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WHAT EXACTLY ARE AUTISM SPECTRUM DISORDERS?

What's going on? Why is the word *autism*—virtually unheard of in your parents' generation—now audible everywhere you turn? It seems as if every family has some concern or connection with autism these days—and there's no avoiding it. In the past year, autism was the subject of 116 articles in the *New York Times*. Do a search on Google and you'll discover over five million hits. Amazon.com offers 1,076 books on the subject. Given this autism overload, no wonder you may feel a sense of dread when you call your child's name and he or she is slow to respond!

Yet despite the publicity that has surrounded this disorder in the last few years, few people know what autism really is. And because a little knowledge can truly be a dangerous thing, the word has become misused and misunderstood—it's become entangled in a web of disorders with similar, often overlapping ranges of symptoms. And because the symptoms of classic autism itself can be present in different combinations and at different levels of severity, the question of whether or not your child has autism—or where your child's behavior fits within the range of possible autistic symptoms—has become very confusing.

The fact is that autism is one type of developmental disorder within a range of what are called Autism Spectrum Disorders. Autism Spectrum Disorders include Autistic Disorder, Asperger's

Disorder, Rett's Disorder, Childhood Disintegrative Disorder, and Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS). Children within the range of Autism Spectrum Disorders show impairments across a number of developmental areas, including difficulty with social interactions, disordered language development, and repetitive activities and behaviors. These symptoms overlap not only from one autism spectrum disorder to another, but also with other nonautistic developmental disorders such as language delay or global developmental delay. This is why it's often difficult for health care professionals* to diagnose autism quickly—and why it is so intolerably frustrating for parents who think they see symptoms that might spell trouble—but might not—and can't get a quick and clear answer.

With other childhood illnesses, we're accustomed to familiar and conventional symptoms and rapid diagnosis. Is your baby crying, suffering a slight fever, and tugging on his ear? A medical evaluation is likely to show signs of an ear infection *(otitis media)* that's treated with a ten-day course of antibiotics. Symptoms, signs, diagnosis, treatment—done. But along come symptoms that the television and print media, family, and friends say look like autism, and suddenly no one can quickly and definitively answer your questions. It's ambiguous; it's hard to pin down; it's alarming.

HOW THIS BOOK CAN HELP YOU

I understand your distress and confusion. I'm a mother. I'm also a psychologist who has worked with hundreds of families who have children with Autism Spectrum Disorders. I have made it my life's work to come up with the information that will help you map your way through the jargon, complexities, and frustration of autism. I

*The term *health care professional* refers to professionals who diagnose and treat mental health or physical health conditions. The diagnosis of autism is most commonly made by clinical psychologists, psychiatrists, and developmental pediatricians.

know that you wouldn't be reading this book if you didn't already have concerns about your child's development, so I have written it to speak directly to you. I imagine sitting with you in the clinic, seeing the worry in your eyes as you ask, "Does my child have autism?" And I understand that you would want an accurate and quick yes-or-no answer. After all, you realize that the sooner you can have a firm diagnosis, the better for your child. And that explains that sense of urgency I hear in your voice.

If you really were sitting here with me, I would be the first to agree that early detection and intervention are so very important for your child. But before I could answer your question, we would both need to gather the facts and then carefully put all the pieces together. And that's exactly what we'll do through the chapters of this book.

In this first chapter, we discuss the signs and symptoms of the Autism Spectrum Disorders. Before you move on to any other chapter, please read this information carefully and then heed this advice: don't jump to conclusions if you see evidence of some of the symptoms in your child. There are many children, for example, who do not speak the expected fifty words by the age of two, yet do not have autism. So wait to get all the facts before you make yourself crazy with worry.

Let's take this exploration of autism one small step at a time. In this first chapter, take time to gain a better understanding of Autism Spectrum Disorders *in general*. This is the foundation you need *before* you can observe and evaluate your own child using the guidelines given in Chapter Two. Then with the Chapter Two checklist in hand, you can share your concerns and observations with your child's pediatrician.

If you and the pediatrician decide that an evaluation is warranted, you can refer to Chapter Three for some guidelines for what to expect during this process. Then, if necessary, you can move on to Chapters Four and Five regarding appropriate interventions and education. Please try to be patient; jumping ahead or to conclusions won't lead you to the information you need to help your child.

