AUTISM OVERVIEW

This review was copied from from UpToDate/patient and the formatting is at times a bit off.

To utilize links please press on control button and then double click to mouse)

Deb Bershel, MD 1/31/10

Please note- This topic can be anxiety-provoking, in part because it tends to be recognized in early childhood and there is a fear that we will be letting innocent children down if they don't receive 'the perfect treatment' at the earliest age. If you have any concerns about a loved one after reading this information, please don't panic but do call the office)

Autism spectrum disorders (ASDs) are a group of developmental disorders that cause lifelong difficulties with socializing, communicating, and behavior. The term "spectrum" refers to the fact that some people have a few mild symptoms while others have severe symptoms that are disabling. There are several types of ASD; the most common include:

- Autistic disorder (classic autism, sometimes called early infantile autism, childhood autism, or Kanner's autism)
- Asperger disorder (also known as Asperger syndrome)
- Pervasive developmental disorder, not otherwise specified (PDD-NOS), including atypical autism

These disorders share some of the same symptoms, but differ in the age at which symptoms begin, the severity of the symptoms, and how the symptoms are expressed. This topic will discuss general information about autism spectrum disorders, as well as the signs and symptoms of the most common type of ASD, autistic disorder.

How does autism develop? — It is not clear how or why autism develops. The general consensus is that autism spectrum disorders are a genetic disorder that affects brain development and impairs the development of social and communication skills. This, in turn, leads to the typical symptoms of ASD. (See 'Symptoms of autistic disorder' below.)

Environmental factors such as toxic exposures before or after birth, complications during delivery, and infections before birth may be responsible for a small percentage of cases. In children with a genetic predisposition, environmental factors may further increase the child's risk of developing autism spectrum disorder. It is not currently possible to test a child's genes to determine if he or she is at risk for ASD.

Some authors have attributed autism to vaccine exposure (particularly measles vaccine and thimerosal, a mercury preservative used in vaccines). However, the overwhelming majority of scientific studies do not support an association between immunizations and autism. (See "Patient information: Childhood immunizations".)

How common is autism? — The number of children diagnosed with autism spectrum disorder in the United States and other countries has increased since the 1970s, and particularly since the late 1990s. It is not clear if the increase is related to changes in the criteria used to diagnose ASD or if the condition has become more common over time. Most experts agree that increased awareness of autism and changes in the definition of ASD account for much of the apparent increase in the prevalence of autism.

Between 2 and 6.7 children per 1000 have autism spectrum disorder, and it affects more boys than girls (approximately four boys for every one girl). Approximately 2 to 8 percent of siblings of children with ASD also have the condition. (So we are talking about 1-2.5% of males can be affected – D. Bershel)

Medical conditions associated with autism — There are a number of medical conditions associated with autism.

- Between 45 and 60 percent of children with autism are mentally retarded.
- Seizures occur in 11 to 39 percent of children with ASD. The risk of seizures is higher in individuals with more severe intellectual disability (mental retardation). (See "Patient information: Seizures in children".)
- A minority (fewer than 10 to 25 percent) of cases of ASD are associated with a medical condition or syndrome, such as phenylketonuria, fetal alcohol syndrome, tuberous sclerosus, fragile X syndrome, or Angelman syndrome. These syndromes are usually diagnosed at or shortly after birth.

SYMPTOMS OF AUTISTIC DISORDER

Symptoms of autistic disorder are usually recognized between two and three years of age, although they may be present earlier. By definition, symptoms must be present by three years of age.

In approximately two-thirds of children with autism, the first sign is a lack of communication skills by two years of age. The remaining children have relatively normal development during the first 12 to 24 months of life, followed by a period of regression (when the child loses skills he or she had previously attained). Reasons for parents to seek help are discussed below. (See 'When to seek help' below.)

Social interaction — Difficulty with or lack of interaction with family and friends is a hallmark of autism spectrum disorder. Components of social interaction include nonverbal behaviors, peer relationships, joint attention, and social reciprocity (which are explained below).

Nonverbal behaviors — Individuals with autism spectrum disorder have difficulty using and interpreting nonverbal behaviors such as eye contact, facial expression, gestures, and body postures. For example, a child may not be able to understand the facial expressions associated with anger or annoyance.

