Intellectual Disabilities: Mental Retardation and Autism

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What is Mental Retardation?

The term Mental Retardation has been used in the past, but in today's society the preferred term is Intellectual Disability. Both mental retardation and intellectual have the same definition.

Intellectual disability is a disability characterized by significant limitations in both **intellectual functioning** and in **adaptive behavior**, which covers many everyday social and practical skills. This disability originates **before the age of 18**.

Intellectual disability is a person with significantly sub average general intellectual functioning, existing concurrently with deficits in adaptive behavior and manifested during the developmental period that adversely affects a child's educational performance.

Intellectual functioning—also called intelligence—refers to general mental capacity, such as learning, reasoning, problem solving, and so on.

Adaptive behavior is the collection of conceptional, social, and practical skills that are learned and performed by people in their everyday lives.

> **Conceptual skills**—language and literacy; money, time, and number concepts; and self-direction.
>
> **Social skills**—interpersonal skills, social responsibility, self-esteem, gullibility, naïveté (i.e., wariness), social problem solving, and the ability to follow rules/obey laws and to avoid being victimized.
>
> **Practical skills**—activities of daily living (personal care), occupational skills, healthcare, travel/transportation, schedules/routines, safety, use of money, use of the telephone.

Intellectual disability is thought to affect about 1% of the population. Of those affected, 85% have mild intellectual disability. This means they are just a little slower than average to learn new information or skills. With the right support, most will be able to live independently as adults.

What are the causes of intellectual disabilities?

Anytime something interferes with normal brain development, intellectual disability can result. However, a specific cause for intellectual disability can only be pinpointed about a third of the time.

There are many causes of intellectual disabilities. The most common causes are:

- **Genetic Conditions** – abnormalities of chromosomes and genes inherited from parents, errors when genes combine, or other reasons. Examples of this are Down syndrome, Fragile X syndrome, and Phenylketonuria (PKU).
- **Problems During Pregnancy** – the baby does not develop normally inside the mother. For example, there may be a problem with the way the way the baby's cells divide as it grows. A woman who drinks alcohol or gets an infection like rubella during pregnancy can also cause the child to have an intellectual disability.
- **Perinatal Problems** - problems during labor and birth, such as not getting enough oxygen, intracranial hemorrhage before or during birth, or a severe head injury.
- **Health Problems –** diseases like whooping cough, measles, or meningitis; extreme malnutrition, not getting proper medical care, and being exposed to certain poisons like lead or mercury.
- **Trauma –** before and after birth
- **Unexplained Reasons –** 75% of causes are unknown

Diagnosing Intellectual Disabilities

Intellectual disability may be suspected for many different reasons. If a baby has physical abnormalities that suggest a genetic or metabolic disorder, a variety of tests may be done to confirm the diagnosis. These include blood tests, urine tests, imaging tests to look for structural problems in the brain, or electroencephalogram (EEG) to look for evidence of seizures.

In children with developmental delays, the doctor will perform tests to rule out other problems, including hearing problems and certain neurological disorders. If no other cause can be found for the delays, the child will be referred for formal testing.

Three things factor into the diagnosis of intellectual disability: interviews with the parents, observation of the child, and testing of intelligence and adaptive behaviors. A child is considered intellectually disabled if he or she has deficits in both IQ *and* adaptive behaviors. To test a child's intelligence, doctors use an IQ (intelligence quotient) test. The average IQ is 100 and a person is considered intellectually disabled if he or she has an IQ of less than 70 to 75. If only one or the other is present, the child is not considered intellectually disabled.

To measure a child's adaptive behaviors, a specialist will observe the child's skills and compare them to other children of the same age. Things that may be observed include how well the child can feed or dress himself or herself; how well the child is able to communicate with and understand others; and how the child interacts with family, friends, and other children of the same age.

After a diagnosis of intellectual disability is made, a team of professionals will assess the child's particular strengths and weaknesses. This helps them determine how much and what kind of support the child will need to succeed at home, in school, and in the community.

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The purpose of establishing a diagnosis of intellectual disability is to determine eligibility for:

- Special education services
- Home and community-based waiver services
- Social Security Administration benefits
- Specific treatment within the criminal justice system (e.g., In 2002, the U.S. Supreme Court ruled in Atkins v. Virginia that executing the mentally retarded violates the Eighth Amendment's ban on cruel and unusual punishment)

Specific Tests

Doctors use a variety of tests to diagnose intellectual disabilities. Some of these testes include:

- Abnormal Denver developmental screening test
- Intelligence quotient (IQ) Test
- Tests used for adaptive behavior
  - Revised Denver Prescreening Developmental Questionnaire
  - Denver Developmental Screening Test
  - Kansas Infant Development Screen
  - Many others
- AAIDD's new Diagnostic Adaptive Behavior Scale (DABS)
  - Scheduled to be released in 2015 provides a comprehensive standardized assessment of adaptive behavior.

- Designed for use with individuals from 4 to 21 years old, DABS provides precise diagnostic information around the cutoff point where an individual is deemed to have "significant limitations" in adaptive behavior. The presence of such limitations is one of the measures of intellectual disability.

Signs and Symptoms of Intellectual Disabilities

There are many different signs of intellectual disability in children. Signs may appear during infancy, or they may not be noticeable until a child reaches school age. It often depends on the severity of the disability. Some of the most common signs of intellectual disability are:

- Rolling over, sitting up, crawling, or walking late
- Talking late or having trouble with talking
- Slow to master things like potty training, dressing, and feeding himself or herself
- Difficulty remembering things
- Inability to connect actions with consequences
- Behavior problems such as explosive tantrums
- Lack of or slow development of motor skills, language skills, and self-help skills, especially when compared to peers
- Difficulty with problem-solving or logical thinking
- Failure to grow intellectually or continued infant-like behavior
- Lack of curiosity
- Failure to adapt (adjust to new situations)
- Difficulty understanding and following social rules

In children with severe or profound intellectual disability, there may be other health problems as well. These problems may include seizures, mental disorders, motor handicaps, vision problems, or hearing problems.

Children with intellectual disabilities may take longer to learn to speak, walk, and take care of their personal needs such as dressing or eating. They are likely to have trouble learning in school. They will learn, but it will take them longer. There may be some things they will be unable to learn.

About 87% of people with intellectual disabilities will only be a little slower than average in new information and skills. When they are children, their limitations may not be obvious. Signs of intellectual disability can range from mild to severe.

Severity of Intellectual Disabilities

Mild: Affects approximately 85% of people with intellectual disabilities. In early childhood, mild intellectual disability (IQ 50–69) may not be obvious, and may not be

identified until children begin school. Even when poor academic performance is recognized, it may take expert assessment to distinguish mild intellectual disability from learning disability or emotional/behavioral disorders. People with mild intellectual disability are capable of learning reading and mathematics skills to approximately the level of a typical child aged nine to twelve. They can learn self-care and practical skills, such as cooking or using the local mass transit system. As individuals with intellectual disability reach adulthood, many learn to live independently and maintain gainful employment.

Moderate: About 10% of people with an intellectual disability have a moderate intellectual disability. Moderate intellectual disability (IQ 35–49) is nearly always apparent within the first years of life. Speech delays are particularly common signs of moderate ID. People with moderate intellectual disability need considerable supports in school, at home, and in the community in order to participate fully. While their academic potential is limited, they can learn simple health and safety skills and to participate in simple activities. As adults they may live with their parents, in a supportive group home, or even semi-independently with significant supportive services to help them, for example, manage their finances. As adults, they may work in a sheltered workshop.

Severe: About 3-4% of the population with intellectual disabilities. IQ scores range from 20-40. People with this severity can master very basic self-care and some communication skills. People with severe intellectual disability need more intensive support and supervision their entire lives. They may learn some activities of daily living. Some require full-time care by an attendant.

Profound: 1-2%. IQ scores range from 20-25. May be able to develop basic self-care skills and communication skills with appropriate training. People with profound intellectual disability need more intensive support and supervision their entire lives. They may learn some activities of daily living. Some require full-time care by an attendant. This is often caused by an accompanying neurological disorder.

Specific Types of Intellectual Disabilities

There are many different types of intellectual disabilities. Listed below are some of the many different disabilities and diseases related to intellectual disabilities.

Down Syndrome

Down syndrome is a chromosomal condition that is associated with intellectual disability, a characteristic facial appearance, and weak muscle tone (hypotonia) in infancy. All

affected individuals experience cognitive delays, but the intellectual disability is usually mild to moderate.

People with Down syndrome may have a variety of birth defects. About half of all affected children are born with a heart defect. Digestive abnormalities, such as a blockage of the intestine, are less common.

Individuals with Down syndrome have an increased risk of developing several medical conditions. These include gastroesophageal reflux, which is a backflow of acidic stomach contents into the esophagus, and celiac disease, which is an intolerance of a wheat protein called gluten. About 15 percent of people with Down syndrome have an underactive thyroid gland (hypothyroidism). The thyroid gland is a butterfly-shaped organ in the lower neck that produces hormones. Individuals with Down syndrome also have an increased risk of hearing and vision problems. Additionally, a small percentage of children with Down syndrome develop cancer of blood-forming cells (leukemia).

Delayed development and behavioral problems are often reported in children with Down syndrome. Affected individuals' speech and language develop later and more slowly than in children without Down syndrome, and affected individuals' speech may be more difficult to understand. Behavioral issues can include attention problems, obsessive/compulsive behavior, and stubbornness or tantrums. A small percentage of people with Down syndrome are also diagnosed with developmental conditions called autism spectrum disorders, which affect communication and social interaction.

People with Down syndrome often experience a gradual decline in thinking ability (cognition) as they age, usually starting around age 50. Down syndrome is also associated with an increased risk of developing Alzheimer disease, a brain disorder that results in a gradual loss of memory, judgment, and ability to function. Approximately half of adults with Down syndrome develop Alzheimer disease. Although Alzheimer disease is usually a disorder that occurs in older adults, people with Down syndrome usually develop this condition in their fifties or sixties.

How common is Down syndrome?

Down syndrome occurs in about 1 in 830 newborns. An estimated 250,000 people in the United States have this condition. Although women of any age can have a child with Down syndrome, the chance of having a child with this condition increases as a woman gets older.

What are the genetic changes related to Down syndrome?

Most cases of Down syndrome result from trisomy 21, which means each cell in the body has three copies of chromosome 21 instead of the usual two copies.

Less commonly, Down syndrome occurs when part of chromosome 21 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) in a parent or very early in fetal development. Affected people have two normal copies of chromosome 21 plus extra material from chromosome 21 attached to another chromosome, resulting in three copies of genetic material from chromosome 21. Affected individuals with this genetic change are said to have translocation Down syndrome.

A very small percentage of people with Down syndrome have an extra copy of chromosome 21 in only some of the body's cells. In these people, the condition is called mosaic Down syndrome.

Researchers believe that having extra copies of genes on chromosome 21 disrupts the course of normal development, causing the characteristic features of Down syndrome and the increased risk of health problems associated with this condition.

Can Down syndrome be inherited?

Most cases of Down syndrome are not inherited. When the condition is caused by trisomy 21, the chromosomal abnormality occurs as a random event during the formation of reproductive cells in a parent. The abnormality usually occurs in egg cells, but it occasionally occurs in sperm cells. An error in cell division called nondisjunction results in a reproductive cell with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of chromosome 21. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 21 in each of the body's cells.

People with translocation Down syndrome can inherit the condition from an unaffected parent. The parent carries a rearrangement of genetic material between chromosome 21 and another chromosome. This rearrangement is called a balanced translocation. No genetic material is gained or lost in a balanced translocation, so these chromosomal changes usually do not cause any health problems. However, as this translocation is passed to the next generation, it can become unbalanced. People who inherit an unbalanced translocation involving chromosome 21 may have extra genetic material from chromosome 21, which causes Down syndrome.

Like trisomy 21, mosaic Down syndrome is not inherited. It occurs as a random event during cell division early in fetal development. As a result, some of the body's cells have

the usual two copies of chromosome 21, and other cells have three copies of this chromosome.

