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For more information, please visit the official AAP Web site for parents, www.HealthyChildren.org/bookstore.

*This book is also available in Spanish.
What Every Parent Needs to Know

Editors
Alan I. Rosenblatt, MD, FAAP
Paul S. Carbone, MD, FAAP
with Winnie Yu

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™
What People Are Saying

Parents are craving knowledge and understanding of autism spectrum disorders and this guide is a wonderful resource to build that knowledge and encourage parents to advocate so that their child lives in a world where the dignity of every person is cherished, respected, and empowered.

Timothy P. Shriver, PhD
Chairman and CEO, Special Olympics

Covers a wide range of topics in a straightforward, commonsense manner, such as diagnosis, different behavioral therapies, family stress issues, and lots of other vital information. Essential reading for parents of young children with autism.

Temple Grandin
Author, Thinking in Pictures: My Life With Autism

Outstanding book and a tremendous resource for parents of children with autism spectrum disorders. It is also a must-read for anyone else working to help children with autism reach their full potential. The AAP and the book’s editors, pediatricians Drs Alan Rosenblatt and Paul Carbone (who also happens to be the parent of a child with autism), know what they’re talking about!

Richard E. Besser, MD, FAAP
Chief Health and Medical Editor, ABC News

Parents looking for a resource after an autism diagnosis will pull Autism Spectrum Disorders: What Every Parent Needs to Know off the shelf with great frequency. The AAP and Drs Carbone and Rosenblatt cover complex issues in an accessible manner. I will recommend this resource to families in need of quality information regarding services and supports for their children with autism.

Patricia Wright, PhD, MPH
National Director, Autism Services, Easter Seals
Autism Spectrum Disorders: What Every Parent Needs to Know is a must-read for every parent with a child on the autism spectrum. How you as a parent can help your child through play, language encouragement, structure, planning and partnering with professionals is outlined at each developmental stage from infancy through adulthood. Chapters on family relationships, your relationship with your pediatrician, and ways that you can become involved as an advocate for both your own child and for systems change, guide and encourage. Stories from multiple families, told in their own words, describe personal experiences and coping strategies that provide hope and comfort throughout the book. Get this book—it’s a complete gem!

Nora Wells, MEd
Director of Programs, Family Voices

Instead of reading like a book, going through the pages of Autism Spectrum Disorders: What Every Parent Needs to Know is like having a casual conversation in your living room with a guru on ASD. The chapters on family, advocacy, and resources are particularly unique and helpful. The information is accurate, comprehensive, well organized, written in a compassionate, reader-friendly style, and interlaced with inspiring stories and endearing humor. Most parents will resonate with some of the stories; many will resonate with all of them. Some stories will bring tears and smiles at the same time. It is a must-have book for parents, service providers, and advocates alike.

Chris Plauché Johnson, MEd, MA, MD, FAAP
Medical Director, Autism Community Network; clinical professor (retired), University of Texas Health Science Center at San Antonio; cochair, American Academy of Pediatrics Autism Subcommittee (2003–2007)
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From the Editors

We dedicate this book to the children and families who inspire, teach, and challenge us daily to seek better answers and solutions.

Dr Rosenblatt thanks his wife, Lisa, and his children, Galit and Shachar, for the blessings and miracles that have led to this moment.

Dr Carbone thanks his wife, Katie, for her love, support, and understanding and also thanks Ben, for being the most wonderful son he could have ever hoped for.

On behalf of the Council on Children With Disabilities Autism Subcommittee, we honor the many contributions of our colleague and friend, Gregory Stephen Liptak, MD, MPH, FAAP (1947–2012), to the field of children with special needs.
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Please Note

The information contained in this book is intended to complement, not substitute for, the advice of your child’s pediatrician. Before starting any medical treatment or program, you should consult with your child’s pediatrician, who can discuss your child’s individual needs and counsel you about symptoms and treatment. If you have questions about how the information in this book applies to your child, speak with your child’s pediatrician.

The names and identifying details in each parent’s story may have been changed to protect the privacy of individuals. The content in these sections is for information only. The experiences may or may not be suitable for each family and do not indicate an exclusive course of treatment. Before starting any medical treatment or program, you should consult with your child’s pediatrician, who can discuss your child’s individual needs and counsel you about symptoms and treatment.

Products mentioned in this book are for informational purposes only. Inclusion in this publication does not constitute or imply a guarantee or an endorsement by the American Academy of Pediatrics.

The information and advice in this book apply equally to children of both sexes (except where noted). To indicate this, we have chosen to alternate between masculine and feminine pronouns throughout the book.
Foreword


Pediatricians are the first to see developmental concerns, so it is fitting that the AAP develop a book on autism spectrum disorders (ASDs) for parents and caregivers. In this book, parents will learn how ASDs are defined and diagnosed, and the types of behavioral and developmental therapies available for treating them. Stories from other parents will help them understand that they are not alone on this journey. This book will help parents understand how ASDs will affect their children as they grow older.

Pediatricians who specialize in ASDs have extensively reviewed this book. Under the direction of our editors, the material in this book was developed with the assistance of numerous reviewers and contributors. Because medical information is constantly changing, every effort has been made to ensure that this book contains the most up-to-date findings. Readers may want to visit the AAP Web site for parents, HealthyChildren.org, to keep current on this and other subjects.

It is the hope of the AAP that this book will become an invaluable resource and reference guide to parents. We are confident that parents and caregivers will find the book extremely valuable. We encourage its use along with the advice and counsel of our readers’ pediatricians, who will provide individual guidance and assistance related to the health of children.