During infancy, parents may notice that the baby resists cuddling, avoids eye contact, or does not spread the arms in anticipation of being picked up; however, these behaviors are not universal.

Peer relationships — Individuals with autism spectrum disorder often have a hard time learning to interact with other people. Younger children may have little or no interest in developing friendships. They may prefer to play alone rather than playing with others, and may involve others in activities only as tools or "mechanical" aids (ie, using the hand of a parent to obtain a desired object without making eye contact).

Older children may become more interested in talking or socializing with other people, but may not understand social conventions or the needs of others. As an example, the child may continue talking about a topic of their own interest with complete disregard for the interests of the listener.

Joint attention — Individuals with autism spectrum disorder are not interested in sharing activities, interests, or achievements; this is referred to as impaired joint attention.

Joint attention is a normal behavior in which an infant or toddler tries to share interest, amusement, or fear with a caretaker. The child does this by purposefully looking back and forth between an object and the eyes of the caretaker (usually by eight to 10 months of age), or by pointing to the object (usually by 14 to 16 months of age). Older children with ASD may not show or bring an object to the caretaker.

Social referencing — Individuals with autism spectrum disorder are sometimes not able to share a pleasurable activity with others. As an example, the child may prefer to play alone amidst a crowd of children engaged in the same activity.

Communication — The second hallmark of autism spectrum disorder is significant difficulty with communication. This is the most common concern of parents, which often leads the parent to seek medical attention. The child may lack the ability to speak or understand and/or may show no interest in communicating.

Language — A delayed or absent ability to speak is a feature of autism. Unlike children with a hearing impairment, children with autism spectrum disorder do not try to compensate for their lack of speech by using alternate means of communication, like gesturing or miming. In most individuals with ASD, the ability to understand is delayed even more than the ability to speak. Children may not respond to their name, and the parent may initially be concerned that the child has a hearing problem. A child may not be able to understand simple questions or directions.

There is wide variability in the severity and quality of language problems in children with autism. The ability to speak never develops in approximately one-half of affected children. In others, the child is able to speak, but language is not used as a tool for communication (eg, it consists of repeating phrases or words spoken by others, called echolalia).

Those who are able to speak may have difficulty starting or sustaining a conversation with others. Their language may have meaning only to people who are familiar with the autistic individual's communication style.

Play — Lack of varied, spontaneous make-believe play or imitation is a characteristic feature of autism. Children with autism spectrum disorder may line up an exact number of playthings in the same manner, without awareness of what the toys represent. They may focus on parts of toys rather than the function of the toy. They may not engage in pretend play, which usually develops by two years of age. Even if pretend play develops, the child may simply be mimicking what has been seen on television or in books.

Behavior — The third hallmark of autism spectrum disorder is repetitive behaviors and interests.

Preoccupations — Younger children may be preoccupied with peculiar sensory objects or experiences, such as spinning objects, shiny surfaces, the edge of objects, lights, odors, or sniffing or licking nonfood objects.

Older children may be preoccupied with the weather, dates, schedules, phone numbers, license plates, cartoon characters, or other items (eg, dinosaurs, dogs, planes).

Rituals — Many children with autism spectrum disorder have specific routines or rituals that must be followed exactly. These may occur as a part of daily life, such as the need to always eat particular foods in a specific order, or to follow the same route from one place to another without deviation. Changes in routine can be upsetting or frustrating, even causing the child to have a tantrum or meltdown.

Motor mannerisms — Another behavioral feature of autism spectrum disorder is repetitive body movements, such as hand or finger flapping or twisting, rocking, swaying, dipping, or walking on tip-toe. These behaviors are seen in 37 to 95 percent of individuals with ASD, and commonly begin during the preschool years. These behaviors are often lifelong.

Cognitive skills — Cognitive skills include the ability to think, remember, and process information. In children with autism, these skills are often uneven, regardless of the child's level of intelligence. The ability to speak clearly is usually not as good as nonverbal skills. The person can often perform tasks that require memorization or putting things together (eg, puzzles), but may have difficulty with tasks that require higher-level skills, such as reasoning, interpretation, or abstract thinking.