By the Numbers

The number of children diagnosed with autism has increased dramatically over the past ten years, making it the fastest-growing developmental disability in the nation. In fact, the Centers for Disease Control and Prevention has found that autism now affects an estimated 1 in 250 births, and that this diagnosis is growing at a rate of 10 to 17 percent each year. At this rate, the Autism Society of America estimates that autism could affect four million Americans in the next decade.[1] The reasons for this increase are complex and include better recognition, earlier diagnosis, and more inclusive diagnostic criteria.

THE MYTH VERSUS THE REALITY

You've probably heard through the media or in discussions with other parents that children with autism live in a world of their own, are unresponsive to their environment, and make odd, repetitive movements, such as rocking or flapping their arms. This is sometimes true—but not necessarily. TV news reports and special programs almost always show the most severe cases; those children make for "good" programming.

I saw this firsthand when a TV crew came to film some children with autism at the TRIAD clinic at Vanderbilt Kennedy Center and Vanderbilt Children's Hospital in Nashville, Tennessee. The children were well behaved despite the presence of so many extra adults in the room and lots of equipment, including hot, bright lights for the camera. I was delighted that we'd be able to show viewers images of children with autism who played with toys or sat at a table to work, rather than spent their time making odd noises or engaging in repetitive actions. But the producer of the show was not so happy. She kept insisting that her reporter get more footage of children who were demonstrating the most unusual behaviors,

and she kept directing the cameraman to focus exclusively on those with the most extreme symptoms—not on the majority of children who were calm and well behaved. Clearly, some members of the media do not want to see children with autism who look just fine.

This need to focus on only the sensational side of a story perpetuates one of the myths surrounding the fear of autism—that these children are entirely nonfunctioning. This is not true. Children with autism can vary tremendously from one another in the way they show the symptoms of this disorder, and that's why such vague terms as *autistic-like*, *autistic tendencies*, and *high-functioning* or *low-functioning autism* are sometimes used to differentiate the symptoms of one child from those of another.

In truth, if you had the opportunity to view a room of young children with autism, you would see some children talking and others using pictures or sign language to communicate. Some children would be sitting with their peers, others sitting by themselves. Some would be working at a table, others running back and forth along a wall or climbing on furniture. Some might be laughing during a tickle game with their parents, others having a tantrum and throwing toys. Looking at this group, you would wonder, "Which of these children have autism?" The simple answer is that they all do.

UNDERSTANDING THE DIFFERENT AUTISM SPECTRUM DISORDERS

The description of children with autism gets confusing because sometimes people use different terms to refer to the same thing, and sometimes they use the same term to refer to different things. The *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV)* is the book that health care professionals use to describe the characteristics necessary for the diagnosis of specific mental health, behavioral, and developmental disorders. The *DSM-IV* has established the umbrella term *Pervasive Developmental Disorders (PDD)* to include autism and four other related disorders that share signs and symptoms. In other words, the terms *Autism Spectrum*

PARENTS SAY
What Autism "Looks" Like

- My son is a beautiful boy; you'd never guess he has any issues just by looking at him. Autism isn't always something that can be noticed by outward appearance, so when he begins to melt down in public, people act as if he's an undisciplined nuisance, and they treat me as if I'm a bad parent. (mother of three-year-old boy)
- Most people don't know how broad the autism spectrum is. They assume autism is much more severe than it is in our son (think Rain Man) and often suspect that we are hypochondriac parents since our son is high functioning. My sister gasped in horror when I first told her about my son's diagnosis—I guess she thought he was somehow doomed to live in an institution or something. (mother of five-year-old boy diagnosed at age two)
- Movie and television characters with autism tend to be on the severe side. I know that only trouble is interesting, but it does my son no good to portray autism as something akin to severe retardation or freakishness. And it tends to keep parents of newly diagnosed children from overcoming their sense of denial. (mother of five-year-old boy)

Disorders and *Pervasive Developmental Disorders* are often used to mean the same thing. Both terms refer to a class of disorders that includes social deficits, communication impairments, and restricted, repetitive activities as their primary characteristics. **For the purpose of this book, I will use the term *Autism Spectrum Disorders* (or ASDs) to refer to autism and related disorders that share the core features,**

and the term *Pervasive Developmental Disorders* (or PDDs) only when describing specific *DSM-IV* categories or diagnostic criteria.

