

Autism

Sharon Witemeyer MD (Pediatrician)

Definition

Autism is a lifelong developmental disability defined by the presence of a cluster of three behaviors: 1) a qualitative impairment in reciprocal (back and forth) social interaction, 2) a qualitative impairment in the development of language and communication, and 3) a restricted (narrow) range of activities and interests.

Introduction

Dr. Leo Kanner, a psychiatrist at Johns Hopkins University, first described autism over 50 years ago. The definition and our understanding of autism have broadened over the years since then. Autism is a spectrum disorder with symptoms that range from severe to very close to normal. It is a developmental disorder and its expression changes with age. It occurs in the range of 5-15 per 10,000 live births. It is 4 times more likely to affect boys than girls. Autism affects people of all socioeconomic, racial and ethnic backgrounds. The onset of symptoms occurs before 3 years of age. Some infants are different from birth, but in many early symptoms may be overlooked and the diagnosis delayed.

No one knows the cause of autism. There are many theories. Some areas of research include food allergies, ear infections, viral infections, genetic predisposition, variations in brain structure, pollutants, vitamin or mineral deficiencies, metabolic imbalances, environmental factors, and exposure to the manmade hormone Pitocin. Research has proven that no psychological factors in a child's upbringing cause autism

Dr. B.J. Freeman from UCLA provides a nice summary of what we look for in an individual with autism when he notes " autism usually manifests itself by the appearance of typical behavioral symptoms in the following areas:

1. Disturbances in the rate of appearance of physical, social and language skills.
2. Abnormal responses to sensations. Any one or a combination of senses or responses are affected, sight, hearing touch, balance, reaction to pain or in the way the child holds his body.
3. Speech, language and non-verbal communication.
4. Abnormal ways of relating to people, objects and to events in the environment."

Autism frequently occurs in association with other syndromes or developmental disabilities. Some of these are:

- Mental Retardation: 70% of individuals with autism score below 70 on IQ tests. Many children with severe/profound mental retardation exhibit some of the behaviors seen in autism.
- Seizure Disorder: By adulthood 25% of individuals with autism have developed seizures. These often begin at puberty and may be accompanied by marked behavioral changes and developmental regression.
- Tuberous Sclerosis: TS is an inherited disorder characterized by mental retardation, facial fibroangiomas (benign tumors) or poorly pigmented (pale) spots of skin or hair and seizures. About 75% of individuals with TS exhibit autism. About 3-4% of individuals with autism have TS.
- Fragile X Syndrome: Fragile X is the most common inherited form of mental retardation. Fewer than 5% of individuals with autism have Fragile X.
- Other Chromosome Abnormalities: About 5% of individuals with autism have a chromosome abnormality.
- Psychiatric Co-morbidities: Obsessive-compulsive disorder/behavior, depression, anxiety disorders, and ADHD are examples.

- Medical Co-Morbidities: Gastroesophageal Reflux Disease (GERD), other gastrointestinal problems, thyroid disease (increased in all individuals with developmental disabilities), obesity (particularly in adults), allergies/"sinus" problems/otitis media.

Diagnosis

Autistic Disorder (299.00)

Diagnostic and Statistical Manual; Fourth Edition (DSM-IV)

- A. A total of six (or more) items from (1), (2), and (3), with at least two from (1) and one each from (2) and (3):
 - 1. Qualitative impairment in social interaction, as manifested
 - a. marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - b. failure to develop peer relationships appropriate to developmental level
 - c. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. by a lack of showing, bringing, or pointing out objects of interest)
 - d. Lack of social or emotional reciprocity
 - 2. Qualitative impairments in communication as manifested by at least one of the following:
 - a. delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
 - b. in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
 - c. stereotyped and repetitive use of language or idiosyncratic language
 - d. lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
 - 3. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by a least one of the following:
 - a. encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 - b. apparently inflexible adherence to specific, nonfunctional routines or rituals
 - c. stereotyped and repetitive motor mannerisms (e.g. hand or finger flapping or twisting, or complex whole-body movements)
 - d. persistent preoccupation with parts of objects
- B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.
- C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

The diagnostic criteria for autism are outlined in the Diagnostic and Statistical Manual, Fourth Edition (DSM-IV). Autism is placed under the general category of Pervasive Developmental Disorders (PDD) that includes Autistic Disorder, Asperger's Syndrome, Rett's Syndrome, Childhood Disintegrative Disorder and Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS). These diagnoses have communication and social impairments in common.

Diagnosis is made on clinical grounds. Several diagnostic instruments are available. Here are some of the screening tests used for young children:

1. Checklist for Autism in Toddlers (CHAT)
2. Autism Screening Questionnaire (ASQ)
3. Pervasive Developmental Disorders Screening Test-Stage 1 (PDDST)
4. Childhood Autism Rating Scale (CARS)

Assessment is best accomplished by a multidisciplinary team and includes a complete developmental history, hearing and vision screening, direct behavioral observations across settings, speech and language evaluation, use of

a standardized autism rating scale and other tests as needed (EEG, MRI, chromosome/DNA probes, selective metabolic studies are some examples.) Tests that are NOT recommended include functional neuro-imaging or MEGs, allergy testing, Immunologic testing, hair analysis for trace elements, urinary peptides, gut permeability studies, stool analysis or erythrocyte glutathione peroxidase.

