much of current research is based on the Human Genome Project, a 13-year scientific study to identify and analyze all the genes in human DNA. If you have questions about a possible genetic link regarding autism in your family,

it is recommended that you consult a geneticist.

Resources

Autism Speaks: www.autismspeaks.com

Autism Society of America: www.autism-society.org

AUCD: www.aucd.org

Collaborative on Health and the Environment: http://healthandenvironment.org/resources/practice_prevention

HelpGuide.org: <http://www.helpguide.org/articles/autism/autism-spectrum-disorders.htm>

Human Genome Project: www.ornl.gov; www.genome.gov