The five disorders categorized as Pervasive Developmental Disorders in *DSM-IV* are (1) Autistic Disorder, (2) Asperger's Disorder, (3) Rett's Disorder, (4) Childhood Disintegrative Disorder, and (5) Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS). These disorders differ from each other on features that include their prevalence, severity, and the way in which their symptoms appear and progress.

As you begin your exploration of autism, it's important to take some time to become familiar with each of the five Pervasive Developmental Disorders. So let's go through them briefly.

Autistic Disorder

Autistic Disorder is the formal term for what is more commonly called "autism." This disorder is characterized by a pattern of severe impairments in three areas: (1) difficulties interacting with others in a reciprocal way, (2) impaired language and communication skills, and (3) a repetitive and restricted range of interests and activities. (See Appendix A for specific *DSM-IV* criteria for Autistic Disorder.) These symptoms emerge before the age of three, but can change over time and vary widely from one child to another. (We'll be looking at each symptom more fully later in the chapter.) For reasons that are unclear at this time, boys are more likely to be diagnosed with Autistic Disorder by a ratio of approximately 4:1.

Asperger's Disorder

The name "Asperger" comes from Hans Asperger, an Austrian physician who first described the disorder in 1944. This PDD describes children (predominantly boys by a 5:1 ratio) who have average intelligence and who do not have a history of delayed language development. They do, however, have social impairments and restricted, repetitive interests. Their social interactions can be

awkward and one-sided, and they often have difficulty understanding the perspectives of others. They may talk unceasingly about some object or topic of fascination, not understanding the give-and-take of conversation or the art of listening.

This diagnosis is used only when the impairments are severe and sustained and interfere with the child's functioning at home, school, or in the community. Many times the symptoms of Asperger's are not evident until the child begins school, which is why this diagnosis is not often made in children under age three.

Rett's Disorder

Named for Dr. Andreas Rett, an Austrian physician who first described the disorder in a journal article in 1966, Rett's Disorder is very rare, affecting girls almost exclusively. Children with this disorder develop normally and on schedule through early infancy. But then sometime between six and eighteen months, they gradually begin to lose skills in different areas of functioning. Children who had been talking, stop. Their ability to interact with others decreases. They also lose the use of their hands to hold and manipulate objects and begin to show repetitive hand movements such as hand rubbing, clasping, or wringing. During this phase of their regression, they show symptoms similar to those seen in Autistic Disorder.

The symptoms of Rett's Disorder are progressive and worsen over time. Rett's Disorder is the only PDD that has an identified genetic cause: an abnormality of a gene on the X chromosome.

Childhood Disintegrative Disorder (CDD)

CDD is a very rare condition that involves a significant regression in skills in children who have had typical development for the first two years of life. (This lengthy period of typical development is a key difference between Rett's Disorder and CDD.) Between the ages of two and ten, children with CDD lose some or all of the skills they have already developed in areas that include language, social skills, play,

and motor skills. Following this period of regression, their behavior stabilizes (also unlike Rett's Disorder), though children usually have severe mental retardation and may also have seizures.

Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS)

PDDNOS is what's called a "diagnosis of exclusion." It is used only when a child demonstrates symptoms of PDD that do not fit the criteria specified for one of the other disorders in this category. Specifically, the diagnosis of PDDNOS is used for children who are impaired in their social interactions along with *either* an impairment in the development of language and communication skills *or* a pattern of restricted or repetitive behaviors and activities.

For example, a child may receive a diagnosis of PDDNOS rather than Autistic Disorder if he or she does not show all the symptoms required for a diagnosis of Autistic Disorder or if his or her symptoms are milder in nature. The diagnostic criteria used for PDDNOS are less exact than those for the other diagnostic classifications within the category of PDD.

I'm sure that the differences between each PDD are now clear as mud to you. For further clarification, you can refer to Table 1.1, but surely you will still wonder, "So what does all this have to do with autism?" and "How can I tell if my child has autism and not one of the other PDDs?" Keep reading!

FOCUSING ON AUTISTIC DISORDER AND PDDNOS

To simplify the answers to those questions, I am going to pull out two of the PDDs to focus on for the rest of this book: Autistic Disorder and PDDNOS. These are the disorders that are seen and diagnosed

TABLE 1.1. Characteristics of Pervasive Developmental Disorders.