Fragile X Syndrome

Fragile X syndrome is a genetic condition that causes a range of developmental problems including learning disabilities and cognitive impairment. Usually, males are more severely affected by this disorder than females.

Affected individuals usually have delayed development of speech and language by age 2. Most males with fragile X syndrome have mild to moderate intellectual disability, while about one-third of affected females are intellectually disabled. Children with fragile X syndrome may also have anxiety and hyperactive behavior such as fidgeting or impulsive actions. They may have attention deficit disorder (ADD), which includes an impaired ability to maintain attention and difficulty focusing on specific tasks. About one-third of individuals with fragile X syndrome have features of autism spectrum disorders that affect communication and social interaction. Seizures occur in about 15 percent of males and about 5 percent of females with fragile X syndrome.

Most males and about half of females with fragile X syndrome have characteristic physical features that become more apparent with age. These features include a long and narrow face, large ears, a prominent jaw and forehead, unusually flexible fingers, flat feet, and in males, enlarged testicles (macroorchidism) after puberty.

How common is Fragile X syndrome?

Fragile X syndrome occurs in approximately 1 in 4,000 males and 1 in 8,000 females.

What genes are related to fragile X syndrome?

Mutations in the FMR1 gene cause fragile X syndrome. The FMR1 gene provides instructions for making a protein called fragile X mental retardation 1 protein, or FMRP. This protein helps regulate the production of other proteins and plays a role in the development of synapses, which are specialized connections between nerve cells. Synapses are critical for relaying nerve impulses.

Nearly all cases of fragile X syndrome are caused by a mutation in which a DNA segment, known as the CGG triplet repeat, is expanded within the FMR1 gene. Normally, this DNA segment is repeated from 5 to about 40 times. In people with fragile X syndrome, however, the CGG segment is repeated more than 200 times. The abnormally expanded CGG segment turns off (silences) the FMR1 gene, which prevents the gene from producing FMRP. Loss or a shortage (deficiency) of this protein disrupts nervous system functions and leads to the signs and symptoms of fragile X syndrome.

Males and females with 55 to 200 repeats of the CGG segment are said to have an FMR1 gene premutation. Most people with a premutation are intellectually normal. In some cases, however, individuals with a premutation have lower than normal amounts of FMRP. As a result, they may have mild versions of the physical features seen in fragile X syndrome (such as prominent ears) and may experience emotional problems such as anxiety or depression. Some children with a premutation may have learning disabilities or autistic-like behavior. The premutation is also associated with an increased risk of disorders called fragile X-associated primary ovarian insufficiency (FXPOI) and fragile X-associated tremor/ataxia syndrome (FXTAS).

How do people inherit fragile X syndrome?

Fragile X syndrome is inherited in an X-linked dominant pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes. (The Y chromosome is the other sex chromosome.) The inheritance is dominant if one copy of the altered gene in each cell is sufficient to cause the condition. X-linked dominant means that in females (who have two X chromosomes), a mutation in one of the two copies of a gene in each cell is sufficient to cause the disorder. In males (who have only one X chromosome), a mutation in the only copy of a gene in each cell causes the disorder. In most cases, males experience more severe symptoms of the disorder than females.

In women, the FMR1 gene premutation on the X chromosome can expand to more than 200 CGG repeats in cells that develop into eggs. This means that women with the premutation have an increased risk of having a child with fragile X syndrome. By contrast, the premutation in men does not expand to more than 200 repeats as it is passed to the next generation. Men pass the premutation only to their daughters. Their sons receive a Y chromosome, which does not include the FMR1 gene.

Fetal Alcohol Syndrome

Fetal alcohol spectrum disorders (FASDs) are a group of conditions that can occur in a person whose mother drank alcohol during pregnancy. These effects can include physical problems and problems with behavior and learning. Often, a person with an FASD has a mix of these problems.

What are the causes?

FASDs are caused by a woman drinking alcohol during pregnancy. Alcohol in the mother's blood passes to the baby through the umbilical cord. When a woman drinks alcohol, so does her baby.

There is no known safe amount of alcohol during pregnancy or when trying to get pregnant. There is also no safe time to drink during pregnancy. Alcohol can cause

problems for a developing baby throughout pregnancy, including before a woman knows she's pregnant. All types of alcohol are equally harmful, including all wines and beer.

How can Fetal Alcohol Syndrome be prevented?

To prevent FASDs, a woman should not drink alcohol while she is pregnant, or when she might get pregnant. This is because a woman could get pregnant and not know for up to 4 to 6 weeks. In the United States, nearly half of pregnancies are unplanned.

If a woman is drinking alcohol during pregnancy, it is never too late to stop drinking. Because brain growth takes place throughout pregnancy, the sooner a woman stops drinking the safer it will be for her and her baby.

What are the signs and symptoms?

FASDs refer to the whole range of effects that can happen to a person whose mother drank alcohol during pregnancy. These conditions can affect each person in different ways, and can range from mild to severe.

A person with an FASD might have:

- Abnormal facial features, such as a smooth ridge between the nose and upper lip (this ridge is called the philtrum)
- Small head size
- Shorter than average height
- Low body weight
- Poor coordination
- Hyperactive behavior
- Difficulty with attention
- Poor memory
- Difficulty in school
- Learning disabilities
- Speech and language delays
- Low IQ
- Poor reasoning and judgment skills
- Vision or hearing problems
- Problems with the heart, kidneys, or bones

Phenylketonuria (PKU)

Phenylketonuria (commonly known as PKU) is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. Phenylalanine is a building block of proteins (an amino acid) that is obtained through the diet. It is found in all proteins and

in some artificial sweeteners. If PKU is not treated, phenylalanine can build up to harmful levels in the body, causing intellectual disability and other serious health problems.

The signs and symptoms of PKU vary from mild to severe. The most severe form of this disorder is known as classic PKU. Infants with classic PKU appear normal until they are a few months old. Without treatment, these children develop permanent intellectual disability. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common. Untreated individuals may have a musty or mouse-like odor as a side effect of excess phenylalanine in the body. Children with classic PKU tend to have lighter skin and hair than unaffected family members and are also likely to have skin disorders such as eczema.

Less severe forms of this condition, sometimes called variant PKU and non-PKU hyperphenylalaninemia, have a smaller risk of brain damage. People with very mild cases may not require treatment with a low-phenylalanine diet.

Babies born to mothers with PKU and uncontrolled phenylalanine levels (women who no longer follow a low-phenylalanine diet) have a significant risk of intellectual disability because they are exposed to very high levels of phenylalanine before birth. These infants may also have a low birth weight and grow more slowly than other children. Other characteristic medical problems include heart defects or other heart problems, an abnormally small head size (microcephaly), and behavioral problems. Women with PKU and uncontrolled phenylalanine levels also have an increased risk of pregnancy loss.

How common is PKU?

The occurrence of PKU varies among ethnic groups and geographic regions worldwide. In the United States, PKU occurs in 1 in 10,000 to 15,000 newborns. Most cases of PKU are detected shortly after birth by newborn screening, and treatment is started promptly. As a result, the severe signs and symptoms of classic PKU are rarely seen.

What genes are related to PKU?

Mutations in the PAH gene cause phenylketonuria.

The PAH gene provides instructions for making an enzyme called phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine to other important compounds in the body. If gene mutations reduce the activity of phenylalanine hydroxylase, phenylalanine from the diet is not processed effectively. As a result, this amino acid can build up to toxic levels in the blood and other tissues. Because nerve cells in the brain are particularly sensitive to phenylalanine levels, excessive amounts of this substance can cause brain damage.

Classic PKU, the most severe form of the disorder, occurs when phenylalanine hydroxylase activity is severely reduced or absent. People with untreated classic PKU

have levels of phenylalanine high enough to cause severe brain damage and other serious medical problems. Mutations in the PAH gene that allow the enzyme to retain some activity result in milder versions of this condition, such as variant PKU or non-PKU hyperphenylalaninemia.

Changes in other genes may influence the severity of PKU, but little is known about these additional genetic factors.

How do people inherit PKU?

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Williams Syndrome

Williams syndrome is a developmental disorder that affects many parts of the body. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems.

People with Williams syndrome typically have difficulty with visual-spatial tasks such as drawing and assembling puzzles, but they tend to do well on tasks that involve spoken language, music, and learning by repetition (rote memorization). Affected individuals have outgoing, engaging personalities and tend to take an extreme interest in other people. Attention deficit disorder (ADD), problems with anxiety, and phobias are common among people with this disorder.

Young children with Williams syndrome have distinctive facial features including a broad forehead, a short nose with a broad tip, full cheeks, and a wide mouth with full lips. Many affected people have dental problems such as teeth that are small, widely spaced, crooked, or missing. In older children and adults, the face appears longer and more gaunt.

A form of cardiovascular disease called supravalvular aortic stenosis (SVAS) occurs frequently in people with Williams syndrome. Supravalvular aortic stenosis is a narrowing of the large blood vessel that carries blood from the heart to the rest of the body (the aorta). If this condition is not treated, the aortic narrowing can lead to shortness of breath, chest pain, and heart failure. Other problems with the heart and blood vessels, including high blood pressure (hypertension), have also been reported in people with Williams syndrome.

Additional signs and symptoms of Williams syndrome include abnormalities of connective tissue (tissue that supports the body's joints and organs) such as joint problems and soft, loose skin. Affected people may also have increased calcium levels in

the blood (hypercalcemia) in infancy, developmental delays, problems with coordination, and short stature. Medical problems involving the eyes and vision, the digestive tract, and the urinary system are also possible.

How common is Williams syndrome?

Williams syndrome affects an estimated 1 in 7,500 to 10,000 people.

What are the genetic changes related to Williams syndrome?

Williams syndrome is caused by the deletion of genetic material from a specific region of chromosome 7. The deleted region includes 26 to 28 genes, and researchers believe that a loss of several of these genes probably contributes to the characteristic features of this disorder.

CLIP2, ELN, GTF2I, GTF2IRD1, and LIMK1 are among the genes that are typically deleted in people with Williams syndrome. Researchers have found that loss of the ELN gene is associated with the connective tissue abnormalities and cardiovascular disease (specifically supravalvular aortic stenosis) found in many people with this disease. Studies suggest that deletion of CLIP2, GTF2I, GTF2IRD1, LIMK1, and perhaps other genes may help explain the characteristic difficulties with visual-spatial tasks, unique behavioral characteristics, and other cognitive difficulties seen in people with Williams syndrome. Loss of the GTF2IRD1 gene may also contribute to the distinctive facial features often associated with this condition.

Researchers believe that the presence or absence of the NCF1 gene on chromosome 7 is related to the risk of developing hypertension in people with Williams syndrome. When the NCF1 gene is included in the part of the chromosome that is deleted, affected individuals are less likely to develop hypertension. Therefore, the loss of this gene appears to be a protective factor. People with Williams syndrome whose NCF1 gene is not deleted have a higher risk of developing hypertension.

The relationship between other genes in the deleted region of chromosome 7 and the signs and symptoms of Williams syndrome is under investigation or unknown.

Can Williams syndrome be inherited?

Most cases of Williams syndrome are not inherited but occur as random events during the formation of reproductive cells (eggs or sperm) in a parent of an affected individual. These cases occur in people with no history of the disorder in their family.

Williams syndrome is considered an autosomal dominant condition because one copy of the altered chromosome 7 in each cell is sufficient to cause the disorder. In a small

percentage of cases, people with Williams syndrome inherit the chromosomal deletion from a parent with the condition.

Sotos Syndrome

Sotos syndrome is a disorder characterized by a distinctive facial appearance, overgrowth in childhood, and learning disabilities or delayed development of mental and movement abilities. Characteristic facial features include a long, narrow face; a high forehead; flushed (reddened) cheeks; and a small, pointed chin. In addition, the outside corners of the eyes may point downward (down-slanting palpebral fissures). This facial appearance is most notable in early childhood. Affected infants and children tend to grow quickly; they are significantly taller than their siblings and peers and have an unusually large head. However, adult height is usually in the normal range.