The AAP is an organization of 60,000 primary care pediatricians, pediatric medical subspecialists, and pediatric surgical specialists dedicated to the health, safety, and well-being of infants, children, adolescents, and young adults. *Autism Spectrum Disorders: What Every Parent Needs to Know* is part of ongoing AAP educational efforts to provide parents and caregivers with high-quality information on a broad spectrum of children's health issues.

Errol R. Alden, MD, FAAP
Executive Director/CEO
American Academy of Pediatrics
CHAPTER 1

What Are Autism Spectrum Disorders?
As a pediatrician whose son has an autism spectrum disorder (ASD), I (Dr Carbone) know all too well about the difficult emotions that often surround a diagnosis of an ASD. My son was diagnosed with an ASD in 2004 at the age of 2. Before the diagnosis, we were concerned about his development, beginning in infancy. At times he seemed uncomfortable with symptoms of acid reflux, and at other times he was extremely quiet and hard to engage. While he has always made forward progress, he reached his developmental milestones later than other children. For example, as a young toddler, he had difficulty using gestures, like pointing, to tell us what he wanted, and didn't begin to talk until he was 24 months old.

While getting the diagnosis was painful, it ultimately helped me to better understand him. It also began the process of knowing how to help him reach his potential.

Although his mother and I are pediatricians, we went through a grieving process just like any parents. At first I thought about the things I did with my father that my son and I might not be able to do, like playing sports. I later realized that although there are some things that are challenging for him, there are many things we do together that bring us both much joy. I have learned during this journey that parenting a child with an ASD is not “better” or “worse” than parenting any other child. It is simply different. My son has helped me appreciate and enjoy those differences.

We have always focused on what our son can do and not on what he can't. Along the way, we have tried to obtain the best therapies possible that allow him to reach even higher. As scientists, we knew that the best evidence-based therapy available for children with ASDs was behavioral therapy, so we began his behavioral therapy program while he was very young. In addition to intensive behavioral therapy, we were open to trying complementary and alternative therapies as long as they were safe. After doing some research, we tried a few different nutritional supplements and the gluten-free/casein-free (GFCF) diet, understanding that there was limited evidence that these treatments would help reduce the symptoms of autism. After some time we came to the conclusion that his progress with behavioral therapy was no better with these interventions than without them, and so we discontinued them. We have continued to support him with ongoing behavioral therapy and have been delighted with his progress.
Now our son is an active participant in his community. With the support of family, friends, educators, therapists, and doctors he enjoys many of the same activities of his peers: swimming, basketball, bowling, summer camp, reading, and discovering. All who take the time to get to know him are drawn in by his gentle demeanor, curious nature, and wonderful sense of humor.

Ellen had always taken pride in her son's intelligence, his expansive vocabulary, and his knowledge of dinosaurs. But at 11, Brian was struggling socially. Classmates found his all-consuming obsession with dinosaurs annoying, and Brian grew impatient with them if they didn't know as much as he did about the prehistoric creatures. He had trouble understanding sarcasm from his peers. He couldn't tell when they were being mean but got overly sensitive when they weren't. He thought nothing of making rude, sarcastic comments during class while the teacher was talking.

Brian also behaved in unusual ways. He was always touching people when he was stuck waiting in lines, falling down at unexpected times, and making loud, inappropriate comments about people within earshot. What concerned Ellen the most was that Brian never seemed to look her in the eye while she was talking to him.

Over time, Ellen grew suspicious that something else was going on with her son, especially when she went back to college to get a degree in psychology and started doing more reading. Though he had already been diagnosed with attention-deficit/hyperactivity disorder (ADHD) at age 7, she began to wonder if he also had an ASD, a diagnosis that a teacher had once suggested but that Ellen had always dismissed. "He didn't fit the profile of what I thought was autism," she says. "I always thought children with autism were unattached, unresponsive, and in their own world."

Ellen had Brian evaluated by a psychologist. A screening test suggested that he had high-functioning autism, a form of ASD marked by an obsession with 1 or 2 topics, challenges with the social aspects of language, and difficulties navigating social relationships. The more she learned about high-functioning autism, the more Ellen was convinced that Brian had it. So far, Ellen has been hesitant about getting a formal diagnosis. She fears that the label will
create a lifelong stigma for her son. “And I’m afraid some people will look at me and think I’m a bad parent,” she says. (Ellen, Price, UT)

Chances are, you’re familiar with some of the concerns that Ellen is facing or the difficult emotions that Dr Carbone has experienced while raising a child with an ASD. Like Ellen, you may be wondering whether you should have your child diagnosed or what a diagnosis will mean for your child’s life. Like Dr Carbone, you may be looking for information about where to find help for your child’s language delays, social challenges, and behavioral problems. Or maybe you suspect your child has an ASD but haven’t addressed your concern with your pediatrician yet.

We hope that reading this book will help provide you with the information you are seeking to make the best decisions for your child. In this book, you will learn how ASDs are defined and diagnosed, and the types of behavioral and developmental therapies available for treating them. You will learn when medications may be required, and whether complementary and integrative medicine may be helpful. We will also help you create a treatment team that includes your pediatrician, and provide information to help you care for your child and get a handle on the types of services and assistance available to him. In addition, we will help you understand the effect of ASDs on you and the rest of your family. Stories from other parents will help you understand that you are not alone on this journey. You will acquire an understanding of how ASDs will affect your child as he grows older and the types of advocacy you can do as the most important part of the treatment team: the parent of a child with an ASD.