Characteristic	*Autistic Disorder*	*Asperger's Disorder*	*PDDNOS*	*Childhood Disintegrative Disorder*	*Rett's Disorder*
Social impairment	X	X	X	X[b]	X
Language and communication disorder	X		X[a]	X[b]	X
Repetitive interests and activities	X	X	X[a]	X[b]	
Average intelligence		X			
Onset prior to 36 months	X				X
Period of normal development followed by loss of skills in several areas				X	X
Relative impairment	Variable	Milder	Milder	More severe	More severe
Relative prevalence	Higher	Intermediate	Higher	Lower	Lower

Note: [a]At least one of these two features must be present.
[b]At least two of these three features must be present.

most commonly in children under three, and are therefore most relevant to you and your young child as you look for the keys to early detection and intervention. Rett's Disorder and Childhood Disintegrative Disorder are quite rare and are very different in course and outcome from Autistic Disorder and PDDNOS. Asperger's Disorder is rarely detected before the age of three, and it does not present the very worrisome early symptoms that send parents rushing to their child's pediatrician for answers. If you are wondering if your child under the age of three has symptoms of autism, Asperger's is unlikely to be the diagnosis.

The differences between Autistic Disorder and PDDNOS can be quite subtle and often difficult to determine in young children. Compared to children diagnosed with Autistic Disorder, children with PDDNOS typically have milder symptoms, atypical symptoms, or both. Still, one health care professional might determine that a young child has PDDNOS, whereas another, who is equally competent, might determine that the same child has Autistic Disorder. **For the purpose of this book, which is focused on the young child, I will use the term *autism* to refer to both Autistic Disorder and PDDNOS.**

COMMON SYMPTOMS OF AUTISM

As indicated previously, there can be a lot of variability in the way that autism is expressed from one child to another. Here's why:

1. No two children are alike, whether they have autism or not. In the same way, each child with autism is an individual, with his or her own personality and unique characteristics.
2. Each child with autism displays a range of behaviors, some of which look just like what we expect for his or her age and some of which are different—or unusual—compared to other children. This behavioral variability is exactly what makes autism hard to identify in young children.

But whatever the range, intensity, or frequency of symptoms, they will include atypical development in these three primary areas: (1) social skills, (2) language and communication skills, and (3) repetitive and restricted behaviors. Although the symptoms in each area can vary from one child with autism to the next, let's take a look at what is typically found in these children.

Symptom 1: Impaired Social Skills

Humans instinctively interact with each other. Even in infancy, babies are interested in faces; they like close physical contact with caregivers; they turn toward voices and smile with recognition at familiar family members. As they grow, babies learn how to be social and interactive by watching how others talk, play, and relate with each other. They enjoy the give-and-take of social engagement and will initiate, maintain, and respond to interactions with others. In fact, they seek out those interactions.

Children with autism, however, often do not show the expected development of early social interaction skills. They seem not to have the same "drive" to interact socially as their peers do. In fact, impaired social interactions are the hallmark of autism and are present in *all* children with this diagnosis. ***If your child does not have difficulty initiating or responding to interactions with others, then your child does not have autism.***

These social impairments affect children's interactions with adults as well as with other children. They affect children's ability to initiate interactions with others as well as to respond to interactions that are initiated by others. For example, unlike other children, children with autism *may not*

- Pay attention to adults, even when they are close by
- Smile in response to praise or an adult's smile
- Respond when an adult calls their name
- Initiate social interactions with adults or peers

- Show enjoyment in interactive or turn-taking games with adults, such as patty-cake or peek-a-boo
- Imitate actions of adults, such as waving good-bye
- Repeat actions that adults respond to with praise or attention
- Show interest in other children, such as watching them or playing near them
- Join another child in play
- Play interactive, back-and-forth games with other children
- Show an interest in making friends
- Imitate the actions of other children
- Initiate play with other children, such as greeting them or handing them a toy

Each of these symptoms of social impairment may vary in frequency and intensity from one child with autism to another, but social deficits are an important marker of the disorder.

