

Why was my child diagnosed with Autism? And what does it mean?

Your child has been diagnosed with autism and you have asked for help. This is an important turning point in a long journey. For some families, it may be the point where, after a long search for answers, you now have a name for something you didn't know what to call, but you knew existed. Perhaps you suspected autism, but held out hope that an evaluation would prove otherwise.

Many families report mixed feelings of sadness and relief when their child is diagnosed. You may feel completely overwhelmed. You may also feel relieved to know that the concerns you have had for your child are valid. Whatever it is you feel, know that thousands of parents share this journey. You are not alone. There is reason to hope. There is help.

Now that you have the diagnosis, the question is, where do you go from here?

This handbook, the *Autism Speaks™* 100 Day Kit, was created to help you make the best possible use of the next 100 days in the life of your child. It contains information and advice collected from trusted and respected experts on autism and parents like you.

In this kit, the umbrella term “Autism” refers to the Pervasive Developmental Disorders, also known as Autism Spectrum Disorders, including Autism, PDD, PDD-NOS, and Asperger’s Syndrome.

Why does my child need a diagnosis of Autism?

Parents are usually the first to notice the early signs of autism. You probably noticed that your child was developing differently from his or her peers. The differences may have existed from birth, or may have become more noticeable later. Sometimes the differences are severe and are obvious to everyone. In other cases they are more subtle and are first recognized by a day-care provider or preschool teacher. Those differences, the symptoms of autism, have led thousands of parents like you to seek answers that have resulted in a diagnosis of autism.

You may wonder: Why does my child need a diagnosis of autism? That’s a fair question to ask - especially when right now, no one is able to offer you a cure.

Autism Speaks is dedicated to funding global biomedical research to find the causes, prevention, treatment and a cure for autism. Great strides have been made and the state of progress is a far cry from the time when parents were given no hope for their children. Some of the most brilliant minds of our time have turned their attention toward this disorder and we work at a constantly increasing pace toward a cure for autism. While indeed, we live in an age of miracles and wonders, we’re not there yet. In the meantime, the best treatments available to us now – the therapies and interventions you will learn about in this handbook – are our chemotherapy, our dialysis, our insulin.

It is important to remember, your child is the same unique, lovable, wonderful person he or she was before the diagnosis. There are however, several reasons why having a diagnosis is important for

Autism Speaks

“Now we know Nicky has Autism. Everyone told me I was worrying about nothing; that he was a late bloomer and would ‘grow out of it.’ I didn’t want to wait and see if he got better. Now that we know, we can help him.”

your child. A thorough and detailed diagnosis provides important information about your child’s behavior and development. It can provide a road map for treatment, by identifying your child’s specific strengths and challenges, providing useful information about which needs and skills to target for intervention. A diagnosis is often required to access autism specific services through early intervention programs or your local school district.

refer the child to Early Intervention and a specialist for a developmental evaluation. Hearing and lead exposure screenings should be performed and an autism-specific screening tool, such as the **Modified Checklist of Autism in Toddlers (MCHAT)** should be used.

The MCHAT, is a list of simple questions about your child. The answers determine whether he or she should be referred to a specialist, usually a **Developmental Pediatrician**, a **Neurologist**, a **Psychiatrist** or a **Psychologist**, for further evaluation. There are other screening tools available, some geared towards older children or specific Autism Spectrum Disorders.

Your child may have been diagnosed by one of the professionals mentioned above. In some cases, a team of specialists may have evaluated your child and provided recommendations for treatment. The team may have included an **Audiologist**, to rule out hearing loss, a **Speech & Language Therapist**, to determine language skills and needs, and an **Occupational Therapist** to evaluate physical and motor skills. If this was not the case for your child, you will want to make sure further evaluations are conducted so that you can learn as much as possible about your child’s strengths and needs.

How is Autism Diagnosed?

Presently, there is not a medical test for autism; a diagnosis is based on observed behavior and educational and psychological testing.

As the symptoms of autism vary, so do the routes to obtaining a diagnosis. You may have raised questions with your pediatrician yourself. Some children are identified as having developmental delays before obtaining a diagnosis of autism and may already receive some **Early Intervention** or **Special Education** services.

Unfortunately, parents’ concerns are sometimes not taken seriously by their doctor and a diagnosis is delayed. *Autism Speaks* and other autism related organizations are working hard to educate parents and physicians so that children with autism are identified as early as possible. From birth to at least 36 months of age, every child should be screened for developmental milestones during routine well visits. If concerns about a child’s development are raised, their doctor should

More information on terms that are printed in **bold** is available in the glossary at the end of this handbook and in the Video Glossary at www.AutismSpeaks.org

What is Autism?