People with Sotos syndrome often have intellectual disability, and most also have behavioral problems. Frequent behavioral issues include attention deficit hyperactivity disorder (ADHD), phobias, obsessions and compulsions, tantrums, and impulsive behaviors. Problems with speech and language are also common. Affected individuals often have a stutter, a monotone voice, and problems with sound production. Additionally, weak muscle tone (hypotonia) may delay other aspects of early development, particularly motor skills such as sitting and crawling.

Other signs and symptoms of Sotos syndrome can include an abnormal side-to-side curvature of the spine (scoliosis), seizures, heart or kidney defects, hearing loss, and problems with vision. Some infants with this disorder experience yellowing of the skin and whites of the eyes (jaundice) and poor feeding.

A small percentage of people with Sotos syndrome have developed cancer, most often in childhood, but no single form of cancer occurs most frequently with this condition. It remains uncertain whether Sotos syndrome increases the risk of specific types of cancer. If people with this disorder have an increased cancer risk, it is only slightly greater than that of the general population.

How common is Sotos syndrome?

Sotos syndrome is reported to occur in 1 in 10,000 to 14,000 newborns. Because many of the features of Sotos syndrome can be attributed to other conditions, many cases of this disorder are likely not properly diagnosed, so the true incidence may be closer to 1 in 5,000.

What genes are related to Sotos syndrome?

Mutations in the NSD1 gene are the primary cause of Sotos syndrome, accounting for up to 90 percent of cases. Other genetic causes of this condition have not been identified.

The NSD1 gene provides instructions for making a protein that functions as a histone methyltransferase. Histone methyltransferases are enzymes that modify structural proteins called histones, which attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones (a process called methylation), histone methyltransferases regulate the activity of certain genes and can turn them on and off as needed. The NSD1 protein controls the activity of genes involved in normal growth and development, although most of these genes have not been identified.

Genetic changes involving the NSD1 gene prevent one copy of the gene from producing any functional protein. Research suggests that a reduced amount of NSD1 protein disrupts the normal activity of genes involved in growth and development. However, it remains unclear exactly how a shortage of this protein during development leads to overgrowth, learning disabilities, and the other features of Sotos syndrome.

How do people inherit Sotos syndrome?

About 95 percent of Sotos syndrome cases occur in people with no history of the disorder in their family. Most of these cases result from new mutations involving the NSD1 gene.

A few families have been described with more than one affected family member. These cases helped researchers determine that Sotos syndrome has an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means one copy of the altered gene in each cell is sufficient to cause the disorder.

Trisomy 18

Trisomy 18, also called Edwards syndrome, is a chromosomal condition associated with abnormalities in many parts of the body. Individuals with trisomy 18 often have slow growth before birth (intrauterine growth retardation) and a low birth weight. Affected individuals may have heart defects and abnormalities of other organs that develop before birth. Other features of trisomy 18 include a small, abnormally shaped head; a small jaw and mouth; and clenched fists with overlapping fingers. Due to the presence of several life-threatening medical problems, many individuals with trisomy 18 die before birth or within their first month. Five to 10 percent of children with this condition live past their first year, and these children often have severe intellectual disability.

How common is trisomy 18?

Trisomy 18 occurs in about 1 in 5,000 live-born infants; it is more common in pregnancy, but many affected fetuses do not survive to term. Although women of all ages can have a

child with trisomy 18, the chance of having a child with this condition increases as a woman gets older.

What are the genetic changes related to trisomy 18?

Most cases of trisomy 18 result from having three copies of chromosome 18 in each cell in the body instead of the usual two copies. The extra genetic material disrupts the normal course of development, causing the characteristic features of trisomy 18.

Approximately 5 percent of people with trisomy 18 have an extra copy of chromosome 18 in only some of the body's cells. In these people, the condition is called mosaic trisomy 18. The severity of mosaic trisomy 18 depends on the type and number of cells that have the extra chromosome. The development of individuals with this form of trisomy 18 may range from normal to severely affected.

Very rarely, part of the long (q) arm of chromosome 18 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) or very early in embryonic development. Affected individuals have two copies of chromosome 18, plus the extra material from chromosome 18 attached to another chromosome. People with this genetic change are said to have partial trisomy 18. If only part of the q arm is present in three copies, the physical signs of partial trisomy 18 may be less severe than those typically seen in trisomy 18. If the entire q arm is present in three copies, individuals may be as severely affected as if they had three full copies of chromosome 18.

Can trisomy 18 be inherited?

Most cases of trisomy 18 are not inherited, but occur as random events during the formation of eggs and sperm. An error in cell division called nondisjunction results in a reproductive cell with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of chromosome 18. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 18 in each of the body's cells.

Mosaic trisomy 18 is also not inherited. It occurs as a random event during cell division early in embryonic development. As a result, some of the body's cells have the usual two copies of chromosome 18, and other cells have three copies of this chromosome.

Partial trisomy 18 can be inherited. An unaffected person can carry a rearrangement of genetic material between chromosome 18 and another chromosome. This rearrangement is called a balanced translocation because there is no extra material from chromosome 18. Although they do not have signs of trisomy 18, people who carry this type of balanced translocation are at an increased risk of having children with the condition.

Rett Syndrome

Rett syndrome is a brain disorder that occurs almost exclusively in girls. The most common form of the condition is known as classic Rett syndrome. After birth, girls with classic Rett syndrome have 6 to 18 months of apparently normal development before developing severe problems with language and communication, learning, coordination, and other brain functions. Early in childhood, affected girls lose purposeful use of their hands and begin making repeated hand wringing, washing, or clapping motions. They tend to grow more slowly than other children and have a small head size (microcephaly). Other signs and symptoms that can develop include breathing abnormalities, seizures, an abnormal side-to-side curvature of the spine (scoliosis), and sleep disturbances.

Researchers have described several variant or atypical forms of Rett syndrome, which can be milder or more severe than the classic form.

How common is Rett syndrome?

This condition affects an estimated 1 in 8,500 females.

What genes are related to Rett syndrome?

Classic Rett syndrome and some variant forms of the condition are caused by mutations in the MECP2 gene. This gene provides instructions for making a protein (MeCP2) that is critical for normal brain function. Although the exact function of the MeCP2 protein is unclear, it is likely involved in maintaining connections (synapses) between nerve cells (neurons). It may also be necessary for the normal function of other types of brain cells.

The MeCP2 protein is thought to help regulate the activity of genes in the brain. This protein may also control the production of different versions of certain proteins in brain cells. Mutations in the MECP2 gene alter the MeCP2 protein or result in the production of less protein, which appears to disrupt the normal function of neurons and other cells in the brain. Specifically, studies suggest that changes in the MeCP2 protein may reduce the activity of certain neurons and impair their ability to communicate with one another. It is unclear how these changes lead to the specific features of Rett syndrome.

Several conditions with signs and symptoms overlapping those of Rett syndrome have been found to result from mutations in other genes. These conditions, including FOXG1 syndrome, were previously thought to be variant forms of Rett syndrome. However, doctors and researchers have identified some important differences between the conditions, so they are now usually considered to be separate disorders.

How do people inherit Rett syndrome?

In more than 99 percent of people with Rett syndrome, the condition is not inherited and there is no history of the disorder in their family. Many of these cases result from new mutations in the MECP2 gene.

A few families with more than one affected family member have been described. These cases helped researchers determine that classic Rett syndrome and variants caused by MECP2 gene mutations have an X-linked dominant pattern of inheritance. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes. The inheritance is dominant if one copy of the altered gene in each cell is sufficient to cause the condition.

Males with mutations in the MECP2 gene often die in infancy. However, a small number of males with a genetic change involving MECP2 have developed signs and symptoms similar to those of Rett syndrome, including intellectual disability, seizures, and movement problems. In males, this condition is described as MECP2-related severe neonatal encephalopathy.

Prevention

Certain causes of intellectual disability are preventable. The most common of these is fetal alcohol syndrome. Pregnant women shouldn't drink alcohol. Getting proper prenatal care, taking a prenatal vitamin, and getting vaccinated against certain infectious diseases can also lower the risk that your child will be born with intellectual disabilities. Preventing exposure to lead, mercury, and other toxins reduces the risk of disability. Teaching women about the risks of alcohol and drugs during pregnancy can also help reduce risk.

Certain tests, such as ultrasound and amniocentesis, can also be performed during pregnancy to look for problems associated with intellectual disability. Although these tests may identify problems before birth, they cannot correct them.

Treatment and Medication Options

Since treatment is not designed to cure intellectual disabilities, there are no specific medications. Therapy goals are to reduce safety risks, and teach appropriate life skills. Interventions are based on the specific needs of the individuals and their families, with the main goal of developing the persons potential to the fullest.

For babies and toddlers, early intervention programs are available. A team of professionals works with parents to write an Individualized Family Service Plan, or IFSP. This document outlines the child's specific needs and what services will help the child

thrive. Early intervention may include speech therapy, occupational therapy, physical therapy, family counseling, training with special assistive devices, or nutrition services.

School-aged children with intellectual disabilities (including preschoolers) are eligible for special education for free through the public school system. This is mandated by the Individuals With Disabilities Education Act (IDEA). Parents and educators work together to create an Individualized Education Program, or IEP, which outlines the child's needs and the services the child will receive at school. The point of special education is to make adaptations, accommodations, and modifications that allow a child with an intellectual disability to succeed in the classroom.

It is important for a specialist to evaluate the person for other physical and mental health problems. Persons with intellectual disability are often helped with behavioral counseling.

The goal of treatment is to develop the person's potential to the fullest. Special education and training may begin as early as infancy. This includes social skills to help the person function as normally as possible.

Parents can also support and help their child who has an intellectual disability. WebMD suggest several steps parents can do to help their child. These steps are:

> Learn everything you can about intellectual disabilities. The more you know, the better advocate you can be for your child.
>
> Encourage your child's independence. Let your child try new things and encourage your child to do things by himself or herself. Provide guidance when it's needed and give positive feedback when your child does something well or masters something new.
>
> Get your child involved in group activities. Taking an art class or participating in Scouts will help your child build social skills.
>
> Stay involved. By keeping in touch with your child's teachers, you'll be able to follow his or her progress and reinforce what your child is learning at school through practice at home.
>
> Get to know other parents of intellectually disabled children. They can be a great source of advice and emotional support.

Specific Needs

The multifaceted habilitation plan for the individual requires input from many different sources. These sources are care providers from multiple disciplines, such as special educators, language therapists, behavioral therapists, occupational therapists, and community services that provide social support and care for families of people with intellectual disability (ID). All of these sources along with the family work together to develop a comprehensive, individualized program for the child. This is started as soon the

diagnosis of ID is suspected. Physical activity and obesity are major concerns for people with ID, and very few programs exist that target healthy lifestyles in those with ID. Annual counseling and referral on these issues to community agencies and programs are recommended.

The child's strengths and weaknesses must be assessed to determine what kind of support is needed. Factors such as physical disabilities, personality problems, mental illness, as well as interpersonal skills are all taken into consideration.

All children with ID benefit from special education. The federal Individuals with Disabilities Education Act (IDEA) requires public schools to provide a free and appropriate education to those children and adolescents with ID or other developmental disorders. Education must be provided in the most inclusive setting possible, so that the child can have every opportunity to interact with non-disabled peers and have equal access to community resources.

Children with IDs usually do the best when living at home. Some families, however, are not able to provide adequate care at home, especially for children with severe disabilities. The decision to care for the children at home is difficult and requires extensive discussion between the family and their entire support team. Having a child with severe disabilities living at home requires dedicated care that some parents are unable to provide. The family might need to seek psychological support. Social workers can organize services to help assist the family in caring for their child. Day care centers, housekeepers, child caregivers, and respite care facilities, may also help provide care. Most adults with IDs live in community-based residences that provide the services needed to most appropriately help with their needs, as well as work and recreational opportunities.