Symptom 2: Impaired Language and Communication Skills

Problems with language and communication can take many forms in children with autism. To understand these symptoms better, you need to understand the difference between communication, language, and speech:

Communication is a *process* through which someone conveys a message to another person. Communication can be verbal, which involves using words, or nonverbal, which involves using other behaviors, such as crying, reaching, gesturing, or facial expressions. In contrast, *language* refers to a *system* of communication in which conventional symbols are used to convey a message. Examples of conventional symbols are sign language, gestures, and words. Crying is not a symbol, nor is pulling on an adult's hand. *Speech* refers

specifically to a *form* of language in which spoken words are used to communicate.

The most commonly recognized characteristic of autism in the domain of language and communication is the delayed development of spoken language. Nearly all children with autism are delayed in reaching their language milestones (see "Language Development Milestones" in Chapter Two). But the problems often go deeper than just language. Many children with autism do not understand the process of communication at all—they don't seem to know that there is a way for them to convey their needs and desires to other people. They don't know how to ask for help or ask for more or make a choice, other than by fussing or crying. Of course this deficit is terribly frustrating for parents—but imagine how frustrating it must be for the child!

Similar to their impairments in the social domain, children with autism have difficulty initiating communication as well as responding to the communication of others. For example, unlike other children with typical language and communication skills, children with autism *may not*

- Look at people in the eye during playful interactions
- Follow another person's point by looking in the direction indicated
- Express their needs or desires to others in conventional ways, such as reaching and vocalizing
- Use nonverbal gestures, such as waving good-bye or nodding or shaking their head
- Look at other people's faces to seek information
- Communicate for the purpose of sharing their interests or achievements with others, such as pointing to objects or holding them up to show to others
- Engage in back-and-forth babble "conversations"

Even children with autism who eventually develop spoken language still have impairments in this area. Like many children learn-

ing to speak, they may echo back words and phrases they hear. But unlike other children, those with autism may mimic the exact intonation of the speaker and persist in this echoing (called echolalia) long after other children have moved on to interactive speech, usually by three years of age.

Sometimes children with autism echo words or phrases they have just heard; this is called immediate echolalia. An example is a child's answering the question, "Do you want to go outside?" with "Go outside." Another type of echolalia occurs when children repeat things they have heard in another context (delayed echolalia). Examples of delayed echolalia occur when children repeat dialogue from videotapes or things that they have heard their teacher say at school. One of the most baffling things about autism is that some children can repeat long segments from favorite videotapes or books, but cannot use words functionally, to achieve their goals, such as by requesting a cookie or favorite snack.

Symptom 3: Restricted Interests and Repetitive Activities

Some children with autism become preoccupied with a particular activity, toy, or interest in a way that is unusual in its intensity. They may, for example, spend hours opening and closing the door on a toy car. Or they may line up puzzle pieces over and over again—never actually putting them into the puzzle.

These children may also find comfort and security in repetition of certain routines, patterns, or rituals. They may insist on following a certain pattern during dressing: socks first, pants next, then shirt, and so on. They may need to have the same plate and cup during mealtime. Sometimes, if their set routine is changed, they will explode in a temper tantrum of frustration.

Young children with autism may also show unusual repetitive movements that seem to serve no particular function. For example, some children may repeatedly flap their hands, flick their fingers, or

spin around in a circle. Much more rarely, they may show self-injurious behaviors such as banging their heads or biting their hands.

Some children with autism also demonstrate unusual sensory responses, which may vary widely from child to child. For example, some children enjoy rubbing certain surfaces or are hypersensitive to the feel of new clothes. Many children avoid foods that have certain textures (though we certainly see picky eating habits in young children without autism as well!). Some children are very attentive to small details, such as a piece of thread on the floor, yet stumble over large objects in their paths. Some children appear distressed when they hear certain sounds, such as a vacuum cleaner or hair dryer, yet do not respond when a parent calls their name.

Despite the variation in the types and extent of restricted interests and repetitive activities, some examples of behaviors that may be observed are

- Engaging in repetitive play activities, such as lining up toys or spinning objects
- Acting out repetitive movements, such as running in circles or flicking their fingers
- Showing prolonged visual interest in objects, such as flapping objects in front of their eyes or staring at mirrors or objects that spin
- Having overly focused interest in one object or activity, such as a fascination with boats or bugs
- Demanding rigid adherence to rituals and routines
- Focusing attention on small parts of toys, such as the wheels on a toy truck, rather than the whole

Repetitive activities often go hand in hand with impaired play skills. Around the age of eighteen months, most children will flex their imaginations by turning a banana into a telephone, a bowl into a hat, or a clothespin into a little man. Because young children with autism often have very literal thinking patterns, they may not be able to see that a stick is anything more than a stick—not

a sword, plane, or totem pole, as other children may imagine. They may not pretend that their teddy bear is injured and in need of a hug.