Autism is a general term used to describe a group of complex developmental brain disorders known as **Pervasive Developmental Disorders (PDD)**. The other pervasive developmental disorders are **PDD-NOS (Pervasive Developmental Disorder – Not Otherwise Specified)**, **Asperger’s Syndrome**, **Rett Syndrome** and **Childhood Disintegrative Disorder**. Many parents and professionals refer to this group as **Autism Spectrum Disorders**.

You may also hear the terms **Classic Autism** or **Kanner’s Autism** (named after the first psychiatrist to describe autism) used to describe the most severe form of the disorder.

The American Psychiatric Association’s **Diagnostic and Statistical Manual of Mental Disorders** is the main diagnostic reference used by mental health professionals and insurance providers in the United States. The current (fourth) edition, which was published in 1994, is commonly referred to as the “**DSM-IV**.” The diagnosis of autism requires at least six developmental and behavioral characteristics are observed, that problems are present before age three, and there is no evidence for certain other conditions that are similar.

DSM-IV criteria for a diagnosis of Autism

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

Source: Diagnostic and Statistical Manual of Mental Disorders; Fourth Edition

How common is Autism?

Today, it is estimated that one in every 150 children is diagnosed with autism, making it more common than childhood cancer, juvenile diabetes and pediatric AIDS combined. An estimated 1.5 million individuals in the U.S. and tens of millions worldwide are affected by autism. Government statistics suggest the prevalence rate of autism is increasing 10-17 percent annually. There is not established explanation for this increase, although improved diagnosis and environmental influences are two reasons often considered. Studies suggest boys are more likely than girls to develop autism and receive the diagnosis three to four times more frequently. Current estimates are that in the United States alone, one out of 94 boys is diagnosed with autism.

What causes Autism?

The simple answer is we don't know. The vast majority of cases of autism are idiopathic, which means the cause is unknown.

The more complex answer is that just as there are different levels of severity and combinations of symptoms in autism, there are probably multiple causes. The best scientific evidence available to us today points toward a potential for various combinations of factors causing autism – multiple genetic components that may cause autism on their own or possibly when combined with exposure to as yet undetermined environmental factors. Timing of exposure during the child's development (before, during or after birth) may also play a role in the development or final presentation of the disorder.

A small number of cases can be linked to genetic disorders such as Fragile X, Tuberous Sclerosis, and Angelman's Syndrome, as well as exposure to environmental agents such as infectious ones (maternal rubella or cytomegalovirus) or chemical ones (thalidomide or valproate) during pregnancy.

There is a growing interest among researchers about the role of the functions and regulation of the immune system, both within the body and the brain, in autism. Piecemeal evidence over the

past 30 years suggests that people with autism may involve inflammation in the central nervous system. There is also emerging evidence from animal studies that illustrates how the immune system can influence behaviors related to autism. *Autism Speaks* is working to extend awareness and investigation of potential immunological issues to researchers outside the field of autism as well as those within the autism research community.

While the definitive cause (or causes) of autism is not yet clear, it is clear that it is not caused by bad parenting. Dr. Leo Kanner, the psychiatrist who first described autism as a unique condition in 1943, believed that it was caused by cold, unloving mothers. Bruno Bettelheim, a renowned professor of child development perpetuated this misinterpretation of autism. Their promotion of the idea that unloving mothers caused their children's autism created a generation of parents who carried the tremendous burden of guilt for their children's disability.

In the 1960s and 70s, Dr. Bernard Rimland, the father of a son with autism, who later founded the Autism Society of America and the Autism Research Institute, helped the medical community understand that autism is not caused by cold parents but rather is a biological disorder.

The best scientific evidence available to us today points toward a potential for various combinations of factors causing autism.



More Information about Symptoms of Autism

Autism affects the way your child perceives the world and makes communication and social interaction difficult. He may also have repetitive behaviors or intense interests. Symptoms, and their severity, are different for each of the affected areas (Communication, Social Interaction, and Repetitive Behaviors). Your child may not have the same symptoms and may seem very different from another child with the same diagnosis. It is sometimes said, that if you know one person with autism; you know *one person* with autism.

The symptoms of autism typically last throughout a person's lifetime. A mildly affected person might seem merely quirky and lead a typical life. A severely affected person might be unable to speak or care for himself. Early intervention can make extraordinary differences in your child's development. How your child is functioning now may be very different from how he or she will function later on in life.