Therapeutic Recreation Implications

The purpose of TR is to facilitate the development, maintenance, and expression of an appropriate leisure lifestyle for the person with physical, intellectual, mental, emotional, or social limitations. An important part of independent function is having and maintaining this lifestyle on a daily basis.

The main point of treatment is to develop interventions and treatments that assist people with intellectual disabilities in developing the skills needed for participation in leisure pursuits. Interventions need to focus on the development of cognitive skills. They should also focus on improving the quality of life for individuals. Interventions should increase physical, social, and emotional development related to leisure participation.

Resources

Support groups can be very helpful for the person with a intellectual disability, as well as their family. Support group information and other helpful information can be found on many websites. Some of these websites include:

- American Association on Intellectual and Developmental Disabilities http://www.aaidd.org
- The Arc http://www.thearc.org
- National Association for Down Syndrome http://www.nads.org
- National Dissemination Center for Children with Disabilities http://www.nichcy.org
- The national Association of Councils on Developmental Disabilities
- Presidents Committee for People with Intellectual Disabilities
- The Pacesetter
- Parents Helping Parents

Autism

What is Autism?

Autism is a brain disorder that often makes it hard to communicate with and relate to others. With autism, the different areas of the brain fail to work together.

Most people with autism will always have some trouble relating to others. But early diagnosis and treatment have helped more and more people who have autism to reach their full potential.

Autism spectrum disorder includes conditions previously called autism, pervasive developmental disorder, and Asperger's syndrome. The causes are not known.

According to Autism Speaks, Autism spectrum disorder (ASD) and autism are both general terms for a group of complex disorders of brain development. These disorders are characterized, in varying degrees, by difficulties in social interaction, verbal and nonverbal communication and repetitive behaviors. With the May 2013 publication of the DSM-5 diagnostic manual, all autism disorders were merged into one umbrella diagnosis of ASD. Previously, they were recognized as distinct subtypes, including autistic disorder, childhood disintegrative disorder, pervasive developmental disorder-not otherwise specified (PDD-NOS) and Asperger syndrome.

ASD can be associated with intellectual disability, difficulties in motor coordination and attention and physical health issues such as sleep and gastrointestinal disturbances. Some persons with ASD excel in visual skills, music, math and art.

Autism appears to have its roots in very early brain development. However, the most obvious signs of autism and symptoms of autism tend to emerge between 2 and 3 years of age. Autism Speaks continues to fund research on effective methods for earlier diagnosis, as early intervention with proven behavioral therapies can improve outcomes. Increasing autism awareness is a key aspect of this work and one in which our families and volunteers play an invaluable role.

What Does It Mean to Be “On the Spectrum”?

Each individual with autism is unique. Many of those on the autism spectrum have exceptional abilities in visual skills, music and academic skills. About 40 percent have average to above average intellectual abilities. Indeed, many persons on the spectrum take deserved pride in their distinctive abilities and “atypical” ways of viewing the world. Others with autism have significant disability and are unable to live independently. About 25 percent of individuals with ASD are nonverbal but can learn to communicate using other means. Autism Speaks’ mission is to improve the lives of all those on the autism spectrum. For some, this means the development and delivery of more effective

treatments that can address significant challenges in communication and physical health. For others, it means increasing acceptance, respect and support.

Prevalence

Autism statistics from the U.S. Centers for Disease Control and Prevention (CDC) identify around 1 in 68 American children as on the autism spectrum–a ten-fold increase in prevalence in 40 years. Careful research shows that this increase is only partly explained by improved diagnosis and awareness. Studies also show that autism is four to five times more common among boys than girls. An estimated 1 out of 42 boys and 1 in 189 girls are diagnosed with autism in the United States.

ASD affects over 3 million individuals in the U.S. and tens of millions worldwide. Moreover, government autism statistics suggest that prevalence rates have increased 10 to 17 percent annually in recent years. There is no established explanation for this continuing increase, although improved diagnosis and environmental influences are two reasons often considered.

Causes

Autism tends to run in families, so experts think it may be something that you inherit. Scientists are trying to find out exactly which genes may be responsible for passing down autism in families.

Other studies are looking at whether autism can be caused by other medical problems or by something in your child's surroundings.

False claims in the news have made some parents concerned about a link between autism and vaccines. But studies have found no link between vaccines and autism. It's important to make sure that your child gets all childhood vaccines. They help keep your child from getting serious diseases that can cause harm or even death.

We do not know all of the causes of ASD. However, we have learned that there are likely many causes for multiple types of ASD. There may be many different factors that make a child more likely to have an ASD, including environmental, biologic and genetic factors.

- Most scientists agree that genes are one of the risk factors that can make a person more likely to develop ASD.
- Children who have a sibling with ASD are at a higher risk of also having ASD.
- ASD tends to occur more often in people who have certain genetic or chromosomal conditions, such as fragile X syndrome or tuberous sclerosis.
- When taken during pregnancy, the prescription drugs valproic acid and thalidomide have been linked with a higher risk of ASD.
- There is some evidence that the critical period for developing ASD occurs before, during, and immediately after birth.

- Children born to older parents are at greater risk for having ASD.

Symptoms

The severity of symptoms varies greatly, but all people with autism have some core symptoms in the areas of:

Social interactions and relationships. Symptoms may include:

- Significant problems developing nonverbal communication skills, such as eye-to-eye gazing, facial expressions, and body posture.
- Failure to establish friendships with children the same age.
- Lack of interest in sharing enjoyment, interests, or achievements with other people.
- Lack of empathy. People with autism may have difficulty understanding another person's feelings, such as pain or sorrow.

Verbal and nonverbal communication. Symptoms may include:

- Delay in, or lack of, learning to talk. As many as 40% of people with autism never speak.[1]
- Problems taking steps to start a conversation. Also, people with autism have difficulties continuing a conversation after it has begun.
- Stereotyped and repetitive use of language. People with autism often repeat over and over a phrase they have heard previously (echolalia).
- Difficulty understanding their listener's perspective. For example, a person with autism may not understand that someone is using humor. They may interpret the communication word for word and fail to catch the implied meaning.

Limited interests in activities or play. Symptoms may include:

- An unusual focus on pieces. Younger children with autism often focus on parts of toys, such as the wheels on a car, rather than playing with the entire toy.
- Preoccupation with certain topics. For example, older children and adults may be fascinated by video games, trading cards, or license plates.
- A need for sameness and routines. For example, a child with autism may always need to eat bread before salad and insist on driving the same route every day to school.
- Stereotyped behaviors. These may include body rocking and hand flapping.

Some more specific symptoms of ASD include:

- not point at objects to show interest (for example, not point at an airplane flying over)
- not look at objects when another person points at them
- have trouble relating to others or not have an interest in other people at all

- avoid eye contact and want to be alone
- have trouble understanding other people's feelings or talking about their own feelings
- prefer not to be held or cuddled, or might cuddle only when they want to
- appear to be unaware when people talk to them, but respond to other sounds
- be very interested in people, but not know how to talk, play, or relate to them
- repeat or echo words or phrases said to them, or repeat words or phrases in place of normal language
- have trouble expressing their needs using typical words or motions
- not play "pretend" games (for example, not pretend to "feed" a doll)
- repeat actions over and over again
- have trouble adapting when a routine changes
- have unusual reactions to the way things smell, taste, look, feel, or sound
- lose skills they once had (for example, stop saying words they were using)

Children

Symptoms of autism are usually noticed first by parents and other caregivers sometime during the child's first 3 years. Although autism is present at birth (congenital), signs of the disorder can be difficult to identify or diagnose during infancy. Parents often become concerned when their toddler does not like to be held; does not seem interested in playing certain games, such as peekaboo; and does not begin to talk. Sometimes, a child with autism will start to talk at the same time as other children the same age, then lose his or her language skills. Parents also may be confused about their child's hearing abilities. It often seems that a child with autism does not hear, yet at other times, he or she may appear to hear a distant background noise, such as the whistle of a train.

Symptoms of autism include:

- A delay in learning to talk, or not talking at all. A child may seem to be deaf, even though hearing tests are normal.
- Repeated and overused types of behavior, interests, and play. Examples include repeated body rocking, unusual attachments to objects, and getting very upset when routines change.

There is no "typical" person with autism. People can have many different kinds of behaviors, from mild to severe. Parents often say that their child with autism prefers to play alone and does not make eye contact with other people.

Autism may also include other problems:

- Many children with autism have below-normal intelligence.
- Teenagers with autism often become depressed and have a lot of anxiety, especially if they have average or above-average intelligence.

- Some children get a seizure disorder such as epilepsy by their teen years.

Teenagers

During the teen years, the patterns of behavior often change. Many teens gain skills but still lag behind in their ability to relate to and understand others. Puberty and emerging sexuality may be more difficult for teens who have autism than for others this age. Teens are at an increased risk for developing problems related to depression, anxiety, and epilepsy.

Adults

Some adults with autism are able to work and live on their own. The degree to which an adult with autism can lead an independent life is related to intelligence and ability to communicate. At least 33% are able to achieve at least partial independence.[2]

Some adults with autism need a lot of assistance, especially those with low intelligence who are unable to speak. Part- or full-time supervision can be provided by residential treatment programs. At the other end of the spectrum, adults with high-functioning autism are often successful in their professions and able to live independently, although they typically continue to have some difficulties relating to other people. These individuals usually have average to above-average intelligence.

Other symptoms

Many people with autism have symptoms similar to attention deficit hyperactivity disorder (ADHD). But these symptoms, especially problems with social relationships, are more severe for people with autism.

About 10% of people with autism have some form of savant skills—special limited gifts such as memorizing lists, calculating calendar dates, drawing, or musical ability.

Many people with autism have unusual sensory perceptions. For example, they may describe a light touch as painful and deep pressure as providing a calming feeling. Others may not feel pain at all. Some people with autism have strong food likes and dislikes and unusual preoccupations.

Sleep problems occur in about 40% to 70% of people with autism.

Almost half of the children who have autism spectrum disorders tend to "wander off" from a caregiver, or "elope." For many caregivers of these children, elopement is one of the most stressful behaviors they must learn to cope with. Studies show that behavioral assessment interventions, such as applied behavioral analysis, may reduce the number of times a child wanders off.

Bottom of Form

Diagnosing Autism

Diagnosing autism spectrum disorder (ASD) can be difficult, since there is no medical test, like a blood test, to diagnose the disorders. Doctors look at the child's behavior and development to make a diagnosis.

ASD can sometimes be detected at 18 months or younger. By age 2, a diagnosis by an experienced professional can be considered very reliable.[1] However, many children do not receive a final diagnosis until much older. This delay means that children with an ASD might not get the help they need.

Diagnosing an ASD takes two steps:

- Developmental Screening
- Comprehensive Diagnostic Evaluation

The American Academy of Pediatrics (AAP) recommends screening children for autism during regularly scheduled well-child visits.[5] This policy helps doctors identify signs of autism early in its course. Early diagnosis and treatment can help the child reach his or her full potential.

When a developmental delay is recognized in a child, further testing can help a doctor find out whether the problem is related to autism, another autism spectrum disorder, or a condition with similar symptoms, such as language delays or avoidant personality disorder. If your primary care provider does not have specific training or experience in developmental problems, he or she may refer your child to a specialist—such as a developmental pediatrician, a psychiatrist, a speech therapist, a psychologist, or a child psychiatrist—for the additional testing.

There are guidelines your doctor will use to see if your child has symptoms of autism. The guidelines put symptoms into categories such as:

- **Social interactions and relationships.** For example, a child may have trouble making eye contact. People with autism may have a hard time understanding someone else's feelings, such as pain or sadness.
- **Verbal and nonverbal communication.** For example, a child may never speak. Or he or she may often repeat a certain phrase over and over.
- **Limited interests in activities or play.** For example, younger children often focus on parts of toys rather than playing with the whole toy. Older children and adults may be fascinated by certain topics, like trading cards or license plates.