With respect to play skills, children with autism *may not*

- Play with a variety of toys
- Use toys the way they're designed to be used (such as stirring a spoon in a bowl)
- Arrange toys in their intended scheme (such as placing toy dishes on a table)
- Show functional play with dolls, stuffed animals, or toy figures (such as feeding a doll or putting a toy figure in a car for a ride)
- Create play sequences (such as putting the little people on the toy bus, driving the bus to its pretend destination, and then taking all the little people out of the bus)
- Play with toys in a variety of ways (such as pushing a toy car back and forth as well as filling the car with gas, driving it around, rolling it down a hill)

Early problems related to the development of social skills, language and communication skills, and restricted and repetitive interests and activities are the primary symptoms used to diagnose autism. Exactly how you and your child's health care professional should evaluate these developmental problems is explained in later chapters. For now, in this early step, be sure you have a clear understanding of how these three symptoms show themselves *in general* among children with autism.

Because the symptoms in each of the three domains that characterize autism can vary so widely from one child to the next, it's far too early in your exploration of autism to make any assumptions about your own child. You'll see in Table 1.2 examples of how autism can show itself in different ways.

Table 1.2. Variability in Symptom Expression: Three Two-Year-Olds with Autism.

Jacob	*Will*	*Amy*
	Social Behaviors	
• Is very attached to his mother; likes to be in the same room with her; will climb up on her lap to twirl her hair or pat her cheek; is very clingy to her in new situations • Is less responsive to his father, but has a favorite game in which he climbs up his father's legs to be flipped over • Shows little interest in other children; sometimes watches them, but keeps his distance	• Mostly stays to himself; can occupy himself for hours on end; doesn't seek out parents as play partners • Shows little interest in his baby brother or older sister; gets upset when the baby cries; will occasionally play a chase game with his sister • Becomes upset in new places or when approached by adults other than his parents; does not like to be held and is not easily calmed when upset	• Is very active and happy; likes to run around the house; will tolerate her parents' presence as long as they don't try to get her to do things • Can sit and watch videotapes for hours, but does not sit at the table during mealtimes for more than a few minutes at a time • Likes to go to the park and run around; does not pay attention to the other children, even when they try to get her to play

Communication Behaviors		
• Constantly jabbers, but is hard to understand; vocalizations seem to be more for his own entertainment than a way to communicate; sometimes seems to be acting out scenes from videotapes • Every now and then will say a new word, but does not use any words consistently	• Is very quiet; rarely makes any sounds at all, other than humming or crying • Cries when he wants something, but does not indicate what he wants by pointing or reaching; parents have to guess what he wants by offering him different objects; will push away things he doesn't want	• Usually tries to get things herself rather than ask for help; will climb on tables and chairs to reach things • Sometimes will pull a parent by the hand to the kitchen when hungry or to the door to go outside • Learned the alphabet by eighteen months; will call out letters when she sees them, but does not use words
Restricted and Repetitive Behaviors		
• Likes to watch certain videos; can independently get the tape he wants and operate the VCR • Will watch the same video for several weeks at a time; covers his ears and runs out of the room during certain parts; will replay the credits over and over • Becomes very upset if he is not able to watch the complete tape	• Favorite toys are cars and any vehicles with wheels • Likes to line up cars and arrange them on the floor and in shoe boxes; becomes very upset if his sister tries to play along with him or disturbs his arrangement • Often lies on the floor to watch the wheels as he rolls the cars near his eyes	• Doesn't play with any of her toys; prefers household objects instead • Favorite activities are to line up and sort forks and spoons and to roll soda bottles back and forth across the floor • Always has to have a fork or spoon in her hand; will sometimes hold it up close to her eyes to examine it or flip it in front of her face

THE CAUSE OF AUTISM

The only honest answer that I can give to the question "What causes autism?" is this: no one knows for sure. This is a very unsatisfying response, I know, but it's the best any of us in the field can come up with at this time.