The information following – about the social symptoms, communication disorders and repetitive behaviors associated with autism – is taken from the National Institute of Mental Health Website.

Social Symptoms

From the start, typically developing infants are social beings. Early in life, they gaze at people, turn toward voices, grasp a finger, and even smile.

By contrast, most children with autism seem to have tremendous difficulty learning to engage in the give-and-take of everyday human interactions. Even in the first year of life, many do not interact and avoid eye contact in a normal way. They may seem indifferent to other people, and prefer being alone. They may resist attention or passively accept hugs and cuddling. Later, they may fail to seek comfort or

respond to parents' displays of anger or affection in a typical way. Research has suggested that although children with autism are attached to their parents, their expression of this attachment is unusual and difficult to "read". To parents, it may seem as if their child is not connected at all. Parents who looked forward to the joys of cuddling, teaching and playing with their child may feel crushed by this lack of the expected and typical attachment behavior.

Children with autism also are slower in learning to interpret what others are thinking and feeling. Subtle social cues such as a smile, a wave, or a grimace may have little meaning to a child with autism. To a child who misses these cues, "Come here" may always mean the same thing, whether the speaker is smiling and extending her arms for a hug or frowning and planting her fists on her hips. Without the ability to interpret gestures and facial expressions, the social world may seem bewildering. To compound the problem, people with autism have difficulty seeing things from another person's perspective. Most five year olds understand that other people have different thoughts, feelings, and goals than they have. A child with autism may lack such understanding. This inability leaves them unable to predict or understand other people's actions.

Although not universal, it is common for people with autism to have difficulty regulating their emotions. This can take the form of "immature" behavior such as crying in class or verbal outbursts that seem inappropriate to those around them. Sometimes they may be disruptive and physically aggressive, making social relationships even more difficult. They have a tendency to "lose control", particularly when they're in a strange or overwhelming environment, or when angry or frustrated. At times, they may break things, attack others or hurt themselves. In their frustration, some bang their heads, pull their hair or bite their arms.

Communication Difficulties

By age three, most children have passed predictable milestones on the path to learning language; one of the earliest is babbling. By the first birthday, a typical toddler says a word or two, turns and looks when he hears his name, points when he wants a toy, and when offered something distasteful, makes it clear that the answer is “no”.

Some people with autism remain mute throughout their lives; although the majority develops spoken language and all eventually learn to communicate in some way. Some infants who later show signs of autism “coo” and babble during the first few months of life, but they stop. Others may be delayed, developing language as late as age five to nine. Some children may learn to use communication systems such as pictures of sign language.

Children with autism who do speak often use language in unusual ways. They seem unable to combine words into meaningful sentences. Some speak only single words, while others repeat the same phrase over and over. They may repeat or “parrot” what they hear, a condition called echolalia. Although many children with autism go through a stage where they repeat what they hear, it normally passes by the time they are three.

Some children with autism who are only mildly affected may exhibit slight delays in language, or even seem to have precocious language and unusually large vocabularies, but have great difficulty in sustaining a conversation. The “give and take” of normal conversations may be hard, although they may often carry on a monologue on a favorite subject, giving others little opportunity to comment. Another common difficulty is the inability to understand body language, tone of voice, or “phrases of speech.” For example, someone with autism might interpret a sarcastic expression such as “Oh, that’s just great” as meaning it really IS great.

While it can be challenging for others to understand what children with autism are less able to say, their body language may also be difficult to understand. Facial expressions, movements, and gestures may not match what they are saying. Also

their tone of voice may fail to reflect their feelings. They may use a high-pitched, sing-song, or flat, robot-like voice. Some children with relatively good language skills speak like little adults, failing to pick up on the “kid-speak” that is common in their peers.

Without meaningful gestures or the language to ask for things, people with autism are less able to let others know what they need. As a result, they may simply scream or grab what they want. Until they are taught better ways to express their needs, children with autism do whatever they can to get through to others. As they grow up, they can become increasingly aware of their difficulties in understanding others and in being understood. As a result, they are at greater risk of becoming anxious or depressed.

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Repetitive Behaviors

Although children with autism usually appear physically normal, odd repetitive motions may set them apart from other children. These behaviors might be extreme and highly apparent or more subtle. Some children and older individuals repeatedly flap their arms or walk on their toes. Some suddenly freeze in a position.

As children, individuals with autism might spend hours lining up their cars and trains in a certain way, rather than using them for pretend play. If someone moves one of the toys, the children may be tremendously upset. Many children with autism need, and demand, absolute consistency in their environment. A slight change in routines, such as mealtimes, dressing, taking a bath, and going to school at a certain time or by the same route, can be extremely stressful.