Doctors assess the following areas:

- **Behavioral assessments.** Various guidelines and questionnaires are used to help a doctor determine the specific type of developmental delay a child has. These include:
    - Medical history. During the medical history interview, a doctor asks general questions about a child's development, such as whether a child shows parents things by pointing to objects. Young children with autism often point to items they want, but do not point to show parents an item and then check to see if parents are looking at the item being pointed out.
    - Diagnostic guidelines for autism. The American Association of Childhood and Adolescent Psychiatry (AACAP) has established guidelines for diagnosing autism. The criteria are designed so a doctor can assess a child's behavior relating to core symptoms of autism.
    - Clinical observations. A doctor may want to observe the developmentally delayed child in different situations. The parents may be asked to interpret whether certain behaviors are usual for the child in those circumstances.
    - Developmental and intelligence tests. The AACAP also recommends that tests be given to evaluate whether a child's developmental delays affect his or her ability to think and make decisions.
- **Physical assessments and laboratory tests.** Other tests may be used to determine whether a physical problem may be causing symptoms. These tests include:
    - Physical exam, including head circumference, weight, and height measurements, to determine whether the child has a normal growth pattern.
    - Hearing tests, to determine whether hearing problems may be causing developmental delays, especially those related to social skills and language use.
    - Testing for lead poisoning, especially if a condition called pica (in which a person craves substances that are not food, such as dirt or flecks of old paint) is present. Children with developmental delays usually continue putting items in their mouth after this stage has passed in normally developing children. This practice can result in lead poisoning, which should be identified and treated as soon as possible.

Other lab tests may be done under specific circumstances. These tests include:

- Chromosomal analysis, which may be done if intellectual disability is present or there is a family history of intellectual disability. For example, fragile X syndrome, which causes a range of below-normal intelligence problems as well as autistic-like behaviors, can be identified with a chromosomal analysis.
- An electroencephalograph (EEG), which is done if there are symptoms of seizures, such as a history of staring spells or if a person reverts to less mature behavior (developmental regression).
- MRI, which may be done if there are signs of differences in the structure of the brain.

Developmental Screening

Developmental screening is a short test to tell if children are learning basic skills when they should, or if they might have delays. During developmental screening the doctor might ask the parent some questions or talk and play with the child during an exam to see how she learns, speaks, behaves, and moves. A delay in any of these areas could be a sign of a problem.

All children should be screened for developmental delays and disabilities during regular well-child doctor visits at:

- 9 months
- 18 months
- 24 or 30 months
- Additional screening might be needed if a child is at high risk for developmental problems due to preterm birth, low birth weight or other reasons.

In addition, all children should be screened specifically for ASD during regular well-child doctor visits at:

- 18 months
- 24 months
- Additional screening might be needed if a child is at high risk for ASD (e.g., having a sister, brother or other family member with an ASD) or if behaviors sometimes associated with ASD are present

It is important for doctors to screen all children for developmental delays, but especially to monitor those who are at a higher risk for developmental problems due to preterm birth, low birth weight, or having a brother or sister with an ASD.

If your child's doctor does not routinely check your child with this type of developmental screening test, ask that it be done.

If the doctor sees any signs of a problem, a comprehensive diagnostic evaluation is needed.

Comprehensive Diagnostic Evaluation

The second step of diagnosis is a comprehensive evaluation. This thorough review may include looking at the child's behavior and development and interviewing the parents. It may also include a hearing and vision screening, genetic testing, neurological testing, and other medical testing.

In some cases, the primary care doctor might choose to refer the child and family to a specialist for further assessment and diagnosis. Specialists who can do this type of evaluation include:

- Developmental Pediatricians (doctors who have special training in child development and children with special needs)
- Child Neurologists (doctors who work on the brain, spine, and nerves)
- Child Psychologists or Psychiatrists (doctors who know about the human mind)

Early Detection

All doctors who see infants and children for well-child visits should watch for early signs of developmental disorders. Developmental screening tools, such as the Ages and Stages Questionnaire or the Modified Checklist for Autism in Toddlers (M-CHAT), can help assess behavior.

If a doctor discovers the following obvious signs of developmental delays, the child should immediately be evaluated:

- No babbling, pointing, or other gestures by 12 months
- No single words by 16 months
- No 2-word spontaneous phrases by 24 months, with the exception of repeated phrases (echolalia)
- **Any** loss of any language or social skills at **any** age

If there are no obvious signs of developmental delays or any unusual indications from the screening tests, most infants and children do not need further evaluation until the next well-child visit.

But children who have a sibling with autism should continue to be closely monitored, because they are at increased risk for autism and other developmental problems.

When socialization, learning, or behavior problems develop in a person at any time or at any age, he or she should also be evaluated.

Types of ASD

Asperger's syndrome

Asperger's syndrome, also called Asperger's disorder, is a type of pervasive developmental disorder (PDD). PDDs are a group of conditions that involve delays in the development of many basic skills, most notably the ability to socialize with others, to communicate, and to use imagination.

Although Asperger's syndrome is similar in some ways to autism -- another, more severe type of PDD -- there are some important differences. Children with Asperger's syndrome typically function better than do those with autism. In addition, children with Asperger's syndrome generally have normal intelligence and near-normal language development, although they may develop problems communicating as they get older.

Asperger's syndrome was named for the Austrian doctor, Hans Asperger, who first described the disorder in 1944. However, Asperger's syndrome was not recognized as a unique disorder until much later.

What are the symptoms of Asperger's syndrome?

The symptoms of Asperger's syndrome vary and can range from mild to severe. Common symptoms include:

- **Problems with social skills:** Children with Asperger's syndrome generally have difficulty interacting with others and often are awkward in social situations. They generally do not make friends easily. They have difficulty initiating and maintaining conversation.
- **Eccentric or repetitive behaviors:** Children with this condition may develop odd, repetitive movements, such as hand wringing or finger twisting.
- **Unusual preoccupations or rituals:** A child with Asperger's syndrome may develop rituals that he or she refuses to alter, such as getting dressed in a specific order.
- **Communication difficulties:** People with Asperger's syndrome may not make eye contact when speaking with someone. They may have trouble using facial expressions and gestures, and understanding body language. They also tend to have problems understanding language in context and are very literal in their use of language.
- **Limited range of interests:** A child with Asperger's syndrome may develop an intense, almost obsessive, interest in a few areas, such as sports schedules, weather, or maps.
- **Coordination problems:** The movements of children with Asperger's syndrome may seem clumsy or awkward.
- **Skilled or talented:** Many children with Asperger's syndrome are exceptionally talented or skilled in a particular area, such as music or math.

What causes Asperger's syndrome?

The exact cause of Asperger's syndrome is not known. However, the fact that it tends to run in families suggests that it may be inherited (passed on from parent to child).

How common is Asperger's syndrome?

Asperger's syndrome has only recently been recognized as a unique disorder. For that reason, the exact number of people with the disorder is unknown. While it is more common than autism, estimates for the United States and Canada range from 1 in every 250 children to 1 in every 10,000. It is four times more likely to occur in males than in females and usually is first diagnosed in children between the ages of 2 and 6.

How is Asperger's syndrome diagnosed?

If symptoms are present, the doctor will begin an evaluation by performing a complete medical history and physical and neurological exam. Many individuals with Asprger's have low muscle tone and dyspraxia, or coordination issues. Although there are no tests for Asperger's syndrome, the doctor may use various tests -- such as X-rays and blood tests -- to determine if there is a physical disorder causing the symptoms.

If no physical disorder is found, the child may be referred to a specialist in childhood development disorders, such as a child and adolescent psychiatrist or psychologist, pediatric neurologist, developmental-behavioral pediatrician, or another health professional who is specially trained to diagnose and treat Asperger's syndrome. The doctor bases his or her diagnosis on the child's level of development, and the doctor's observation of the child's speech and behavior, including his or her play and ability to socialize with others. The doctor often seeks input from the child's parents, teachers, and other adults who are familiar with the child's symptoms.

How is Asperger's syndrome treated?

Right now, there is no cure for Asperger's syndrome, but treatment may improve functioning and reduce undesirable behaviors. Treatment may include a combination of the following:

- **Special education:** Education that is structured to meet the child's unique educational needs.
- **Behavior modification:** This includes strategies for supporting positive behavior and decreasing problem behaviors.
- **Speech, physical, or occupational therapy:** These therapies are designed to increase the child's functional abilities.
- **Social skills therapies:** Run by a psychologist, counselor, speech pathologist, or social worker, these therapies are invaluable ways to build social skills and the ability to read verbal and non-verbal cues that is often lacking in those with Asperger's.
- **Medication :** There are no medications to treat Asperger's syndrome itself, but drugs may be used to treat specific symptoms such as anxiety, depression, hyperactivity, and obsessive-compulsive behavior.

Childhood Disintegrative Disorder

Childhood disintegrative disorder is also known as Heller's syndrome. It's a very rare condition in which children develop normally until at least two years of age, but then demonstrate a severe loss of social, communication and other skills.

Childhood disintegrative disorder is part of a larger category called autism spectrum disorder. However, unlike autism, someone with childhood disintegrative disorder shows severe regression after several years of normal development and a more dramatic loss of skills than a child with autism does. In addition, childhood disintegrative disorder can develop later than autism does.

Treatment for childhood disintegrative disorder involves a combination of medications, behavior therapy and other approaches.

Symptoms

Children with CDD have at least two years of normal development in all areas—language understanding, speech, skill in the use of large and small muscles, and social development. After this period of normal growth, the child begins to lose the skills he or she has acquired. This loss usually takes place between ages three and four, but it can happen any time up to age ten.

The loss of skills may be gradual, but more often occurs rapidly over a period of six to nine months. The transition may begin with unexplained changes in behavior, such as anxiety, unprovoked anger, or agitation. Behavioral changes are followed by loss of communication, social, and motor skills. Children may stop speaking or revert to single words. They often lose bowel or bladder control and withdraw into themselves, rejecting social interaction with adults or other children. They may perform repetitious activities and often have trouble moving from one activity to the next.

In this way CDD resembles autism. In autism, however, previously acquired skills are not usually lost. According to the *Handbook of Autism and Pervasive Developmental Disorders* , virtually all children with CDD lose speech and social skills. About 90% lose self-help skills (the ability to feed, wash, and toilet themselves); and about the same number develop non-specific overactivity. After a time, the regression stops, but the child does not usually regain the skills that were lost.

Children with childhood disintegrative disorder typically show a dramatic loss of previously acquired skills in two or more of the following areas:

Language, including a severe decline in the ability to speak and have a conversation
Social skills, including significant difficulty relating to and interacting with others
Play, including a loss of interest in imaginary play and in a variety of games and activities

Motor skills, including a dramatic decline in the ability to walk, climb, grasp objects and perform other movements
Bowel or bladder control, including frequent accidents in a child who was previously toilet trained

Loss of developmental milestones may occur abruptly over the course of days to weeks or gradually over an extended period of time.

Children typically develop at their own pace, but any loss of developmental milestones is cause for concern. If your child has suddenly lost previously acquired language, social, motor, play, thinking (cognitive) or self-help skills, such as toilet training and feeding, contact your doctor. In addition, if you suspect that your child has gradually shown a loss in any area of development, talk with your doctor.

Pervasive Developmental Disorder-Not Otherwise Specified

PDD-NOS stands for Pervasive Developmental Disorder-Not Otherwise Specified. Psychologists and psychiatrists sometimes use the term "pervasive developmental disorders" and "autism spectrum disorders" (ASD) interchangeably. As such, PDD-NOS became the diagnosis applied to children or adults who are on the autism spectrum but do not fully meet the criteria for another ASD such as autistic disorder (sometimes called "classic" autism) or Asperger Syndrome.

Like all forms of autism, PDD-NOS can occur in conjunction with a wide spectrum of intellectual ability. Its defining features are significant challenges in social and language development.