I've seen the books that dedicate entire chapters to the exploration of a cause. Perhaps, they say, it is caused by fragile X syndrome or phenylketonuria or neurofibromatosis; or it may be caused by viral infections, such as congenital rubella, cytomegalovirus, or herpes encephalitis; or maybe it's due to metabolic conditions, such as abnormalities of purine synthesis or carbohydrate metabolism; it might go back to problems during pregnancy; or it may be caused by the child's immune system or sensitivity to certain foods; and of course we've all heard the possibility that there is a link between autism and the use of thimerosal, a mercury-based preservative present in the measles-mumps-rubella (MMR) vaccine. (Although mercury is no longer used in childhood vaccines in the United States and many large-scale studies have failed to show a link between thimerosal and autism, the theory persists.)

So what is the answer? We do know that autism affects the way the brain develops, because the brain is responsible for the functions that are impaired in autism. And we know that parents do not cause their child's autism. Scientists involved in autism research believe that there is no one single cause of autism that operates for all children. More than likely, we will find that there are different combinations of factors that can cause this disorder in different children—perhaps explaining why the symptoms differ so much from one child to the next.

Fortunately, the scientific community is now beginning to unlock some of the mysteries that surround autism. For example, we do know that genetic factors can increase a child's vulnerability or risk for having autism. This type of genetic influence is different from that seen in other disorders such as Down syndrome or cystic fibrosis, in which a genetic mutation actually causes the disorder.

Testing of twins has found that if one identical twin (twins from the same fertilized egg and therefore genetically identical) has autism, it is highly likely (better than a 50:50 chance) that the other twin will also have autism. If autism were purely genetic, the risk would be 100 percent. But autism does run in families. When one child in the family has autism, his or her siblings are more likely also to have autism compared to siblings in nonaffected families. However, scientists have not yet identified the specific combination of genes that act together to increase children's vulnerability to autism.

Many families will question how their child with autism could have inherited the disorder, when no one else in their extended family has autism. In these cases, it's not unusual to find that there are some family members who, upon close examination, do have some behaviors that are consistent with autism-like symptoms. For example, they may be socially awkward, may have had language delays as a child, may have highly restricted interests, or may have some combination of these. These symptoms are often milder than those seen in individuals diagnosed with autism and are referred to as the "broader autism phenotype." (A phenotype is the physical or behavioral expression of genes.)

The genetics of autism are complicated because the inherited vulnerability for autism may not result in behavioral symptoms of autism for all children. Scientists believe that a genetic tendency toward autism must operate in combination with other, noninherited factors—such as environmental influences—in order for a child to express the characteristic behaviors of autism.

For example, early environmental factors might include interruption of oxygen to the baby's brain during birth, the mother having German measles during the pregnancy, or even exposure to a pesticide. Each of these conditions may negatively influence brain development, but I have to emphasize that the exact combination of underlying genetic and environmental factors that causes autism is one of the large missing pieces in this complex puzzle.

Researchers have also determined that a child is not at increased risk for autism because of his or her racial, ethnic, geographical, or

socioeconomic background. For unknown reasons, however, boys are three to four times more likely than girls to have autism. But keep in mind that males are at higher risk than females for many developmental problems, including Attention-Deficit/Hyperactivity Disorder and learning disabilities.

There are far more unknowns than knowns about the cause of autism at this time. I believe the discovery of the elusive cause (more likely, causes) will come only after a long and complex search that involves collaboration among researchers specializing in the biological, brain, and behavioral sciences.

THE IMPORTANCE OF EARLY DETECTION AND INTERVENTION

The cause of autism is not yet known, and there is no known cure at this time. But all is not hopeless. Research has identified many educational practices that are effective in helping children with autism improve their skills and behavior (see Chapter Four). The outcomes for children with autism who are diagnosed early and who receive specialized early intervention services are much better today than in the past—in terms of their cognitive development, language ability, social skills, and overall behavioral functioning. In fact, some children who receive a diagnosis of autism at age two no longer have the diagnosis two or three years later.

It's true! I used to tell parents of children with autism that their child would probably have the disorder for the rest of his or her life. But I no longer have to say that, now that we are diagnosing autism so much earlier, and intervening more effectively. If young children diagnosed with autism receive appropriate intervention, their path of development can be altered in very positive ways. Early intervention can change the way the brain develops in these children. The percentage of children with autism who "leave the spectrum" is still small, and we don't yet have a way to predict which children will leave their diagnosis behind, but research is underway to help answer this question.