Repetitive behavior sometimes takes the form of a persistent, intense preoccupation. These strong interests may be unusual because of their content (e.g. Being interested in fans or toilets) or because of the intensity of the interest (e.g. knowing much more detailed information about Thomas the Tank Engine than peers). For example, a child with autism might be obsessed with learning all about vacuum cleaners, train schedules, or lighthouses. Often older children with autism have a great interest in numbers/ letters, symbols, dates or science topics.

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Unique Abilities that may Accompany Autism

You may have recognized that your child with autism has some unusual skills and abilities. The information that follows, adapted from Sally Ozonoff, Geraldine Dawson and James McPartland's book, *A Parent's Guide to Asperger's Syndrome and High-Functioning Autism*, and the article, "How can my child have Autism when he seems so smart?" From *Does My Child Have Autism?* By Wendy Stone, highlight some of them.

Consider incorporating some of these abilities into your child's treatment plan, where possible and appropriate, to take advantage of them.

From A Parent's Guide to Asperger Syndrome and High-Functioning Autism

Just as individuals with autism have a variety of difficulties they also have some distinctive strength. Some of the strengths that individuals with autism may have include:

- Ability to understand concrete concepts, rules and sequences
- Strong long term memory skills
- Math skills
- Computer skills
- Musical ability
- Artistic ability
- Ability to think in a visual way
- Ability to decode written language at an early age (This ability is called **Hyperlexia**. Some children with autism can decode written language earlier than they can comprehend written language.)
- Honesty – sometimes to a fault
- Ability to be extremely focused – if they are working on a preferred activity
- Excellent sense of direction

How can my child have Autism when he seems so smart?

Right now you might be thinking about all the things your child with autism learned at a much younger age than other children you know. And yes, you are right: there are also things that children with autism learn on their own much faster than their typically developing peers or siblings. For example:

They can be very good at learning to pick out their favorite DVD from a stack, even when it's not in its case.

They may learn at a very young age how to operate the remote controls to the TV and DVD player so that they can rewind their videos to their favorite parts (or fast forward through the parts they don't like).

They can be very creative in figuring out ways to climb up on the counter to reach a cabinet that has their favorite cereal, or even how to use the key to unlock the dead bolt on the back door so they can go outside to play on the swing.

Clearly, these are not behaviors that you would even think about trying to teach a two-year-old child. And yet some children with autism somehow manage to acquire these skills on their own.

How can we understand this inconsistency between the things children with autism do and don't learn? How can a child who can't put different shapes into a shape sorter learn to turn on the TV and DVD player, put a DVD in, and push the play button? How can a child who can't understand a simple direction like "Get your coat" figure out how to unlock a door to get outside?

What accounts for this unique learning style? In a word: motivation. We all pay attention better to the things that interest us, so we become much more proficient at learning them.

Understanding what is motivating to your child (all children are different) will be one of the keys to increasing their learning and their skills.

Physical and Medical Issues

that may Accompany Autism

Seizure Disorders

Seizure Disorder, also called **Epilepsy**, occurs in as many as 39% of people with autism. It is more common in children who also have cognitive deficits than those without. Some researchers have suggested that it is more common when the child has shown a regression or loss of skills.

There are different types and subtypes of seizures and a child with autism may experience more than one type. The easiest to recognize are large “grand mal” (or tonic-clonic) seizures. Others include “petit mal” (or absence) seizures and subclinical seizures, which may only be apparent in an EEG (Electroencephalogram). It is not clear whether subclinical seizures have effects on language, cognition, and behavior.

The seizures associated with autism usually start either early in childhood or during adolescence, but may occur at any time. If you are concerned that your child may be having seizures you should see a neurologist. The neurologist may order tests which may include an EEG, an MRI (Magnetic Resonance Imaging), CT (Computed Axial Tomography) and a CBC (Complete Blood Count).

Children and adults with epilepsy are typically treated with anticonvulsant or seizure medicines to reduce or eliminate occurrence. If your child has epilepsy, you will work closely with a neurologist to find the medicine that works the best with the fewest side effects and to learn the best ways to ensure your child’s safety during a seizure.

Genetic Disorders

A small number of children with autism may also have an identifiable neurogenetic condition such as **Fragile X Syndrome**, **Angelman’s Syndrome**, a neurocutaneous disorder called **Tuberous Sclerosis**, **Chromosome 15 Duplication Syndrome** or another chromosomal abnormality.