Autism spectrum disorders are a group of biologically based neurodevelopmental disorders that affect a child’s behavior and social and communication skills. They belong to a group of disorders known as pervasive developmental disorders (PDDs), a distinction that includes ASDs, Asperger syndrome, and pervasive developmental disorder–not otherwise specified (PDD-NOS). These subcategories were used in the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV), a manual published by the American Psychiatric Association to provide diagnostic criteria for behavioral conditions. They will
be eliminated in the new DSM-5, which will be published in 2013. You will read more about the differences later in this chapter.

For most children, these conditions are chronic and require lifelong management. Some children—approximately 3% to 25%, according to studies—improve over time to a point where they no longer meet diagnostic criteria for ASDs. In general, these children are the ones who have typical learning abilities and have received behavioral therapy (see Chapter 4). However, most children who improve still have other developmental and behavioral symptoms.

No doubt, we certainly hear a great deal about ASDs these days. A study by the Centers for Disease Control and Prevention (CDC) estimated that ASDs affect 1 in 88 children, about 1% of all children. Boys are 5 times as likely to be affected as girls, and white children are more often diagnosed than African American and Hispanic children.

A major reason for the dramatic increase in the diagnosis of ASDs has to do with changes in the way the conditions are diagnosed. In 1994, the diagnosis was changed to include children with milder symptoms, including those whose language is closer to normal cognitive milestones. In addition, a growing body of research showing the importance of early, intensive behavioral treatment in helping children with ASDs prompted the federal government to emphasize early detection, so that more children could receive services at a younger age. The emphasis on importance of early diagnosis and intervention inspired several major public education campaigns to teach parents about ASDs and the importance of early diagnosis.

In spite of all the public interest in and attention on autism, figuring out whether your child has an ASD is not easy. These conditions are remarkably complex and difficult to diagnose. No 2 children exhibit the same symptoms, and severity varies widely. Some cases may be subtle, while others may be more straightforward. In most cases, the process of determining

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<th>CAN A CHILD EVER FULLY RECOVER FROM AN AUTISM SPECTRUM DISORDER?</th>
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<td>Symptoms of an autism spectrum disorder vary greatly from one child to the next. While it’s possible for some children—studies range from 3% to 25%—to improve to the point where they no longer meet diagnostic criteria, most continue to have some degree of developmental or behavioral symptoms.</td>
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whether a child has an ASD usually begins with parents who are concerned about their child’s development. But in some cases, the early sign of an ASD first comes to the attention of a pediatrician or child care provider who observes something different in the way the child plays, learns, speaks, or acts.

We’ll discuss more about diagnosis in Chapter 3. First, we’ll go back in time to see how autism emerged as a major health concern.

**A Brief History of Autism**

Autism was first described in 1943 by Dr Leo Kanner, a child psychiatrist at Johns Hopkins University School of Medicine. It was Dr Kanner who first coined the term *autism*, borrowed from the Swiss psychiatrist Eugen Bleuler, who used the word to describe the idiosyncratic, self-centered thinking he saw in schizophrenia. Kanner used *autism* to describe 11 children in his practice who seemed to prefer isolation to social engagement. The children all displayed extreme aloofness and total indifference to other people. They made little eye contact and did not engage in imaginary play. Some displayed an amazing ability for rote memory. Others were obsessed with routines, spinning toys, and mechanical objects. Dr Kanner believed that autism was an inborn disorder and that children with this condition entered the world without biological underpinnings for social interaction. These were children who lived in their own world. Even today, Dr Kanner’s descriptions of autism are highly regarded and considered some of the best ever written.

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<th>FASCINATING FACT</th>
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<td>Donald Triplett, the first child cited in Dr Kanner’s now-famous report on autism, was profiled in <em>The Atlantic</em> in October 2010. At the time, he was 77, living in Forest, MS. Although he faced many challenges throughout his life, he was embraced by his community and enjoyed doing activities such as playing golf. Read his story online at <a href="http://www.theatlantic.com/magazine/archive/2010/10/autism-8217-s-first-child/8227">www.theatlantic.com/magazine/archive/2010/10/autism-8217-s-first-child/8227</a>.</td>
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In the 1950s, Freudian psychoanalysts put a new spin on autism, contending that the condition resulted from the emotional withdrawal of a baby born to a cold and emotionally distant parent. In particular, they focused on mothers and called these parents “refrigerator mothers.” Bruno Bettelheim, then the director of the Orthogenic School in Chicago, became fascinated with children who had autism and advanced this theory. (Bettelheim had a PhD in philosophy but was widely cited as a child psychologist. He lectured on psychology at the University of Chicago, despite the lack of any formal training.) Bettelheim’s most famous patient was a boy named Joey, whom he described in 1959 as a “mechanical boy” in the popular magazine *Scientific American*. At 18 months, Joey was unable to speak and was described by his grandparents as “remote and inaccessible.” Joey became fascinated with mechanical objects and learned to take apart and reassemble an electric fan. By the age of 4, Joey was spending a great deal of his time rocking back and forth and becoming completely consumed with mechanical objects.

Like many of his colleagues at the time, Bettelheim blamed Joey’s unusual behaviors on his parents. Bettelheim claimed that their aloof parenting style forced Joey to withdraw into his own world and marked the beginning of his descent into schizophrenia. In fact, autism was classified as a form of childhood schizophrenia in the first 2 editions of *DSM*.

Bettelheim’s views persisted for years until experts began to consider autism from more biological perspectives. In 1964, a research psychologist named Bernard Rimland described infantile autism as a neurologic disorder with a strong genetic component. Rimland and his wife were personally acquainted with autism—they were the parents of a child with autism, whom they had diagnosed themselves.