RESEARCH TODAY

There has been a great deal of discussion and research on the possibility of a link between childhood vaccines and autism. The U.S. Centers for Disease Control and Prevention (CDC) conducts and supports many of the federal studies of large populations (epidemiological studies) that have closely examined this possibility. The most carefully conducted research on the connection between autism and vaccines has not found a link. For accurate and up-to-date information about this research, visit the following Web site: www.cdc.gov/nip/vacsafe/concerns/autism/default.htm.

You can also find out more about this issue from an article called "FAQs About MMR Vaccine and Autism," published by the National Immunization Program, at www.cdc.gov/nip/vacsafe/concerns/autism/autism-mmr.htm and from another called "Autism and the MMR Vaccine," published by the National Institute of Child Health and Human Development, at www.nichd.nih.gov/publications/pubs/autism/mmr/index.htm.

Knowing this, parents can focus on what their children need right now to help with their development; they can hold on to the hope that their child will be one of those who have every indication of autism at age two but then don't have it a few years later. Most often, the children who leave the autism spectrum still have some developmental issues, such as language impairments or developmental delays, but their behavioral improvements can be truly remarkable.

FREQUENTLY ASKED QUESTIONS

Here and at the end of each chapter, I answer some of the questions about autism that I am often asked by parents of young children.

Are there more children with autism today than in the past?

It's very hard to determine whether there are more children with autism now or whether we are just better at identifying and diagnosing the disorder. Although autism was first identified in 1943, it is very possible that many individuals with autism were misdiagnosed until the 1970s or even 1980s. The higher-functioning children who used language and had average cognitive skills may have gone through the regular education system in school and may have been considered "odd" by their classmates. Others may have been diagnosed with schizophrenia due to their unusual behavior. Children functioning at lower cognitive levels were likely to have been classified as having mental retardation rather than autism. There wasn't as much effort as now to distinguish between the subtleties among the various developmental disorders.

It is definitely the case that there are more children *diagnosed* with autism today than there were in years past. This is happening in part because there is greater awareness of the signs and symptoms of this disorder in the health care community (and among parents!), so now more children are getting the proper diagnosis. Also, we are now able to diagnose autism at a much younger age than we were in years past. Adding two- and three-year-olds to the number of older children with the diagnosis will naturally raise the total number.

Finally, the criteria for diagnosing autism have changed over time to be more inclusive. Children with milder impairments are now being identified, which also contributes to the larger numbers of children with the diagnosis. So . . . yes, the number of children *diagnosed* with autism has increased, but it's less clear whether the actual number of children with autism has changed dramatically over the years.

How do I know that my two-year-old child doesn't just have a speech delay?

This is an important question, because the methods used to treat delays in spoken language are not the same as those used for

autism. There are two important ways that young children with autism differ from children with speech delays alone. First, children with speech delays (who don't have autism) are able to communicate with others—they just have difficulty using words to do so. They manage to figure out nonverbal ways of communicating their needs, likes, and dislikes, such as by pointing to things they want or wrinkling their noses to let their parents know what they *don't* like. In contrast, young children with autism typically have difficulty communicating both nonverbally and verbally.

Second, children with delays in spoken language do not show the impairments in social relating and reciprocity that children with autism do. They have more social interest and enjoy getting their parents' attention by acting silly to make them laugh or showing them a puzzle they have completed. Like other children without autism, they seek out interactions with others and enjoy the give-and-take of engaging with other people.

My wife and I have a five-year-old boy who has autism, and we just had our second child, a baby boy. How likely is it that our baby will have autism?

Because autism is a disorder in which increased risk can be inherited, your second child does have a greater chance of having autism than other children do—but the risk is still fairly low. The recurrence risk of autism for later-born siblings of children with autism ranges from 3 percent to 8 percent. That means that somewhere between three and eight children out of every hundred children born to families that already have a child with autism will receive a diagnosis of autism. Later-born siblings also have an increased risk of showing some features of autism (that is, the "broader phenotype") without demonstrating the full-blown disorder.

Don't let the information in this introductory chapter scare you. It's intended to give you a solid understanding of the characteristics

of autism and their many manifestations *in general*. This should help you separate the media hype from the facts and give you a base to stand on as you read the next chapter. It's time to turn the page and find out how to apply the facts about autism to your own young child.