Some developmental health professionals refer to PDD-NOS as "subthreshold autism." In other words, it's the diagnosis they use for someone who has some but not all characteristics of autism or who has relatively mild symptoms. For instance, a person may have significant autism symptoms in one core area such as social deficits, but mild or no symptoms in another core area such as restricted, repetitive behaviors.

As a diagnosis, PDD-NOS remains relatively new, dating back only 15 years or so. As a result, some physicians and educators may not be familiar with the term or may use it incorrectly.

The current Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) spells out the criteria for a diagnosis of PDD-NOS. Unfortunately, this description consists of a single paragraph, which mainly asserts what it is not:

"This category should be used when there is severe and pervasive impairment in the development of reciprocal social interaction associated with impairment in either verbal or nonverbal communication skills or with the presence of stereotyped behavior, interests, and activities, but the criteria are not met for a specific Pervasive

Developmental Disorder, Schizophrenia, Schizotypal Personality Disorder, or Avoidant Personality Disorder. For example, this category includes "atypical autism" – presentations that do not meet the criteria for Autistic Disorder because of late age at onset, atypical symptomatology, or subthreshold symptomatology, or all of these."

More helpful, perhaps, are studies suggesting that persons with PDD-NOS can be placed in one of three very different subgroups:

- A high-functioning group (around 25 percent) whose symptoms largely overlap with that of Asperger syndrome, but who differ in terms of having a lag in language development and mild cognitive impairment. (Asperger syndrome does not generally involve speech delay or cognitive impairment).

- A second group (around 25 percent) whose symptoms more closely resemble those of autistic disorder, but do not fully meet all its diagnostic signs and symptoms.

- A third group (around 50 percent) who meet all the diagnostic criteria for autistic disorder, but whose stereotypical and repetitive behaviors are noticeably mild.

As these findings suggest, individuals with PDD-NOS vary widely in their strengths and challenges.

Symptoms

It is common for individuals with PDD-NOS to have more intact social skills and a lower level of intellectual deficit than individuals with other PDDs. Characteristics of many individuals with PDD-NOS are:

- Communication difficulties (e.g., using and understanding language)
- Difficulty with social behavior
- Difficulty with changes in routines or environments
- Uneven skill development (strengths in some areas and delays in others)
- Unusual play with toys and other objects
- Repetitive body movements or behavior patterns

Treatment

Treatment for autism involves special behavioral training. Behavioral training rewards appropriate behavior (positive reinforcement) to teach children social skills and to teach them how to communicate and how to help themselves as they grow older.

With early treatment, most children with autism learn to relate better to others. They learn to communicate and to help themselves as they grow older. With early and intensive treatment, most children improve their ability to relate to others, communicate, and help

themselves as they grow older. Contrary to popular myths about children with autism, very few are completely socially isolated or "live in a world of their own."

Depending on the child, treatment may also include such things as speech therapy or physical therapy. Medicine is sometimes used to treat problems such as depression or obsessive-compulsive behaviors.

Exactly what type of treatment your child needs depends on the symptoms, which are different for each child and may change over time. Because people with autism are so different, something that helps one person may not help another. So be sure to work with everyone involved in your child's education and care to find the best way to manage symptoms.

Early diagnosis and treatment helps young children with autism develop to their full potential. The primary goal of treatment is to improve the overall ability of the child to function.

Symptoms and behaviors of autism can combine in many ways and vary in severity. Also, individual symptoms and behaviors often change over time. For these reasons, treatment strategies are tailored to individual needs and available family resources. But in general children with autism respond best to highly structured and specialized treatment. A program that addresses helping parents and improving communication, social, behavioral, adaptive, and learning aspects of a child's life will be most successful.

The American Academy of Pediatrics (AAP) recommends the following strategies for helping a child to improve overall function and reach his or her potential:

- **Behavioral training and management**. Behavioral training and management uses positive reinforcement, self-help, and social skills training to improve behavior and communication. Many types of treatments have been developed, including Applied Behavioral Analysis (ABA), Treatment and Education of Autistic and Related Communication Handicapped Children (TEACCH), and sensory integration.
- **Specialized therapies.** These include speech, occupational, and physical therapy. These therapies are important components of managing autism and should all be included in various aspects of the child's treatment program. Speech therapy can help a child with autism improve language and social skills to communicate more effectively. Occupational and physical therapy can help improve any deficiencies in coordination and motor skills. Occupational therapy may also help a child with autism to learn to process information from the senses (sight, sound, hearing, touch, and smell) in more manageable ways.
- **Medicines**. Medicines are most commonly used to treat related conditions and problem behaviors, including depression, anxiety, hyperactivity, and obsessive-compulsive behaviors.
- **Community support and parent training**. Talk to your doctor or contact an advocacy group for support and training.

Many people with autism have sleep problems. These are usually treated by staying on a routine, including a set bedtime and time to get up. Your doctor may try medicines as a last resort.

Treatment Options for Toddlers and Preschool Children

Scientific studies have demonstrated that early intensive behavioral intervention improves learning, communication and social skills in young children with autism. While the outcomes of early intervention vary, all children benefit. Researchers have developed a number of effective early intervention models. They vary in details, but all good early intervention programs share certain features. They include:

- The child receives structured, therapeutic activities for at least 25 hours per week.
- Highly trained therapists and/or teachers deliver the intervention. Well-trained paraprofessionals may assist with the intervention under the supervision of an experienced professional with expertise in autism therapy.
- The therapy is guided by specific and well-defined learning objectives, and the child's progress in meeting these objectives is regularly evaluated and recorded.
- The intervention focuses on the core areas affected by autism. These include social skills, language and communication, imitation, play skills, daily living and motor skills.
- The program provides the child with opportunities to interact with typically developing peers.
- The program actively engages parents in the intervention, both in decision making and the delivery of treatment.
- The therapists make clear their respect for the unique needs, values and perspectives of the child and his or her family.
- The program involves a multidisciplinary team that includes, as needed, a physician, speech-language pathologist and occupational therapist.

An important part of your child's treatment plan is making sure that other family members get training about autism and how to manage symptoms. Training can reduce family stress and help your child function better. Some families need more help than others.

Specific Types of Treatment

Applied Behavioral Analysis

A notable treatment approach for people with an ASD is called applied behavior analysis (ABA). ABA has become widely accepted among health care professionals and used in many schools and treatment clinics. ABA encourages positive behaviors and discourages negative behaviors in order to improve a variety of skills. The child's progress is tracked and measured.

Behavior analysis is a scientifically validated approach to understanding behavior and how it is affected by the environment. In this context, “behavior" refers to actions and skills. "Environment" includes any influence – physical or social – that might change or be changed by one's behavior.

On a practical level, the principles and methods of behavior analysis have helped many different kinds of learners acquire many different skills – from healthier lifestyles to the mastery of a new language. Since the 1960s, therapists have been applying behavior analysis to help children with autism and related developmental disorders.

There are different types of ABA. Following are some examples:

- Discrete Trial Training (DTT)
  DTT is a style of teaching that uses a series of trials to teach each step of a desired behavior or response. Lessons are broken down into their simplest parts and positive reinforcement is used to reward correct answers and behaviors. Incorrect answers are ignored.
- Early Intensive Behavioral Intervention (EIBI)
  This is a type of ABA for very young children with an ASD, usually younger than five, and often younger than three.
- Pivotal Response Training (PRT)
  PRT aims to increase a child’s motivation to learn, monitor his own behavior, and initiate communication with others. Positive changes in these behaviors should have widespread effects on other behaviors.
- Verbal Behavior Intervention (VBI)
  VBI is a type of ABA that focuses on teaching verbal skills.

What is ABA?

Behavior analysis focuses on the principles that explain how learning takes place. Positive reinforcement is one such principle. When a behavior is followed by some sort of reward, the behavior is more likely to be repeated. Through decades of research, the field of behavior analysis has developed many techniques for increasing useful behaviors and reducing those that may cause harm or interfere with learning.

Applied behavior analysis (ABA) is the use of these techniques and principles to bring about meaningful and positive change in behavior.

As mentioned, behavior analysts began working with young children with autism and related disorders in the 1960s. Early techniques often involved adults directing most of the instruction. Some allowed the child to take the lead. Since that time, a wide variety of ABA techniques have been developed for building useful skills in learners with autism – from toddlers through adulthood.

These techniques can be used in structured situations such as a classroom lesson as well as in "everyday" situations such as family dinnertime or the neighborhood playground.

Some ABA therapy sessions involve one-on-one interaction between the behavior analyst and the participant. Group instruction can likewise prove useful.

How Does ABA Benefit Those with Autism?

Today, ABA is widely recognized as a safe and effective treatment for autism. It has been endorsed by a number of state and federal agencies, including the U.S. Surgeon General and the New York State Department of Health. Over the last decade, the nation has seen a particularly dramatic increase in the use of ABA to help persons with autism live happy and productive lives. In particular, ABA principles and techniques can foster basic skills such as looking, listening and imitating, as well as complex skills such as reading, conversing and understanding another person's perspective.

What Does Research Tell Us About ABA and Autism?

Autism Speaks has funded and continues to fund research on developing and validating ABA techniques.

A number of completed studies have demonstrated that ABA techniques can produce improvements in communication, social relationships, play, self care, school and employment. These studies involved age groups ranging from preschoolers to adults. Results for all age groups showed that ABA increased participation in family and community activities.

A number of peer-reviewed studies have examined the potential benefits of combining multiple ABA techniques into comprehensive, individualized and intensive early intervention programs for children with autism. "Comprehensive" refers to interventions that address a full range of life skills, from communication and sociability to self-care and readiness for school. "Early intervention" refers to programs designed to begin before age 4. "Intensive" refers to programs that total 25 to 40 hours per week for 1 to 3 years.

These programs allow children to learn and practice skills in both structured and unstructured situations. The "intensity" of these programs may be particularly important to replicate the thousands of interactions that typical toddlers experience each day while interacting with their parents and peers.

Such studies have demonstrated that many children with autism experience significant improvements in learning, reasoning, communication and adaptability when they participate in high-quality ABA programs. Some preschoolers who participate in early intensive ABA for two or more years acquire sufficient skills to participate in regular classrooms with little or no additional support. Other children learn many important skills, but still need additional educational support to succeed in a classroom.

Across studies, a small percentage of children show relatively little improvement. More research is needed to determine why some children with autism respond more favorably to early intensive ABA than others do. Currently, it remains difficult to predict the extent to which a particular child will benefit.

In some studies, researchers compared intensive ABA with less intensive ABA and/or other early intervention or special education programs for children with autism. Generally, they found that children who receive intensive ABA treatment make larger

improvements in more skill areas than do children who participate in other interventions. In addition, the parents of the children who receive intensive ABA report greater reductions in daily stress than do parents whose children receive other treatments.

ABA and Adults with Autism

A number of recent studies confirm that ABA techniques are effective for building important life skills in teens and adults with autism. Many comprehensive autism support programs for adults employ and combine ABA techniques to help individuals transition successfully into independent living and employment. However, the benefits of intensive ABA programs remain far less studied in teens and adults than they have been with young children. This is a research area of particular interest to Autism Speaks and its supporters.

What Does ABA Intervention Involve?

Effective ABA intervention for autism is not a "one size fits all" approach and should never be viewed as a "canned" set of programs or drills. On the contrary, a skilled therapist customizes the intervention to each learner's skills, needs, interests, preferences and family situation. For these reasons, an ABA program for one learner will look different than a program for another learner. That said, quality ABA programs for learners with autism have the following in common:

Planning and Ongoing Assessment

* A qualified and trained behavior analyst designs and directly oversees the intervention.
* The analyst's development of treatment goals stems from a detailed assessment of each learner's skills and preferences and may also include family goals.
* Treatment goals and instruction are developmentally appropriate and target a broad range of skill areas such as communication, sociability, self-care, play and leisure, motor development and academic skills.
* Goals emphasize skills that will enable learners to become independent and successful in both the short and long terms.
* The instruction plan breaks down desired skills into manageable steps to be taught from the simplest (e.g. imitating single sounds) to the more complex (e.g. carrying on a conversation).
* The intervention involves ongoing objective measurement of the learner's progress.
* The behavior analyst frequently reviews information on the learner's progress and uses this to adjust procedures and goals as needed.
* The analyst meets regularly with family members and program staff to plan ahead, review progress and make adjustments as needed.