Studies in the early 1970s showed that despite similar symptoms, autism was a disorder distinct from childhood schizophrenia. In 1977, the first study of twins and autism was published in the *Journal of Child Psychology and Psychiatry*. The study found a strong genetic influence in identical twins who had autism. If one twin had autism, the other twin was much more likely to have other cognitive differences too. Finding a genetic connection to autism meant that autism needed to be described more precisely so that it could be properly studied and better understood. That became possible in 1980, when *infantile autism* finally received its own separate category in the third edition of *DSM*. 
### EARLY SIGNS OF AUTISM SPECTRUM DISORDERS

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<th>Social Differences</th>
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<tr>
<td>• Resists snuggling when picked up; arches back instead</td>
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<td>• May have temperament differences during infancy, such as being described as a very quiet or very fussy baby</td>
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<td>• Makes little or no eye contact</td>
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<td>• Shows no or less expression in response to parent’s smile or other facial expressions</td>
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<td>• May avoid following a parent’s gaze or finger to see what parent is looking at or pointing to</td>
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<tr>
<td>• No or less pointing to objects or events to get parents to look at them</td>
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<tr>
<td>• Less likely to bring objects to show to parents just to share his interest</td>
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<tr>
<td>• Less likely to show appropriate facial expressions</td>
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<tr>
<td>• Difficulty in recognizing what others might be thinking or feeling by looking at their facial expressions</td>
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<tr>
<td>• Less likely to show concern (empathy) for others</td>
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<td>• Has difficulty in establishing and keeping friendships</td>
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<th>Communication Differences</th>
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<td>• Says no single words by 15 months or 2-word phrases by 24 months</td>
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<td>• May repeat exactly what others say without understanding its meaning (parroting or echolalia)</td>
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<tr>
<td>• Responds to sounds (like a car horn or a cat’s meow) but less likely to respond to name being called</td>
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<td>• May refer to self as “you” and others as “I” (pronoun reversal)</td>
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<td>• Shows no or less interest in communicating</td>
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<td>• Less likely to start or continue a conversation</td>
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<tr>
<td>• Less likely to use toys or other objects to represent people or real life in pretend play</td>
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<tr>
<td>• May have a good rote memory, especially for numbers, songs, TV jingles, or a specific topic</td>
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<td>• May lose language milestones, usually between the ages of 15 and 24 months in some children (regression)</td>
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<th>Behavioral Differences (Stereotypic, Repetitive, and Restricted Patterns)</th>
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<tr>
<td>• May rock, spin, sway, twirl fingers, or flap hands (stereotypic behavior)</td>
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<td>• Likes routines, order, and rituals</td>
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<td>• May be obsessed with a few activities, doing them repeatedly during the day</td>
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<tr>
<td>• More likely to play with parts of toys instead of the whole toy (for example, spinning wheels of a toy truck)</td>
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(continued on next page)
### EARLY SIGNS OF AUTISM SPECTRUM DISORDERS, CONTINUED

**Behavioral Differences (Stereotypic, Repetitive, and Restricted Patterns), continued**

- May have splinter skills, such as the ability to read at an early age but often without understanding what it means
- May not cry if in pain or seem to have any fear
- May be very sensitive or not sensitive at all to smells, sounds, lights, textures, and touch (sensory processing differences)
- May have unusual use of vision or gaze (for example, looks at objects from unusual angles)
- May have unusual or intense but narrow interests

### WHAT AN AUTISM SPECTRUM DISORDER MIGHT LOOK LIKE

It isn’t always easy for parents to know if a child has an autism spectrum disorder (ASD). Some of the symptoms of ASDs may be seen in children with other types of developmental or behavioral problems or, to a lesser extent, in children with typical development. Also, not all of the symptoms are seen in all children. Some children may only display a few of the symptoms. This is what makes the process of diagnosing ASDs difficult. But here are some examples that may help distinguish a child with an ASD from other children.

**At 12 Months**

A child with typical development will turn his head when he hears his name.

A child with an ASD might not turn to look, even after his name is repeated several times, but will respond to other sounds.

**At 18 Months**

A child with delayed speech skills will point, gesture, or use facial expressions to make up for her lack of talking.

A child with an ASD might make no attempt to compensate for delayed speech or might limit speech to parroting what is heard on TV or what she just heard.

**At 24 Months**

A child without an ASD brings a picture to show his mother and shares his joy from it with her.

A child with an ASD might bring her a bottle of bubbles to open but doesn’t look at his mom’s face when he does or share in the pleasure of playing together.
Defining Autism Spectrum Disorders Today

Even now, as we go to press, the definition of ASDs is evolving. To understand how it will change, we need to look at DSM-IV, which was published in 1994, and the soon-to-be published DSM-5, which is in the process of being written. In short, the disorder remains unchanged, but how it is classified and described will be different.

In DSM-IV, ASDs are listed as 1 of 5 PDDs. The other PDDs are Asperger syndrome, PDD-NOS, childhood disintegrative disorder, and Rett syndrome. Here is how these conditions are defined.

Autistic Spectrum Disorder

Autistic spectrum disorder is what most people know as autism. Children who have ASDs have problems relating to others socially. They may have trouble making eye contact, building friendships, and sharing things they enjoy with other people. As infants, they often display limited or no joint attention, a behavior that involves enjoying an object or event with another person by looking back and forth between the two.

Many children with ASDs have delayed language skills or use language in ways that are out of the ordinary. Rather than use language to connect with others, they may use words to meet basic needs, or they may just use their vocabulary for labeling but not to indicate their needs. For instance, a child may have a vocabulary of 20 or more words for labeling objects yet not be able to use those words to ask for an item at an appropriate time. In some cases, children may not develop the ability to use verbal communication.

