WHY WAS MY CHILD DIAGNOSED WITH AUTISM? WHAT DOES IT MEAN?

Your child has been diagnosed with autism or autism spectrum disorder and you have asked for help. This is an important turning point in a long journey. For some families, it may be the point when, 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.

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 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 and a cure, along with effective prevention and treatment methods. Great strides have been made and the current 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 are working at a constantly increasing pace toward a cure for autism. While 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 that your child is the same unique, lovable, wonderful person he or she was before the diagnosis.

There are however, several reasons having a diagnosis is important for your child. A thorough and detailed diagnosis provides important information about your child’s behavior and development. It can help create a road map for treatment, by identifying your child’s specific strengths and challenges and providing useful information about which needs and skills should be targeted for effective intervention. A diagnosis is often required to access autism specific services through early intervention programs or your local school district.

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. 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 as a result, 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 visits. The American Academy of Pediatrics recommends that all children be screened for autism at their 18- and 24-month well-baby check-ups. If concerns about a child’s development are raised, his or her doctor should 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. (http://www.dbpeds.org/media/mchat.pdf)
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. A multi-disciplinary evaluation is important for diagnosing autism and other challenges that often accompany autism, such as delays in motor skills. If your child has not been evaluated by a multi-disciplinary team, you will want to make sure further evaluations are conducted so that you can learn as much as possible about your child’s individual strengths and needs. For more information you can visit The Autism Treatment Network at: www.autismspeaks.org/science/programs/atn/

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 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 that at least six developmental and behavioral characteristics are observed, that problems are present before the age of three, and that there is no evidence for certain other conditions that are similar. The DSM-IV is currently being revised. The DSM-V will group together the subtypes of autistic disorder, PDD-NOS, and Asperger Syndrome under the umbrella term “Autism Spectrum Disorder” because research has not shown these categories to be distinct. Rather, they are part of a broad continuum of disorders that involve difficulties in social and communication skills.
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.
• Apparent 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 88 children is diagnosed with autism in the United States, making it more common than childhood cancer, juvenile diabetes and pediatric AIDS combined. This marks a 23% increase in prevalence in the past two years. There is no established explanation for this increase, although improved diagnostic methods 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 54 boys is diagnosed with autism.

**What causes Autism?**

It is important to keep in mind that autism is not one disorder with one cause. Rather, it is a group of related disorders with many different causes. In most instances, autism is likely caused by a combination of genetic risk factors that interact with environmental risk factors. Several autism susceptibility genes have been identified, meaning that an individual will be more likely to develop autism if they have a specific variant of this gene, or in some cases, a rare mutation in the gene. Many genes likely contribute to autism. These specific genes are believed to interact with certain environmental factors. A great deal of research is currently focused on identifying how both genetic and environmental risk factors contribute to autism. Although some genetic factors have been identified, less is known about the specific environmental factors that contribute to autism. Exposure to environmental agents such as infectious agents (maternal rubella or cytomegalovirus) or chemical agents (thalidomide or valproate) during pregnancy can cause autism. About 10-15% of cases have a specific, identifiable genetic cause, such as such as Fragile X Syndrome, Tuberous Sclerosis, and Angelman’s Syndrome.

There is a growing interest among researchers about the role of the function 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 experience 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 causes of autism are complex, 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 a biological disorder, and is not caused by cold parents.
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 child in 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.

Although autism is usually a life-long condition, the symptoms of autism can change over time. The long term outcome is highly variable. Some children lose their diagnosis over time, while others remain severely affected. Many have normal cognitive skills, despite challenges in social and language abilities. Most individuals with autism develop speech and learn to communicate with others. 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 partially 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 smile. By contrast, many children with autism have difficulty learning to engage in the give-and-take of everyday human interactions. Even in the first year of life, many may prefer playing with objects and may fail to initiate social interaction or engage in communicative babbling and imitative play. Eye contact may be fleeting. Most have difficulty using gestures such as pointing, waving bye-bye, and showing objects to others. 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.

Children with autism are also 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 thoughts, feelings,
and goals that are different from their own. 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 they are 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.

Fortunately, children with autism can be taught how to socially interact, use gestures, and recognize facial expressions. Also, there are many strategies that can be used to help the child with autism deal with frustration so that they don’t have to resort to challenging behaviors. We will discuss this later.

**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”. Although a minority of people with autism doesn’t use speech, the large majority develops spoken language, and all eventually learn to communicate in some way. Most infants who later show signs of autism “coo” and babble during the first few months of life, but over time, 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 or 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 turn 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 still have great difficulty in sustaining a conversation. The “give and take” of normal conversations may be hard, but 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. It can be challenging sometimes for others to understand what children with autism are saying, as well as what their body language means. 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 have difficulty letting others know what they need. As a result, they may simply scream or grab what they want. Fortunately, children with autism can be taught to communicate in more appropriate way.
Children with autism have difficulty letting others know what they want or need until they are taught how to communicate through speech, gestures, or other means.

**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 with autism 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 become tremendously upset. Many children with autism need and demand absolute consistency in their environment. A slight change in routines, such as eating a meal, getting dressed, 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. fans or toilets) or because of the intensity of the interest (e.g. extremely detailed information about Thomas the Tank Engine). 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.

*Many children with autism need, and demand, absolute consistency in their environment.*
UNIQUE ABILITIES THAT MAY ACCOMPANY AUTISM

Along with the challenges that autism involves, you may have noticed that your child also exhibits areas of strength. Although not all children have special talents, it is not uncommon for individuals with autism to have exceptional skills in math, music, art, and reading, among others. These areas of expertise can provide great satisfaction and pride for the child with autism. If possible, incorporate your child’s areas of expertise into his everyday activities and use them whenever possible as a way for he or she to learn and excel.

*Adapted from Sally Ozonoff, Geraldine Dawson and James McPartland’s, A Parent’s Guide to Asperger’s Syndrome and High-Functioning Autism*

*Just as individuals with autism have a variety of difficulties, they also have some distinctive strengths. Some of the strengths that individuals with autism have may 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?”
From Does My Child Have Autism? by Wendy Stone

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. Your child’s special talents may be part of his unique and inherent learning style and nature.
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 who do not. Some researchers have suggested that seizure disorder 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.

You can find more information about autism and epilepsy at the following link: http://fiddle.readvantage.com/news/attach/DJF-EpilepsyBrochure.pdf

Genetic Disorders

About 10-15% of children with autism have an identifiable neurogenetic condition such as Fragile X Syndrome, Angelman’s Syndrome, a neurocutaneous disorder called Tuberous Sclerosis, Chromosome 15 Duplication Syndrome or other chromosomal abnormalities. 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 slightly 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 as 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 prevalence of gastrointestinal problems, such as gastritis, chronic constipation, colitis, and esophagitis in individuals with autism is unknown. Surveys have suggested that between 46 and 85% of children with autism have problems such as chronic constipation or diarrhea. One 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. 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 like rocking, or outbursts of aggression or self-injury. Bear in mind that your child may not have the language skills to communicate the pain caused by GI issues. Treating GI problems may result in improvement in your child’s behavior. Anecdotal evidence suggests that some children may be helped by dietary intervention for GI issues, including 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 January 2010, Autism Speaks initiated a campaign to inform pediatricians about the diagnosis and treatment of GI problems associated with autism.

For additional information from the Official Journal of American Academy of Pediatrics go to:
http://pediatrics.aappublications.org/cgi/content/full/125/Supplement_1/S1

For information that can be shared with your child’s doctor, go to:

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 research 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 an 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 for 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.