ABA Techniques and Philosophy

* The instructor uses a variety of behavior analytic procedures, some of which are directed by the instructor and others initiated by the learner.
* Parents and/or other family members and caregivers receive training so they can support learning and skill practice throughout the day.
* The learner's day is structured to provide many opportunities – both planned and

naturally occurring - to acquire and practice skills in both structured and unstructured situations.
* The learner receives an abundance of positive reinforcement for demonstrating useful skills and socially appropriate behaviors. The emphasis is on positive social interactions and enjoyable learning.
* The learner receives no reinforcement for behaviors that pose harm or prevent learning.

What Kind of Progress Can Be Expected with ABA?

Competently delivered ABA intervention can help learners with autism make meaningful changes in many areas. However, changes do not typically occur quickly. Rather, most learners require intensive and ongoing instruction that builds on their step-by-step progress. Moreover, the rate of progress – like the goals of intervention – varies considerably from person to person depending on age, level of functioning, family goals and other factors.

Some learners do acquire skills quickly. But typically, this rapid progress happens in just one or two particular skill areas such as reading, while much more instruction and practice is needed to master another skill area such as interacting with peers.

Who Is Qualified to Provide ABA Intervention?

Just as a medical treatment program should be directed by a qualified medical professional, ABA programs for learners with autism should be designed and supervised by qualified professionals, which include either licensed clinical psychologists with training in applied behavior analysis or behavior analysts, who are board certified with supervised experience providing ABA treatment for autism or who can clearly document that they have equivalent training and experience.

Because of the huge demand for ABA intervention for autism, many individuals and programs now claim to provide ABA. Some are private practitioners or agencies that offer services in a family's home. Others operate private schools. And still others provide consultation services to public schools.
Unfortunately, some who claim to offer ABA lack the field's established minimum requirements in education and practical experience. Family members, teachers and others involved in developing an individual's therapy and support program should keep the following in mind when choosing an ABA program or practitioner:

Always check credentials of those who claim to be qualified in behavior analysis. For example, for licensed clinical psychologists, you should inquire about the level of training in behavioral interventions for autism, including training in applied behavior analysis. For behavior analysts, you should determine whether the person has been credentialed with the Behavior Analyst Certification Board or the Association of Professional Behavior Analysts. These professionals often supervise other people, including paraprofessionals, who will be working directly with your child. Thus, it is important that you feel confident that the licensed clinical psychologist or behavior analyst is providing regular supervision to anyone working directly with your child.

Parents, guardians and other caregivers should monitor the program by observing sessions and participating in training sessions and consultations.

Early Start Denver Model

What Is the Early Start Denver Model?

The Early Start Denver Model (ESDM) is a comprehensive behavioral early intervention approach for children with autism, ages 12 to 48 months. The program encompasses a developmental curriculum that defines the skills to be taught at any given time and a set of teaching procedures used to deliver this content. It is not tied to a specific delivery setting, but can be delivered by therapy teams and/or parents in group programs or individual therapy sessions in either a clinic setting or the child's home.

Psychologists Sally Rogers, Ph.D., and Geraldine Dawson, Ph.D., developed the Early Start Denver Model as an early-age extension of the Denver Model, which Rogers and colleagues developed and refined. This early intervention program integrates a relationship-focused developmental model with the well-validated teaching practices of Applied Behavior Analysis (ABA). Its core features include the following:

- Naturalistic applied behavioral analytic strategies
- Sensitive to normal developmental sequence
- Deep parental involvement
- Focus on interpersonal exchange and positive affect
- Shared engagement with joint activities
- Language and communication taught inside a positive, affect-based relationship

Who can benefit from the Early Start Denver Model? What Has Research Shown?

The Early Start Denver Model is the only comprehensive early intervention model that has been validated in a randomized clinical trial for use with children with autism as young as 18 months of age. It has been found to be effective for children with autism spectrum disorder (ASD) across a wide range of learning styles and abilities. Children with more significant learning challenges were found to benefit from the program as much as children without such learning challenges. A randomized clinical trial published in the journal *Pediatrics* showed that children who received ESDM therapy for 20 hours a week (15 hours by trained therapists, 5 hours by parents) over a 2-year span showed greater improvement in cognitive and language abilities and adaptive behavior and fewer autism symptoms than did children referred for interventions commonly available in their communities.

Who is qualified to provide ESDM?

An ESDM therapist may be a psychologist, behaviorist, occupational therapist, speech and language pathologist, early intervention specialist or developmental pediatrician. What's important is that they have ESDM training and certification.

How can professionals become trained in ESDM?

Qualified professionals attend a training workshop and then submit videotapes showing them using ESDM techniques in therapy sessions. Certification requires that the therapist demonstrates the ability to implement ESDM techniques reliably and according to high standards set by leading ESDM therapists.

This ensures that a certified professional has the knowledge and skills to successfully use the teaching strategies with children with autism.

How can parents be trained in the techniques?

Parental involvement is a crucial part of the ESDM program. If your child is receiving ESDM therapy, the instructor will explain and model the strategies for you to use at home.

In addition, Drs. Dawson and Rogers saw the need for a separate training "manual" for parents. Earlier this year, they published *An Early Start for Your Child with Autism*, with coauthor Laurie Vismara, PhD. The book has useful tips and hands-on strategies that integrate smoothly into daily activities and play. I think you'll find it useful whether or not your child's therapist is trained in ESDM techniques. You can even use it while waiting for your child to begin therapy.

Pivotal Response Treatment

Pivotal Response Treatment (PRT) is one of the best studied and validated behavioral treatments for autism. Derived from applied behavioral analysis (ABA), it is play based and child initiated. Its goals include the development of communication, language and positive social behaviors and relief from disruptive self-stimulatory behaviors.

Rather than target individual behaviors, the PRT therapist targets "pivotal" areas of a child's development. These include motivation, response to multiple cues, self-management and the initiation of social interactions. The philosophy is that, by targeting these critical areas, PRT will produce broad improvements across other areas of sociability, communication, behavior and academic skill building.

Motivation strategies are an important part of the PRT approach. These emphasize "natural" reinforcement. For example, if a child makes a meaningful attempt to request, say, a stuffed animal, the reward is the stuffed animal – not a candy or other unrelated reward.

Though used primarily with preschool and elementary school learners, studies show that PRT can also help adolescents and young adults. Indeed, autism-affected persons of all ages may benefit from its techniques. In all age groups, the learner plays a crucial role in determining the activities and objects that will be used in a PRT exchange.

What is the History of PRT?

Pivotal response treatment was developed in the 1970s by educational psychologists Robert Koegel, Ph.D., and Lynn Kern Koegel, Ph.D., at the University of California, Santa Barbara. The Koegels are now director and clinical director, respectively, of the UCSB Koegel Autism Research Center.

Since its inception, Pivotal Response Treatment has been called Pivotal Response Training, Pivotal Response Teaching, Pivotal Response Therapy, Pivotal Response Intervention and the Natural Language Paradigm. These terms all refer to the same treatment delivery system.

Who provides PRT?

Many psychologists, special education teachers, speech therapists and other providers pursue training in PRT. The Koegel Autism Research Centers offers training and certification.

What is a typical PRT therapy session like?

Each program is tailored to meet the goals and needs of the individual learner and his or her school and home routines. A session typically involves six segments during which language, play and social skills are targeted with both structured and unstructured interactions. As the learner progresses, the focus of each session changes to accommodate more advanced goals and needs.

What is the time commitment involved?

PRT programs usually involve 25 or more hours per week for the learner as well as instruction for parents and other caregivers. Indeed, everyone involved in the learner's life is encouraged to use PRT methods consistently. PRT has been described as a "lifestyle" adopted by the affected family.

Verbal Behavior Therapy

Verbal Behavior Therapy teaches communication using the principles of Applied Behavior Analysis and the theories of behaviorist B.F. Skinner. By design, Verbal Behavior Therapy motivates a child, adolescent or adult to learn language by connecting words with their purposes. The student learns that words can help obtain desired objects or other results.

Therapy avoids focusing on words as mere labels (cat, car, etc.) Rather, the student learns how to use language to make requests and communicate ideas. To put it another way, this intervention focuses on understanding *why* we use words.

In his book *Verbal Behavior*, Skinner classified language into types, or "operants." Each has a different function. Verbal Behavior Therapy focuses on four word types. They are:

- **Mand.** A request. Example: "Cookie," to ask for a cookie.
- **Tact.** A comment used to share an experience or draw attention. Example: "airplane" to point out an airplane.

- **Intraverbal.** A word used to answer a question or otherwise respond. Example: Where do you go to school? “Castle Park Elementary.”
- **Echoic.** A repeated, or echoed, word. Example: "Cookie?" “Cookie!” (important as the student needs to imitate to learn)

Verbal Behavior Therapy begins by teaching mands, or requests, as the most basic type of language. For example, the individual with autism learns that saying "cookie" can produce a cookie. Immediately after the student makes such a request, the therapist reinforces the lesson by repeating the word and presenting the requested item. The therapist then uses the word again in the same or similar context.

Importantly, students don’t have to say the actual word to receive the desired item. In the beginning, they simply need to signal requests by any means. Pointing at the item represents a good start.

This helps the student understand that *communicating* produces positive results. The therapist builds on this understanding to help the student shape the communication toward saying or signing the actual word.

Importantly, Verbal Behavior Therapy uses “errorless learning.” The therapist provides immediate and frequent prompts to help improve the student’s communication. These prompts become less intrusive as quickly as possible, until the student no longer needs prompting. Take, for example, the student who wants a cookie. The therapist may hold the cookie in front of the student’s face and say “cookie,” to prompt a response from the child. Next, the therapist would hold up the cookie and make a “c” sound, to prompt the response. After that, the therapist might simply hold a cookie in the child’s line of sight and wait for the request. The ultimate goal, in this example, is for the student to say “cookie” when he or she wants a cookie – without any prompting.

In a typical Verbal Behavior Therapy session, the teacher asks a series of questions that combine easy and hard requests. This increases the frequency of success and reduces frustration. Ideally, the teacher varies the situations and instructions in ways that catch and sustain the student’s interest.

Most programs involve a minimum of one to three hours of therapy per week. More-intensive programs can involve many more hours. In addition, instructors train parents and other caregivers to use verbal-behavior principles throughout the student’s daily life.

Who Responds to Verbal Behavior Therapy?

Reports suggest that Verbal Behavior Therapy can help both young children beginning to learn language and older students with delayed or disordered language. It likewise helps many children and adults who sign or use visual supports or other forms of assisted communication.

Floortime Therapy

Floortime therapy derives from the Developmental Individual-difference Relationship-based model (DIR) created by child psychiatrists Stanley Greenspan, M.D. and Serena Wieder, PhD. Its premise is that adults can help children expand their circles of communication by meeting them at their developmental level and building on their strengths.

According to the organization Greenspan Floortime Approach, the technique challenges children with autism to push themselves to their full potential. It develops "who they are," rather than "what their diagnosis says."

As its name suggests, Floortime encourages parents to engage children literally at their level – by getting on the floor to play. Families can combine it with other behavioral therapies or use it as an alternative approach.

In Floortime, therapists and parents engage children through the activities each child enjoys. They enter the child's games. They follow the child's lead. Therapists teach parents how to direct their children into increasingly complex interactions. This process, called "opening and closing circles of communication," remains central to the Floortime approach.

Overall, Floortime aims to help children reach six developmental milestones crucial for emotional and intellectual growth. They are:

- Self-regulation and interest in the world
- Intimacy, or engagement in human relations
- Two-way communication
- Complex communication
- Emotional ideas
- Emotional thinking

Floortime does not target speech, motor or cognitive skills in isolation. Rather, it addresses these areas through its focus on emotional development.