While social difficulties may appear in the first year, communication problems may not become obvious until the second year. A 2-year-old may lack words to communicate or may not be able to use the words he does have for meaningful interaction, and by age 3, may have no phrases or sentences. Over time, these communication challenges become even more noticeable. Some children may have trouble knowing how to start a conversation. Others may have echolalia, in which they repeat what people say to them. Still others may constantly recite scripts from favorite videos or TV shows.
When children with ASDs play, they rarely use their imagination. They may not act out scenarios or pretend that an object is something else (using a pencil for a laser sword or a banana for a telephone, for instance). When they do play, they may prefer to arrange their toys or play with parts of a toy—opening doors on a car door instead of driving it, for example. Some children form attachments to hard objects such as a ballpoint pen or flashlight instead of stuffed animals.

Children with ASDs may become rigidly fixated on topics that most other people would consider unusual and have trouble letting go of these topics. For instance, they may be interested in movie credits, license plates, or addresses.

Some may become consumed with following rigid routines that may not have any useful purpose or have a strong insistence on sameness. For example, children with ASDs may prefer that certain activities be done in a precise order and may become highly anxious if a routine or ritual is broken. Likewise, they may have difficulty with transitions if they have not been prepared ahead of time.

In addition, children with ASDs may engage in motor activities that appear unusual and to serve no purpose. These may include hand flapping, rocking in place, or walking on tiptoe. Some children become unusually intrigued with parts of objects rather than the objects themselves.

Most of these problems will emerge before the age of 3. According to the current DSM, diagnosing an ASD involves first ruling out 2 other PDDs that we will discuss on page 14, Rett syndrome and childhood disintegrative disorder.

**Asperger Syndrome**

Asperger syndrome is named for Hans Asperger, an Austrian pediatrician. In 1944, Asperger—who did not know about Dr Kanner’s work—published an article describing children whose symptoms were much like those Dr Kanner detailed. But the children Asperger described were typical in their verbal and cognitive skills.

Children with Asperger syndrome share traits with those who have ASDs. Both groups have difficulties with social interactions. They often have trouble looking people in the eye, rarely
use gestures or facial expressions, and have trouble knowing how close to stand to others. They also may have less interest in engaging others and may not share objects or experiences of interest with others.

Like children with ASDs, those with Asperger syndrome may fixate on narrow interests, sometimes to the exclusion of other topics. They may prefer rituals and routines, and may become anxious and upset when those are altered or disrupted. They may also engage in repetitive behaviors such as spinning or rocking for long periods. Children with Asperger syndrome may become preoccupied with parts of objects too.

A key difference, however, is in language skills. Early language milestones are not delayed in children with Asperger syndrome. In fact, these children may even be early readers who speak in an overly formal way and have an impressive vocabulary. Some may affectionately be referred to as “little professors.” Even so, their language skills may be quite unique or different. They may talk continuously about a limited number of topics; have difficulty understanding certain types of humor, figures of speech, and jokes; and have less understanding of the social use of language, such as how to start and maintain a conversation or how to end one.

Another difference is that children with Asperger syndrome do not have cognitive delays, which may or may not occur in autism. The DSM specifies that children with Asperger syndrome are also capable of doing age-appropriate self-help skills like bathing and dressing and will be curious about their environment in childhood.

**Pervasive Developmental Disorder–Not Otherwise Specified**

A child may be diagnosed with PDD-NOS when she has some of the signs and symptoms of ASDs or Asperger syndrome but doesn't meet the strict criteria used to diagnose those conditions. Children with PDD-NOS may have poor social skills because of limited verbal or nonverbal skills, or persistent and repetitive interests, activities, and behaviors. But they differ from children with ASDs or Asperger syndrome in that their symptoms may not appear until they are older.
Some people feel that PDD-NOS is a subthreshold or atypical form of ASD because children who have it show certain symptoms but not others. For instance, a child may have difficulties with social interactions and communication but have no persistent, repetitive behaviors. Symptoms may also be milder. Still, *DSM* specifies that PDD-NOS involves a “severe and pervasive impairment” in developing social skills. This means it can create many of the same challenges as ASDs and Asperger syndrome. This may be especially true if the child also has an intellectual disability that affects her cognitive functioning or other behavioral challenges, such as ADHD.

**Childhood Disintegrative Disorder**

Childhood disintegrative disorder is a rare disorder that affects 2 in 100,000 children. The condition is also known as Heller syndrome. Children with childhood disintegrative disorder develop normally until age 2 or 3, then lose social skills, expressive or receptive language, play skills, motor skills, or bowel and bladder control. They will go on to have difficulties with social interactions or communication and may also develop stereotyped behaviors seen with ASDs. Because childhood disintegrative disorder is so similar to ASD with regression (see box on next page), it has been proposed that it be eliminated from *DSM-5*.

**Rett Syndrome**

Also known as Rett disorder, Rett syndrome is a genetic condition that causes behavioral changes that may initially look like ASDs. An infant with Rett syndrome has typical development and head circumference until about 5 months of age, when growth in head size may begin to slow and early motor skills start to disappear. Most children develop repetitive hand movements like tapping, wringing, and clapping. They also have trouble with gross motor skills and may lose the ability to walk. Rett syndrome is a possible cause of ASDs. It occurs primarily in girls and usually appears before age 2 years. Rett syndrome will likely not be included in *DSM-5* as a subtype of ASDs. See Chapter 2 for a more detailed description of this syndrome.
My son is 20 months old and was developing normally until recently. Someone suggested he might be regressing to an autism spectrum disorder (ASD). What does that mean?

