

Overview of Autism

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It has been over 50 years since Dr. Leo Kanner, a psychiatrist at Johns Hopkins University, wrote the first paper applying the term 'autism' to a group of children who were self-absorbed and who had severe social, communication, and behavioral problems. This paper provides a general overview of the complexity of this developmental disability by summarizing many of the major topics in autism.

Prevalence

For many years, the most cited statistic is that autism occurs in 4.5 out of 10,000 live births. This was based on large-scale surveys conducted in the United States and England. More recently, estimates on the prevalence of autism have ranged been as high as 1/4% to 1/2% of the population. These estimates typically include those with autism, Asperger syndrome, and PDD.

Autism is three times more likely to affect males than females. This gender difference is not unique to autism since many developmental disabilities have a greater male to female ratio.

Major characteristics

Many autistic infants are different from birth. Two common characteristics they may exhibit include arching their back away from their caregiver to avoid physical contact and failing to anticipate being picked up (i.e., becoming limp). As infants, they are often described as either passive or overly agitated babies. A passive baby refers to one who is quiet most of the time making little, if any, demands on his/her parents. An overly agitated baby refers to an infant who cries a great deal, sometimes non-stop, during his/her waking hours. During infancy, many begin to rock and/or bang their head against the crib; but this is not always the case.

In the first few years of life, some autistic toddlers reach developmental milestones, such as talking, crawling, and walking, much earlier than the average child; whereas others are considerably delayed. Approximately one-half of autistic children develop normally until somewhere between 1 1/2 to 3 years of age; then autistic symptoms begin to emerge. These individuals are often referred to as having 'regressive' autism. Some people in the field believe that candida albicans, vaccinations, exposure to a virus, or the onset of seizures may be responsible for this regression. It is also thought that some children with 'regressive' autism may have Landau-Kleffner Syndrome (see next section).

During childhood, autistic children may fall behind their same-aged peers in the areas of communication, social skills, and cognition. In addition, dysfunctional behaviors may start to appear, such as self-stimulatory behaviors (i.e., repetitive, non-goal directed behavior, such as rocking, hand-flapping), self-injury (e.g., hand-biting, headbanging), sleeping and eating problems, poor eye contact, insensitivity to pain, hyper-/hypo-activity, and attention deficits.

One characteristic which is quite common in autism is the individual's 'insistence on sameness' or 'perseverative' behavior. Many children become overly insistent on routines; if one is changed, even slightly, the child may become upset and tantrum. Some common examples are: drinking and/or eating the same food items at every meal, wearing certain clothing or insisting that others wear the same clothes, and going to school using the same route. One possible reason for 'insistence on sameness' may be the person's inability to understand and cope with novel situations.

Autistic individuals sometimes have difficulty with the transition to puberty. Approximately 25% have seizures for the first time during puberty which may be due to hormonal changes. In addition, many behavior problems can become more frequent and more severe during this period. However, others experience puberty with relative ease.

In contrast to 20 years ago when many autistic individuals were institutionalized, there are now many flexible living arrangements. Usually, only the most severe individuals live in institutions. In adulthood, some people with autism live at home with their parents; some live in residential facilities; some live semi-independently (such as in a group home); and others live independently. There are autistic adults who graduate from college and receive graduate degrees; and some develop adult relationships and may marry. In the work environment, many autistic adults can be reliable and conscientious workers. Unfortunately, these individuals may have difficulty getting a job. Since many of them are socially awkward and may appear to be 'eccentric' or 'different,' they often have difficulty with the job interview.

Subgroups and Related Disorders

There is no adjective which can be used to describe every type of person with autism because there are many forms of this disorder. For example, some individuals are anti-social, some are asocial, and others are social. Some are aggressive toward themselves and/or aggressive toward others. Approximately half have little or no language, some repeat (or echo) words and/or phrases, and others may have normal language skills. Since there are no physiological tests at this time to determine whether a person has autism, the diagnosis of autism is given when an individual displays a number of characteristic behaviors.

In the last five years, research has shown that many people who engage in autistic behaviors have related but distinct disorders. These include: Asperger Syndrome, Fragile X Syndrome, Landau-Kleffner Syndrome, Rett Syndrome, and Williams Syndrome. Asperger Syndrome is characterized by concrete and literal thinking, obsession with certain topics, excellent memories, and being 'eccentric.' These individuals are considered high-functioning and are capable of holding a job and of living independently.

Fragile X Syndrome is a form of mental retardation in which the long arm on the X chromosome is constricted. Approximately 15% of people with Fragile X Syndrome exhibit autistic behaviors. These behaviors include: delay in speech/language, hyperactivity, poor eye contact, and hand-flapping. The majority of these individuals function at a mild to moderate level. As they grow older, their unique physical facial features may become more prominent (e.g., elongated face and ears), and they may develop heart problems.

People with Landau-Kleffner Syndrome also exhibit many autistic behaviors, such as social withdrawal, insistence on sameness, and language problems. These individuals are often thought of as having 'regressive' autism because they appear to be normal until sometime between ages 3 and 7. They often have good language skills in early childhood but gradually lose their ability to talk. They also have abnormal brain wave patterns which can be diagnosed by analyzing their EEG pattern during an extended sleep period.