If your child has clinical features, such as a family history or physical symptoms, that are characteristic of one of these disorders, your pediatrician may order tests or may refer you to a developmental pediatrician, a geneticist and/or a child neurologist for testing. The chance of having one of these abnormalities is a little

higher if your child also has cognitive deficits or mental retardation. It is also higher if your child has certain physical features that are characteristic of a given syndrome.

While none of these conditions is curable, it is important to know if your child has one of these syndromes because there may be other medical issues that go along with some of them. Having a known genetic cause for autism may also change your risk of having another child with autism.

Gastrointestinal Disorders

Many parents report gastrointestinal (GI) problems in their children with autism. The exact number of children with both gastrointestinal issues such as gastritis, chronic constipation, colitis, celiac disease and esophagitis and autism is unknown. Surveys have suggested that between 46 and 85% of children with autism have problems such as chronic constipation or diarrhea.

One recent study identified a history of gastrointestinal symptoms (such as abnormal pattern of bowel movements, frequent constipation, frequent vomiting, and frequent abdominal pain) in 70% of the children with autism, compared with 42% of children with other developmental disabilities and 28% of children without developmental disabilities.

If your child has symptoms such as chronic or recurrent abdominal pain, vomiting, diarrhea, or constipation, you will want to consult a **gastroenterologist** (preferably one that works with people with autism). Your child’s physician may be able to help you find an appropriate specialist. Pain caused by GI issues is sometimes recognized because of a change in a child’s behavior, such as an increase in self-soothing behaviors such as rocking or outbursts of aggression or self-injury. Bear in mind that your child may not have the language skills to communicate pain caused by GI issues. Treating GI problems may result in improvement in your child’s behavior.

A popular dietary intervention for GI issues includes the elimination of dairy and gluten containing foods (for more information see Gluten Free Casein Free diet in the treatment section of this kit). As with any treatment it is best to consult your child’s physician to develop a comprehensive plan.

In February 2007, Autism Speaks initiated a campaign to inform pediatricians about the diagnosis and treatment of GI problems associated with autism. For additional information that can be

shared with your child's doctor, go to the following link on the *Autism Speaks* website: www.AutismSpeaks.org/docs/Autism_Speaks_GI_Statement_1-07.pdf

Sleep Dysfunction

Is your child having trouble getting to sleep or sleeping through the night? Sleep problems are common in children and adolescents with autism. Having a child with sleep problems can affect the whole family. It can also have an impact on the ability of your child to benefit from therapy.

Sometimes sleep issues may be caused by medical issues such as **obstructive sleep apnea** or **gastroesophageal reflux** and addressing the medical issues may solve the problem. In other cases, when there is no medical cause, sleep issues may be managed with behavioral interventions including “sleep-hygiene” measures such as limiting the amount of sleep during the day, and establishing regular bedtime routines.

There is some evidence of abnormality of **melatonin** regulation in children with autism. While melatonin may be effective for improving the ability of children with autism to fall asleep, more study is needed. Melatonin or sleep aids of any kind should not be given without first consulting with your child's physician.

Sensory Integration Dysfunction

Many children with autism experience unusual responses to **sensory stimuli**, or input. These responses are due to difficulty in processing and integrating sensory information. Vision, hearing, touch, smell, taste, the sense of movement (**vestibular system**) and the sense of position (**proprioception**) can all be affected. This means that while information is sensed normally, it may be perceived much differently. Sometimes stimuli that seem “normal” to others can be experienced as painful, unpleasant or confusing by the child with **Sensory Integration Dysfunction (SID)**, the clinical term for this characteristic. (SID may also be called **Sensory Processing Disorder** or **Sensory Integration Disorder**.)

SIDs can involve hypersensitivity, also known as **sensory defensiveness**, or **hyposensitivity**. An example of hypersensitivity would be the inability to tolerate wearing clothing, being touched, or being in a room with normal lighting. Hyposensitivity might be apparent in a child's increased tolerance of pain or a constant need for sensory stimulation.

Treatment for **Sensory Integration Dysfunction** is usually addressed with occupational therapy and/or sensory integration therapy.

Pica

Pica is an eating disorder involving eating things that are not food. Children between 18 and 24 months old often eat non food items, but this is typically a normal part of development. Some children with autism and other developmental disabilities persist beyond the developmentally typical time frame and continue to eat items such as dirt, clay, chalk or paint chips.

Children showing signs of persistent mouthing of fingers or objects, including toys, should be tested for elevated blood levels of lead, especially if there is a known potential for environmental exposure to lead.