Regression occurs in about 25% of children with ASDs. These children may appear to develop normally, then experience a gradual or sudden loss of social or communication skills. Studies suggest that this is most likely to occur between 18 and 24 months. They may stop talking if they’ve already started using words and stop turning their heads when their names are called. They may withdraw into their own world and appear more distant and less interested in their surroundings. They may become more irritable. For parents, the abrupt change is alarming.

A close look at children’s behavior before diagnosis shows that some of the delays in developmental milestones might have been present before regression, at least to a mild degree. When researchers looked at the home videos of these children at their first birthday parties, they saw subtle signs in some children before regression became obvious and an ASD was detected. The most common sign they noticed was that these children did not consistently turn their heads when their names were called.

The New Definition

In the new DSM-5, due out in 2013, it’s been proposed that a single diagnostic category for ASDs replace the PDDs category. All children who have ASDs, Asperger syndrome, childhood disintegrative disorder, or PDD-NOS will be consolidated under the single diagnosis of ASD, and those other terms will be eliminated. The new DSM-5 will provide a simplified way of defining autism.

To be diagnosed with an ASD in DSM-5, a child must have problems in 2 main areas, social forms of communication and repetitive behaviors. More specifically, these are

- Persistent struggles with social communication and social interactions in various situations that cannot be explained by developmental delays. These may include problems with give-and-take in normal conversations, difficulties making eye contact, a lack of facial expressions, and difficulties adjusting behaviors to fit different social situations.
- Obsessive and repetitive patterns of behavior, interests, or activities. These may include unusual and constant movements, strong attachment to rituals and routines, and fixations on unusual objects and interests. These may also include sensory abnormalities,
which have always been commonly seen in children with ASDs but are not used currently to diagnose an ASD or Asperger syndrome. Children with sensory abnormalities may be hypersensitive to certain sounds, textures, or lights. They may also be unusually insensitive to things in the environment that usually cause pain, heat, or cold.

The new criteria note that symptoms must begin in early childhood and disrupt a child's day-to-day functioning. In addition, diagnosis must take into account an individual's age, stage of development, intellectual abilities, and language level.

If you have any concerns about the diagnosis your child receives or questions about DSM classification or terminology, talk with your child's pediatrician.

**What We Know and Don’t Know**

We're a long way from the days when Bruno Bettelheim blamed parents for a child's autism. We now know that it is a neurodevelopmental disorder, something that occurs in the early formation of the brain. We also know the importance of early diagnosis and treatment, and now have the tools to help us determine whether a child has or is at risk for ASDs. To that end, organizations like the CDC and American Academy of Pediatrics have waged successful public awareness campaigns like “Learn the Signs. Act Early.” (www.cdc.gov/actearly) that have promoted surveillance and screening leading to early diagnosis. This has made early treatment possible for the benefit of millions of children. In addition, we know that certain therapies are more effective than others at treating symptoms.

For example, developmental and behavioral interventions are the mainstay of supporting individuals with ASDs. Behavioral interventions focus on changing specific behaviors and symptoms. As these behaviors change, social relationships and mastery of basic developmental capacities improve. There are several different types of behavioral interventions (see Chapter 4). Studies have consistently shown that children with ASDs who use intensive and systematic behavioral principles to reinforce developmentally appropriate skills have improved social communication, intelligence, language, behavior, and self-help skills when compared with children with ASDs who did not.
Throughout this book we discuss resources and services available today that did not exist just a few years ago and provide strategies on how to access them. People with special needs are more widely accepted in today’s society, and the desire to include them in family and community life has opened doors and allowed them to participate in activities and go to venues that were previously inaccessible. Children born with ASDs today have greater hope for full and rewarding lives than they ever did in previous generations.

Even so, there are still many unknowns. The exact cause of the condition, for instance, has not been determined, although it is now clear that ASD has many different causes, some of which can be identified. In addition, all children with ASDs are unique, so there is no single treatment that works for every child. While scientists do know that the rate of ASD diagnosis is rising rapidly, they have yet to figure out the rate at which ASD itself is increasing. The search for answers has led to unfounded theories and unproven remedies.

Take vaccines. Research shows that almost 20% of parents worry that vaccines are not safe for their children and might lead to health problems like autism or diabetes. In particular, some people worry about the measles-mumps-rubella (MMR) vaccine. Their belief is bolstered by the fact that regression in children later diagnosed with ASDs might occur weeks to months after the MMR vaccine is given. The timing has led many parents to mistakenly assume a cause-and-effect relationship between the MMR vaccine and ASDs. Research has long since shown that vaccines do not cause ASDs, but unfortunately, the concern about an association still persists. (For more detailed information, see Chapter 2.)

Many people also worried about thimerosal, a mercury-based preservative used in vaccines to prevent bacterial contamination. That fear was also proven false. Since 2001, most routine children’s vaccines made in the United States have had no thimerosal except for some flu vaccines. But the rates at which ASDs have been diagnosed have continued to climb.

The search for a cause has also led to theories about abnormalities in the gastrointestinal (GI) tract causing changes in the brain. The belief that ASDs are a GI disorder has given rise to nutritional therapies that have limited scientific backing.

Of particular interest has been the GFCF diet. This diet is based on the belief that gluten (a molecule found in barley, rye, oats, and wheat) and casein (a molecule found in milk
My friend says her sister cured her son of autism with the gluten-free/casein-free (GFCF) diet. Should we try it?