Rett Syndrome is a degenerative disorder which affects mostly females and usually develops between 1/2 to 1 1/2 years of age. Some of their characteristic behaviors include: loss of speech, repetitive hand-wringing, body rocking, and social withdrawal. Those individuals suffering from this disorder may be severely to profoundly mentally retarded.

Williams Syndrome is characterized by several autistic behaviors including: developmental and

language delays, sound sensitivity, attention deficits, and social problems. In contrast to many autistic individuals, those with Williams Syndrome are quite sociable and have heart problems.

Causes

Although there is no known unique cause of autism, there is growing evidence that autism can be caused by a variety of problems. There is some indication of a genetic influence in autism. For example, there is a greater likelihood that two monozygotic twins (i.e., identical twins) will have autism than two dizygotic twins (i.e., fraternal twins). In the case of monozygotic twins, there is a 100% overlap in genes; whereas in dizygotic twins, there is a 50% overlap in genes, the same overlap as in non-twin siblings. Currently, a great deal of research has focused on locating the 'autism gene;' however, many researchers speculate that three to five genes will likely be associated with autism. There is also evidence that the genetic link to autism may be a weakened or compromised immune system. Other research has shown that depression and/or dyslexia are quite common in one or both sides of the family when autism is present.

There is also evidence that a virus can cause autism. There is an increased risk in having an autistic child after exposure to rubella during the first trimester of the pregnancy. Cytomegalovirus has also been associated with autism. Additionally, there is also a growing concern that viruses associated with vaccinations, such as the measles component of the MMR vaccine and the pertussis component of the DPT shot, may cause autism.

There is growing concern that toxins and pollution in the environment can also lead to autism. There is a high prevalence of autism in the small town of Leominster, Massachusetts, where a factory manufacturing sunglasses was once located. Interestingly, the highest proportion of autism cases were found in the homes down-wind from the factory smokestacks. Recently, a large proportion of autistic children were identified in Brick Township, New Jersey. Several agencies are now attempting to uncover the reason(s) for the high proportion of autism in this community.

Physical abnormalities

Researchers have located several brain abnormalities in individuals with autism; however, the reasons for these abnormalities is not known nor is the influence they have on behavior. These abnormalities can be classified into two types--dysfunctions in the neural structure of the brain and abnormal biochemistry of the brain. It will be important for future researchers to examine the relationship between these two types of abnormalities.

With respect to brain structure, Drs. Bauman and Kemper examined post-mortem brains of several autistic individuals and have located two areas in the limbic system which are underdeveloped--the amygdala and the hippocampus. These two areas are responsible for emotions, aggression, sensory input, and learning. These researchers also found a deficiency of Purkinje cells in the cerebellum. Using Magnetic Resonance Imaging, Dr. Courchesne has found two areas in the cerebellum, vermal lobules VI and VII, which are significantly smaller than normal in autistic individuals. Interestingly, there are some autistic individuals whose vermal lobules VI and VII are larger than normal. One or both of these areas of the cerebellum are believed to be responsible for attention.

With respect to biochemistry, many autistic individuals have elevated levels of serotonin in their blood and cerebral spinal fluid, whereas others have relatively low levels of serotonin. It should be mentioned that other disorders, such as Down Syndrome, attention deficit/hyperactivity disorder, and unipolar depression are also associated with abnormal levels of serotonin. There is also evidence that some

autistic individuals have elevated levels of beta-endorphins, an endogenous opiate-like substance in the body. It is felt that those individuals who have an increased pain tolerance may likely be due to elevated levels of beta-endorphins.

A dysfunctional immune system has also been associated with autism. It is thought that a viral infection or an environmental toxin may be responsible for damaging the immune system. As mentioned above, there is also evidence of a genetic association to a compromised immune system. Researchers have found that many autistic individuals have a decreased number of helper t-cells which help the immune system fight infection.

There is growing evidence that the gut or intestinal tract of autism children is impaired. Researchers have documented yeast overgrowths (*candida albicans*), low levels of phenyl sulfur transferase, and measles virus in their intestinal tract.

Sensory impairments

Many autistic individuals seem to have an impairment in one or more of their senses. This impairment can involve the auditory, visual, tactile, taste, vestibular, olfactory (smell), and proprioceptive senses. These senses may be hypersensitive, hyposensitive, or may result in the person experiencing interference such as in the case of tinnitus, (a persistent ringing or buzzing in the ears). As a result, it may be difficult for individuals with autism to process incoming sensory information properly.

Sensory impairments may also make it difficult for the individual to withstand normal stimulation. For example, some autistic individuals are tactilely defensive and avoid all forms of body contact. Others, in contrast, have little or no tactile or pain sensitivity. Furthermore, some people with autism seem to 'crave' deep pressure. Another example of sensory abnormalities is hypersensitive hearing. Approximately 40% of autistic individuals experience discomfort when exposed to certain sounds or frequencies. These individuals often cover their ears and/or tantrum after hearing sounds such as a baby's cry or the sound of a motor. In contrast, some parents suspect their children of being deaf because they appear unresponsive to sounds.