The other Pervasive Developmental Disorder identified in the DSM- IV are:

1. Asperger's Syndrome- There is disagreement among the experts as to whether Asperger's Syndrome is a mild form of autism like "high functioning autism" or a separate category entirely. These individuals show social and communication deficits and unusual circumscribed interests but not early language developmental delays. The DSM-IV criteria state "there must be no significant associated delay in either general cognitive functioning, self-help/adaptive skills, interest in the environment, or overall language development.
2. Rett's Syndrome- This condition occurs only in girls. They have normal development in the first year of life, but then head growth slows. Over the next two years they loose purposeful hand skills and verbal skills. They develop "hand-wringing" or "hand-washing" movements and social impairments. They develop seizures. Loss of skills is persistent throughout life and progressive.
3. Atypical Autism-PDD-NOS – Pervasive Developmental Disorder Not Otherwise Specified is used when there is a severe impairment in social interaction, communication and stereotyped interests but the criteria for a specific Pervasive Developmental Disorder is not met.
4. Childhood Disintegrative Disorder – These children have normal development for at least 2 years and then exhibit developmental regression and behavioral changes over a period of several months. They loose expressive and receptive language, social skills, adaptive behaviors, and bowel or bladder control. They develop restricted, repetitive and stereotyped patterns of behavior and marked social withdrawal. The changes must have their onset before 10 years of age.

Other conditions that need to be ruled out before a diagnosis of autism is made include Schizophrenia, Deafness, Elective Mutism, Landau Kleffner Syndrome, Psychosocial Deprivation and Receptive Language Disorder.

Treatment

Because we do not know the cause of autism we have no effective way to prevent the condition.

There is no cure for autism. Research on the cause of and treatments for autism is on going. There is a great deal of information and misinformation about autism available. On the Internet one can find over 100,000 sites devoted to this topic. So far, the only treatment that has been proved effective for all children with autism in the long run are structured educational programs. The earlier and the more intensive the intervention the better.

No other treatment, including prescription medications, vitamin/mineral supplements, DMG, dietary restrictions, secretin, patterning, AIT, or behavior modification programs has proved effective in all individuals with autism. Some of these methods do seem to benefit some individuals. Prescription medications may be helpful in individual cases. Examples are Ritalin for attention deficit problems, Clonidine for sleep disturbance, anticonvulsants for seizures, and psychoactive medications for depression, anxiety, SIB or behavior problems. According to Dr. Stephen Edelson from the Center for the Study of Autism "Vitamin B6 taken with magnesium has been shown to increase general well-being, awareness, and attention in approximately 45% of autistic children." On the other hand, recent scientific studies of the hormone secretin have not supported earlier reports of benefit to individuals with autism. New treatments for autism appear on a regular basis, and it is important to approach each new treatment with caution. Above all we hope to do no harm to individuals with autism.

Emergency Situations – What can go wrong?

Autism is a neuro-developmental disorder that in itself does not lead to emergency situations. However, there are two areas in particular in which associated problems could lead to emergencies. These are seizure disorders and psychiatric co-morbidities.

Seizure Disorders can lead to:

Status epilepticus – prolonged seizure activity such as a seizure that lasts for more than 10 minutes or several seizures that occur one after another for 20-30 minutes.

Injury – including bruising, concussion, fractures or even drowning if the seizure occurs during a bath.

Trouble breathing – individual's lips may turn blue.

What to Do:

1. Clear the area around the individual, stay with him/her to prevent injury. DO NOT put anything in his/her mouth.
2. Try to write down what happened before, during and after the seizure and how long the seizure lasts.
3. Notify agency nurse/supervisor as soon as possible.
4. Call 911 if the seizure lasts longer than 5 minutes, if individual is injured or if he/she stops breathing.
5. If individual stops breathing, start rescue breathing if you are certified to so.

The psychiatric co-morbidity most likely to lead to an emergency is:

Depression and Suicide Attempt

What to Do:

1. Insure the individual's safety if possible.
2. Call 911.
3. Notify agency and individual's physician (psychiatrist and/or PCP) as soon as possible.

Conclusion

Autism is a lifelong developmental disability defined by a behavior triad of (1) a qualitative impairment in reciprocal social interaction, (2) a qualitative impairment in the development of language and communication and (3) a restricted range of activities and interests has its onset before 3 years of age. Symptoms range from severe to nearly normal. A great deal of research is being done to discover the cause of autism and to find a cure or treatment for autism. At this time the cause of autism is unknown and there is no cure. The only treatment that has proved effective for all children with the condition is structured educational programs. Many other treatments have been suggested, but none has been effective in all individuals with autism. Some do seem to benefit some individuals. When considering a new treatment modality for autism one must use caution before trying it. Many suggested treatments have not been tested scientifically. Above all DO NO HARM.

References

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Sharon Witemeyer MD (Pediatrician)