Although the American Academy of Pediatrics does not endorse the GFCF diet, it does understand that some parents may want to consider trying it. Before doing so, it's important to discuss your child's nutritional needs with your pediatrician. For instance, a diet without casein restricts the amount of calcium and vitamin D a child gets and puts her at risk for osteoporosis later in life. Therefore, it's essential to find other ways to meet your child's needs for calcium, vitamin D, and iron. You also have to make sure your child meets her caloric needs. Your pediatrician might refer you to a registered dietitian.

Before going on the GFCF diet, make sure to identify target behaviors and how you will measure changes in those behaviors. By doing so, you will be able to gauge whether the diet is working. Finally, make sure to keep your child's food preferences and routines in mind. Children with ASDs are often resistant to change. And your child may not like the idea of eating foods that are different from what she's used to eating and what the rest of the family eats. You may need to use behavioral strategies to get her to eat new foods. If your child has improvement in irritability or intestinal symptoms but no change in symptoms of ASDs, it may be that she has a common intestinal problem like lactose intolerance.

products) cause substances produced by these foods to enter the bloodstream and travel to the brain, triggering symptoms we see as ASDs. You'll find more information on nutritional therapies and other alternative remedies in Chapter 7.

The Bottom Line

As the parent of a child who has an ASD—or a child whom you suspect has an ASD—you probably want to learn more about ASDs. In this book, we will do our best to provide you with the most up-to-date facts and information about ASDs. We'll also share stories from other parents, who are often the best sources of information as well as emotional support. Our goal is to empower you with the knowledge you need as you embark on this journey with your child. We encourage you to partner with your child's pediatrician, who can help you every step of the way. Armed with the appropriate facts and information, you will be able to make the best possible choices for your child. And that's reassuring for any parent.
Autism Champion: Carmen Pingree

Back in 1979, when Carmen Pingree’s son Brian was diagnosed with autism, 95% of children with autism were institutionalized. The only program for children with autism near their home in Salt Lake City, UT, had 4 students in it, with 12 on a waiting list. And getting a diagnosis typically took years.

Even getting answers for Brian had been a struggle despite all the classic signs—Brian rocked for hours at a time, ignored the affection and attention of his family, and became fascinated with windshield wipers, heights, and lights. He had a penchant for unscrewing objects and undoing latches and locks. Experts labeled him “abused,” “emotionally disturbed,” and “mentally retarded.”

When he was nearly 4 years old, he was finally diagnosed, much to Carmen’s relief. “We finally had a name for what we were dealing with,” she says. “We read books, attended conferences, met other parents, and found a small behavior therapy program with young dedicated professionals. The information lowered our frustration and gave us tools to work with Brian at home until we could find a way to expand the preschool program.”

Together with other parents, Carmen obtained a school building that was being closed. Then she turned her energies toward getting funding from the Utah state legislature. She went back to college to get a master’s degree in special education and political science, a perfect melding of the task that lay ahead. She invited legislators to visit the program, learn about autism, and hear budget requests over lunch.

Her husband and even her other 4 children got involved in the effort, which ultimately succeeded in securing funds for autism services in Utah, including the preschool program, an adolescent teaching home, and a residential treatment program.

Carmen didn’t just lobby. She also began to pursue federal and community funding for autism research and helped create a 5-year joint epidemiologic and genetic study between the University of Utah and the University of California, Los Angeles. Through the years, she became an autism research consultant for Stanford University and Utah State University, the president of the Autism Society of Utah, and a frequent lecturer and researcher whose studies have appeared in numerous scientific journals.
When a new building was constructed in 2003 for the expanded preschool program (which by then included elementary-aged children with autism), it was named The Carmen B. Pingree Center for Children with Autism. An active research site, the center also offers support for parents and siblings of children with ASDs and serves as a training ground for graduate students specializing in ASDs, as well as pediatric and psychiatry residents.

Brian is in his thirties now, but Carmen continues to advocate for families with children with ASDs and serves on the advisory board of the center as well as the board of the program where her son resides. “Brian enjoys his role as uncle to the 20 Pingree grandchildren,” Carmen says. “The whole family continues to involve Brian in their lives and to support programming for individuals with autism. They consider these opportunities for service a great blessing in their lives.”
CHAPTER 2

What Causes Autism Spectrum Disorders?
Chances are, you already knew someone with an autism spectrum disorder (ASD) before you learned or suspected that your child had one. Or maybe you knew someone who worked with children and adults who have ASDs. The number of children being diagnosed with an ASD has been rising steadily in recent decades. In fact, in early 2012, the Centers for Disease Control and Prevention (CDC) reported that the total number of ASD cases in a given population at a specific time had increased 78% in the short span of just 6 years. The CDC found that 1 in 88 children had an ASD in 2008, well above the rate of 1 in 110 that had been found in 2006. That meant that more than 1% of all children in the United States had an ASD.

The report gave credence to a study released in 2009 using the National Survey of Children’s Health, which was conducted by interviewing parents in 2007. That study found that 1 in 91 children had been diagnosed with an ASD. Clearly, ASDs have become a major issue, one that prompted the authors of the CDC report to declare autism “an urgent public health concern.”

The latest increase was not really news, though the extent of it certainly was. The reason for the increase, however, was—and remains—open to debate. Some of it, of course, can be traced to greater public awareness of the disorder and improvements in screening tools that have made parents and health care professionals alike more adept at spotting and identifying children with ASDs. Efforts like the “Learn the Signs. Act Early.” campaign by the CDC and several efforts by the American Academy of Pediatrics have certainly made a difference. Intense media coverage and the publication of a growing body of research in professional journals have fueled that awareness too. Changes in diagnostic criteria that broadened the definition of an ASD also contributed to the surge. Since 1980, when autism first landed in the third edition of the *Diagnostic and Statistics Manual of Mental Disorders* as its own separate entity, criteria have been expanded to include milder cases of the condition such as Asperger syndrome and pervasive development disorder—not otherwise specified.