Some individuals have special skills (ie, "savant" skills) in memory, mathematics, music, art, or puzzles, despite significant difficulties in other areas. Other special skills can include calendar calculation (determining the day of the week for a given date) and hyperlexia (the ability to read written words that are far above the person's reading level). However, the person does not usually understand what is being read or the purpose of reading.

Sensory perception — Many people with autism spectrum disorder perceive sounds, tastes, or touch abnormally. For example, the person may be overly sensitive to normal noise levels or have no response to loud noises.

Other examples include:

- Refusal to eat foods with certain tastes or textures, or eating only foods with certain tastes and textures. These dietary obsessions can cause gastrointestinal symptoms, such as weight loss, diarrhea, or constipation. Malnutrition can also occur, even if the child eats an adequate number of calories per day. This can lead to serious deficiencies of important vitamins such as calcium, vitamin D, and/or vitamin C.
- Resistance to being touched or increased sensitivity to certain kinds of touch; light touch may be experienced as painful, whereas deep pressure may provide a sense of calm. This may include resistance to the feel of certain clothing textures or colors next to the skin.
- Apparent indifference to pain.
- Hypersensitivity to certain frequencies or types of sound (eg, distant fire engines) and lack of response to sounds close by or sounds that would startle other children (eg, firecrackers).

Macrocephaly — Approximately one-fourth of children with autism spectrum disorder have a larger than normal-size head. The medical term for this is macrocephaly. This may be related to abnormalities in early brain development, which contribute to the signs and symptoms of autism discussed above.

AUTISM DIAGNOSIS

If a child has symptoms of autism spectrum disorder, s/he is usually evaluated by a team that has expertise in diagnosing and managing the condition. This team often includes a child psychologist, developmental-behavioral pediatrician, neurologist, psychiatrist, speech therapist, and other professionals.

The evaluation usually includes a complete medical history (of the child and family), physical examination, neurologic examination, and testing of the child's social, language, and cognitive skills. In addition, the parent(s) will have time to discuss the child's behavior and any other concerns.

The purpose of the evaluation includes the following:

- Determine if the child has ASD or if another condition could be causing the child's symptoms.
- Determine if the child has any ASD-associated medical problems that should be evaluated or treated.
- Determine the child's strengths, weaknesses, and level of functioning.

WHEN TO SEEK HELP

Some common symptoms of autism spectrum disorder are listed in table 1 (table 1).

Parents who notice that their child has one or more symptoms of autism spectrum disorder should talk the child's healthcare provider. The provider should screen the child for ASD according to the American Academy of Pediatrics algorithm (algorithm 1).

If the provider's evaluation raises red flags for autism, the child should be referred for a complete evaluation for autism spectrum disorder. Early diagnosis and treatment of ASD can modify some autistic behaviors and improve socialization. (See 'Autism diagnosis' above.)

Even before the complete evaluation, the child should be referred for a hearing test (if not done previously) and for early intervention services. Early intervention is a support system that provides appropriate therapies for children with disabilities. It can help to minimize delays and maximize the child's chance of reaching normal milestones in development. Even if the child is not diagnosed with autism, early intervention services can help to address parents' concerns (eg, delayed language skills, temper tantrums).

AUTISM TREATMENT

Autism cannot be cured. However, a healthcare provider can work with parents to develop a treatment plan to help the child reach his or her full potential. The optimal treatment plan depends upon the child's age, diagnosis, underlying medical problems, and other individual factors.

The American Academy of Pediatrics recommends a plan that provides structure, direction, and organization for the child [1]. In the United States, services are often provided through an early intervention program, administered by the individual states. Information about services for children with autism is available through the National Dissemination Center for Children with Disabilities (www.nichcy.org).

Other resources for parents and providers are listed below. (See 'Where to get more information' below.)

WHERE TO GET MORE INFORMATION

Your child's healthcare provider is the best source of information for questions and concerns related to your child's medical problem. Because no two people are exactly alike and recommendations can vary from one person to another, it is important to seek guidance from a provider who is familiar with your child's situation.

This discussion will be updated as needed every four months on our web site (www.uptodate.com/patients). Additional topics as well as selected discussions written for healthcare professionals are also available for those who would like more detailed information.