How does Floortime work?

Ideally, Floortime takes place in a calm environment. This can be at home or in a professional setting. Formal treatment sessions range from two to five hours a day. They include training for parents and caregivers as well as interaction with the child. Therapists encourage families to use Floortime principals in their daily lives.

Floortime sessions emphasize back-and-forth play interactions. This establishes the foundation for shared attention, engagement and problem solving. Parents and therapists help the child maintain focus to sharpen interactions and abstract, logical thinking.

For example, if the child is tapping a toy truck, the parent might tap a toy car in the same way. To encourage interaction, the parent might then put the car in front of the child's truck or add language to the game.

As children mature, therapists and parents tailor the strategies to match a child's developing interests and higher levels of interaction. For example, instead of playing with toy trucks, parents can engage with model airplanes or even ideas and academic fields of special interest to their child.

Relationship Development Intervention

Relationship Development Intervention® (RDI) is a family-based, behavioral treatment designed to address autism's core symptoms. Developed by psychologist Steven Gutstein, Ph.D., it builds on the theory that "dynamic intelligence" is key to improving quality of life for individuals with autism. Dr. Gutstein defines dynamic intelligence as the ability to think flexibly. This includes appreciating different perspectives, coping with change and integrating information from multiple sources (e.g. sights and sounds).

RDI aims to help individuals with autism form personal relationships by gradually strengthening the building blocks of social connections. This includes the ability to form an emotional bond and share experiences.

RDI's six objectives are:

- **Emotional referencing**: The ability to learn from the emotional and subjective experiences of others

- **Social coordination**: The ability to observe and control behavior to successfully participate in social relationships

- **Declarative language**: The ability to use language and non-verbal communication to express curiosity, invite interactions, share perceptions and feelings and coordinate with others
- **Flexible thinking**: The ability to adapt and alter plans as circumstances change
- **Relational information processing**: The ability to put things into context and solve problems that lack clear cut solutions
- **Foresight and hindsight**: The ability to anticipate future possibilities based on past experiences

Though designed for in-home use, RDI is also used by classroom teachers and behavioral therapists. Training typically begins with the parent or other caregiver attending educational sessions led by an RDI consultant. Certified RDI consultants operate in the

U.S., Canada and many other countries. Alternately, caregivers can learn the principles of RDI through a variety of books.

The consultant may also assess the child and his or her interactions with parents or teachers. Based on this information, the consultant designs a personalized teaching plan. It includes developing communications styles that best suit the child.

The initial goal is to build a "guided participation" relationship between parents and child, with the child as a "cognitive apprentice." Once this relationship is in place, the family advances through a series of developmental goals for their child. According to Dr. Gutstein, this process improves “neural connectivity,” or brain function.

Parents, teachers and other caretakers continue to apply the principles of RDI in the child’s daily life. They use positive reinforcement to help the child improve social skills, adaptability and self-awareness.

In its initial stages, RDI involves one-on-one work between caregiver and child. In the next stage of the intervention, the child begins spending time with a peer at a similar level of relationship development. Gradually, additional children join the group. With guidance, they meet and play in a variety of settings. This allows them to practice forming and maintaining relationships in different contexts.

What is a typical RDI therapy session like?

Parents or other caregivers apply a set of stepwise, developmentally appropriate objectives to everyday life situations. For instance, in the early stages of training, they may limit spoken language to encourage eye contact and non-verbal communication. They gradually advance these goals as the child’s abilities increase.

TEAACH Autism Program

The TEACCH® Autism Program is a clinical, training, and research program based at the University of North Carolina – Chapel Hill. TEACCH, developed by Drs. Eric Schopler and Robert Reichler in the 1960s, was established as a statewide program by the North Carolina legislature in 1972, and has become a model for other programs around the world.

TEACCH offers a set of core clinical services at seven community-based centers throughout North Carolina, a supported employment program and an integrated vocational and residential program for adults with ASD. TEACCH’s **clinical services** include diagnostic evaluations; parent training and parent support groups, intervention groups, and individual counseling for higher-functioning clients. TEACCH services are grounded in empirical research, enriched by extensive clinical expertise, and notable for flexible and individualized support of individuals with ASD and their families.

TEACCH developed the intervention approach called “**Structured TEACCHing**”, an array of teaching or treatment principles and strategies based on the learning characteristics of individuals with ASD, including strengths in visual information

processing, and difficulties with social communication, attention, and executive function. In response to this profile of strengths and challenges, Structured TEACCHing includes:

1. External organizational supports to address challenges with attention and executive function
2. Visual and/or written information to supplement verbal communication
3. Structured support for social communication

Structured TEACCHing is not a curriculum, but instead is a framework to support achievement of educational and therapeutic goals. This framework includes:

1. Physical organization
2. Individualized schedules
3. Work (Activity) systems
4. Visual structure of materials in tasks and activities

The goal of Structured TEACCHing is to promote meaningful engagement in activities, flexibility, independence, and self-efficacy. We integrate Structured TEACCHing strategies into other evidenced-based practices.

Training

TEACCH is committed to developing training programs for students and professionals who serve individuals with ASD. In addition to training University-based graduate students, TEACCH conducts training nationally and internationally and provides consultation for teachers, residential care providers, and other professionals from a variety of disciplines. TEACCH uses a unique training model that combines traditional lecture-style activities with hands-on learning activities to train professionals to employ Structured TEACCHing techniques

TEACCH Autism Program has established a comprehensive **Professional Certification Program** in response to the increasing number of professionals trained by TEACCH and the growing demand for quality services for individuals with Autism Spectrum Disorder. This certification program is for educators, psychologists, social workers, speech therapists and other service providers in the field of autism.

This new TEACCH Professional Certification Program will help protect the integrity and quality of the TEACCH model and provide educators and clinicians with a professional certification that documents their knowledge and use of Structured TEACCHing Intervention strategies, evidence-based practices. The program includes two certification levels, **Practitioner** and **Advanced Consultant**, progressing professional from a skilled practitioner to a consultation role within their work setting to an advanced consultant who is trained to provide lectures to groups inside and outside their work setting.

Social Communication/Emotional Regulation/Transactional Support SCERTS

SCERTS is an educational model developed by Barry Prizant, PhD, Amy Wetherby, PhD, Emily Rubin and Amy Laurant. SCERTS uses practices from other approaches including ABA (in the form of PRT), TEACCH, Floortime and RDI. The SCERTS Model differs most notably from the focus of ?traditional? ABA, by promoting child-initiated communication in everyday activities. SCERTS is most concerned with helping children with autism to achieve ?Authentic Progress,? which is defined as the ability to learn and spontaneously apply functional and relevant skills in a variety of settings and with a variety of partners.

The acronym SCERTS refers to the focus on:

SC: Social Communication - Development of spontaneous, functional communication, emotional expression and secure and trusting relationships with children and adults.

ER: Emotional Regulation - Development of the ability to maintain a well-regulated emotional state to cope with everyday stress, and to be most available for learning and interacting.

TS: Transactional Support - Development and implementation of supports to help partners respond to the child's needs and interests, modify and adapt the environment, and provide tools to enhance learning (e.g., picture communication, written schedules, and sensory supports).

Specific plans are also developed to provide educational and emotional support to families, and to foster teamwork among professionals.

What does a SCERTS session look like?

The SCERTS Model favors having children learn with and from children who provide good social and language models in inclusive settings as much as possible. SCERTS is implemented using transactional supports implemented by a team, such as environmental accommodations, learning supports (schedules or visual organizers).

Who provides SCERTS?

SCERTS is usually provided in a school setting by SCERTS-trained special education teachers, speech therapist.

Families and Autism

Having a child with autism requires taking a proactive approach to learning about the condition and its treatment while working closely with others involved in your child's care. You also need to take care of yourself so that you are able to face the many challenges of having a child with autism.

Take advantage of every kind of help you can find. Talk to your doctor about what help is available where you live. Family, friends, public agencies, and autism organizations are all possible resources.

Remember these tips:

- Plan breaks. The daily demands of caring for a child with autism can take their toll. Planned breaks will help the whole family.
- Get extra help when your child gets older. The teen years can be a very hard time for children with autism.
- Get in touch with other families who have children with autism. You can talk about your problems and share advice with people who will understand.

Raising a child with autism is hard work. But with support and training, your family can learn how to cope.

Educate Yourself

Ask your doctor or contact autism groups to find training about autism and how to manage symptoms. Parent and family education can reduce family stress and improve a child's functioning. Understanding the condition and knowing what to expect is an important part of helping your child develop independence.

Become informed about your child's educational rights. Federal laws require services for handicapped children, including those with autism. Also, there may be state and local laws or policies to aid children who have autism. Find out what services are available in your area.

Learning about autism will also help prepare you for when your child reaches adulthood. Some adults with autism can live by themselves, work, and be as independent as other people their age. Others need continued support.

Work closely with others who care for your child

Close communication with others involved in your child's education and care will help all concerned. The best treatment for children with autism is a team approach and a consistent, structured program. Everyone involved needs to work together to set goals for:

- Education.
- Identifying and managing symptoms of autism and any related conditions.

- Behavior and interactions with family and peers, adjustment to different environments, and social and communication skills.

Work closely with the health professionals involved in your child's care. It is important that they take time to listen to your concerns and are willing to work with you.

Promote healthy growth and development

Children as young as preschool age benefit from exercise and fitness as much as adults do. The same is true for children with autism spectrum disorders (ASDs), such as autism. Not only does physical activity promote a healthy weight and body, but it also provides opportunities to build self-esteem, confidence, and friendships with other children. For children with ASDs, these social benefits may be especially important. Work with your child's doctors to learn how physical activities may be best worked into your child's routine.

Children with ASDs may be especially interested in video games, computers, or other screen-based media such as TV. If possible, keep televisions, video games, and computers out of your child's bedroom. When children with ASDs have these devices in their bedroom, they are more likely to sleep fewer hours. This is especially true when video games are in the bedroom. If your child doesn't get enough sleep, his or her ASD symptoms may be worse.

Children with autism often have picky eating habits or may take a long time to acquire tastes for new foods. This can be frustrating for parents. One reason for picky eating may not be because of how the food tastes, but because of how it feels or its texture. Children with autism are very sensitive to textures. You may try preparing the food in a different way, such as blending a banana in a smoothie instead of having your child eat the banana.

Take care of yourself

Learn ways to handle the normal range of emotions, fears, and concerns that go along with raising a child who has autism. The daily and long-term challenges put you and your other children at an increased risk for depression or stress-related illnesses. The way you handle these issues influences other family members.

- Get involved in a hobby, visit with friends, and learn ways to relax.
- Seek and accept support from others. Consider using respite care, which is a family support service that provides a break for parents and siblings. Also, support groups for parents and siblings are often available. People who participate in support groups can benefit from others' experiences. For more information on support groups in your area, contact the Autism Society of America at www.autism-society.org.
- Talk with a doctor about whether counseling would help if you or one of your children is having trouble handling the strains related to having a family member with autism.

TR Implications

TR professionals can help improve the lives of people with Autism in many areas. Some of these areas are social and interpersonal skills. People with ASD relate poorly to people and have difficulty understanding how other people feel. They also have fears other people don't have. They typically avoid eye contact, want to be alone, demonstrate odd or inappropriate social behavior, and lack of empathy or understanding of how other people feel. Leisure activities and therapeutic recreation can play a significant role in helping improve those areas and build their skills.

Resources

National Institute of Neurological Disorders and Stroke
www.ninds.nih.gov

Autism Speaks
https://www.autismspeaks.org/

National Dissemination Center for Children with Disabilities
Htttp://www.nichcy.org

MAAP Services for Autism, Asperger Syndrome, and PDD
http://www.maapservices.org

Autism Research Institute
http://www.autismresearchinstitute.com

Autism Network International
http://www.ani.ac

Autism National Committee
http://autcom.org

National Institue of Menal Health
http://www.nimh.nih.gov

National Institute of Child Health and Human Information Resource Center
http://www.nichd.nih.gov

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