Cognition

"Theory of mind" refers to one's inability to realize that other people have their own unique point of view about the world. Many autistic individuals do not realize that others may have different thoughts, plans, and perspectives than their own. For example, a child may be asked to show a photograph of an animal to another child. Rather than turning the picture around to face the other child, the autistic child may, instead, show the back of the photograph. In this example, the autistic child can view the picture but does not realize that the other child has a different perspective or point of view.

About 10% of autistic individuals have savant skills. This refers to an ability which is considered remarkable by most standards. These skills are often spatial in nature, such as special talents in music and art. Another common savant skill is mathematical ability in which some autistic individuals can multiply large numbers in their head within a short period of time; others can determine the day of the week when given a specific date in history or memorize complete airline schedules.

Many autistic individuals also have a narrow or focused attention span; this has been termed 'stimulus overselectivity.' Basically, their attention is focused on only one, often irrelevant, aspect of an object. For example, they may focus on the color of a utensil, and ignore other aspects such as the shape. In this case, it may be difficult for a child to discriminate between a fork and a spoon if he/she attends only to the color. Since attention is the first stage in processing information, failure to attend to the

relevant aspects of an object or person may limit one's ability to learn about objects and people in one's environment.

Interventions

Over the years, families have tried various types of traditional and non-traditional treatments to reduce autistic behaviors and to increase appropriate behaviors. Although some individuals are given medications to improve general well-being, there is no primary drug which has been shown to be consistently effective in treating symptoms of autism. The most widely prescribed medication for autistic children is Ritalin, (a stimulant used to treat Attention Deficit/Hyperactivity Disorder). However, there are no double-blind controlled studies to demonstrate its effectiveness for those with autism.

The two treatments which have received the most empirical support are Applied Behavior Analysis (ABA; behavior modification) and the use of vitamin B6 with magnesium supplements. Behavior modification involves a variety of strategies, (e.g., positive reinforcement, time-out), to increase appropriate behaviors, such as communication and social behavior, and to decrease inappropriate behaviors, such as self-stimulatory and self-injurious behavior.

Vitamin B6 taken with magnesium has been shown to increase general well-being, awareness, and attention in approximately 45% of autistic children. There are also a number of recent reports about the benefits of another nutritional supplement, Di-methylglycine (DMG). DMG also seems to help the person's general well-being, and there are many anecdotal reports of it enhancing communication skills.

Some people with autism have excessive amounts of a type of yeast called 'candida albicans' in their intestinal tract. It is thought that high levels of candida albicans may be a contributing factor to many of their behavioral problems. One scenario is that when a child develops a middle ear infection, the antibiotics that help fight the infection may destroy microbes that regulate the amount of yeast in the intestinal tract. As a result, the yeast grows rapidly and releases toxins in the blood; and these toxins may influence the functioning of the brain. Excessive candida albicans can be treated with rather mild medications such as Nystatin.

Food intolerances and food sensitivities are beginning to receive much attention as possible contributors to autistic behaviors. Many families have observed rather dramatic changes after removing certain food items from their children's diet. Researchers have recently detected the presence of abnormal peptides in the urine of autistic individuals. It is thought that these peptides may be due to the body's inability to breakdown certain proteins into amino acids; these proteins are gluten (e.g., wheat, barley, oats) and casein (found in human and cow's milk). Many parents have removed these substances from their children's diets and have, in many cases, observed dramatic, positive changes in health and behavior.

As mentioned earlier, many autistic individuals have sensory impairments. Sensory integration techniques are often used to treat dysfunctional tactile, vestibular, and proprioceptive senses. Some of the techniques involve swinging a child on a swing in various ways to help normalize the vestibular sense and rubbing different textures on the skin to normalize the tactile sense. In addition, an autistic woman, Dr. Temple Grandin, developed a hug machine which provides the individuals with deep pressure which appears to have a calming effect on the person.

Many autistic individuals are also sensitive to sounds in their environment. They may hear sounds beyond the normal range and/or certain sounds may be perceived as painful. Auditory integration training, (listening to processed music for ten hours), is an intervention which is often used to reduce these sensitivities. Visual training is another sensory intervention designed to normalize one's vision.

There are several different methods of visual training. One popular program, developed by Dr. Melvin Kaplan, involves wearing ambient (prism) lenses and performing movement exercises which appear to reorganize and normalize the visual system.

Conclusion

Autism is a very complex disorder; and the needs of these individuals vary greatly. After 50 years of research, traditional and contemporary approaches are enabling us to understand and treat these individuals. It is also important to mention that parents and professionals are beginning to realize that the symptoms of autism are treatable--there are many interventions that can make a significant difference.

The logo for the national parent support group, the Autism Society of America, is a picture of a child embedded in a puzzle. Most of the pieces of the puzzle are on the table, but we are still trying to figure out how they fit together. We must also keep in mind that these pieces may fit several different puzzles.