Legislative maneuvers had an effect too, namely passage of the Individuals with Disabilities Education Act (IDEA) in 1990. Before IDEA was enacted, children might have received educational classifications such as *intellectual disability, learning disability, speech or language impairment*, or *emotional disturbance* before they could be eligible for special education services. Once autism was recognized as an educational classification by IDEA, the numbers of
children labeled with these other categories went down, while the number of children under the autism classification went up.

That same year, Congress passed the Americans with Disabilities Act, which required states to administer their programs for children with disabilities in the most integrated settings possible. This federal legislation led to closure of some institutions that once housed children with disabilities and instead encouraged local governments to support families who were raising their children at home. As a result, children with autism, especially those who also had other behavior problems and intellectual disability, began to attend public schools. Instead of being hidden away in institutions, these children were now being counted among those in special education programs.

But some experts have concluded that these factors account for only some of the increase. Changes in how an ASD is diagnosed, for instance, could explain only 25% of the rise in autism cases in California between 1992 and 2005, according to a study from Columbia University. That meant that 1 in 4 children diagnosed with an ASD today would not have been given that diagnosis in 1993. On the other hand, a recent study from England estimated the rate of ASDs in adults to be similar to that of children. This would seem to indicate that the true risk for developing an ASD is not increased for today’s children. So while some recent events explain why an ASD diagnosis is made more often, there still is vigorous debate whether the true risk of a child having an ASD is increasing.

As part of this discussion, it’s important to appreciate the difference between an increased risk for an ASD (such as older age of parents) and what causes the condition (ie, certain medical and genetic syndromes). A risk factor, such as parental age, does not directly cause an ASD but makes it more likely for the child to have an ASD compared with those without the risk factor. The increased risk for an ASD means that parental age is probably one factor among many that may contribute to the child's diagnosis but by itself is not the cause. In congenital rubella syndrome (see page 33), an infection during pregnancy with the rubella virus causes the ASD, whereas in fragile X syndrome (FXS) (see page 28), the cause of the ASD is a particular sequence of DNA on the X chromosome.
Challenges of Autism Spectrum Disorders

Truth is, we are just beginning to learn about what causes an ASD. In about 15% of children with an ASD, the condition is associated with a clear underlying cause such as a chromosome abnormality, a genetic syndrome, or a known environmental cause. In most children, however, the underlying cause of the ASD is not obvious.

What we do know about ASDs is that they are a group of biologically based neurodevelopmental disorders with a strong genetic component. In short, an ASD is the result of something that occurs in the development of the brain. Exactly what triggers the event (or events) in the brain is still not fully understood. Scientists are fairly certain that autism is the result of complex interaction between genetic risk and environmental exposure, but they do not yet know exactly how much is genetic and how much is environmental. It’s also quite possible that there are several genetic factors being influenced by the environment. But these environmental risk factors are still poorly understood and have not been sufficiently identified yet.

One thing scientists are certain about: ASDs come in many forms and have multiple causes. Understanding the causes of ASDs has been a major research challenge that as of now still eludes us.

Genetics of Autism Spectrum Disorders

To understand hereditary influences on autism, it helps to have some basic knowledge of genetics. Genetics is the study of heredity, the passing on of cellular instructions from parents to their offspring, which determine traits of the offspring such as hair color, eye color, and height. All our body cells have 46 chromosomes—2 sex chromosomes and 22 pairs of non-sex or autosomal chromosomes. We inherit half of our chromosomes from each of our parents. Chromosomes are made up of DNA, which is shaped like a double helix or spiral ladder. Genes are the units of DNA that code instructions for making proteins that enable each cell to do what it’s assigned to do. In other words, genes provide body cells with instruction manuals on what they need to do to make our bodies function. Changes in DNA can affect how genes work. A change in an individual gene, called a mutation, can lead to certain diseases and disorders.
### BRAIN ABNORMALITIES IN AUTISM SPECTRUM DISORDERS

Children with autism spectrum disorders (ASDs) have distinct differences in their brain that are not seen in children without ASDs. And while these differences don’t explain the cause of ASDs, they do explain some of the social differences, language issues, and motor skill difficulties that children with ASDs often display.

Among the key differences are

- Larger than normal head size, also known as macrocephaly.
- Greater brain volume.
- Fewer number of Purkinje cells in the cerebellum. Purkinje cells are large neurons in the brain responsible for coordinating motor skills. Recent studies have shown they are also involved in language, attention, and mental imagery.
- Abnormal maturation of the forebrain limbic system. The limbic system is made up of several brain structures, including the hippocampus, amygdala, thalamic nuclei, and limbic cortex. These structures are associated with emotion, behavior, long-term memory, and smell.
- Abnormalities in the frontal and temporal lobes. The frontal lobes are involved in various aspects of higher cognitive functioning, control of emotions and behavioral impulses, as well as the ability to transform thoughts into words. The temporal lobes play an important role in auditory perception and processing, visual processing, and the formation of long-term memory. Sensations of touch and taste also seem to be integrated with memory in the temporal lobes.
- Brainstem abnormalities. The brainstem connects the brain to the spinal cord. It carries sensory signals from the body to the brain and motor signals from the brain back to the rest of the body. The brainstem is also involved in regulating cardiac and respiratory functions, the sleep cycle, and consciousness.
- Neocortical malformations. The neocortex is the thin outer layer of the brain involved in higher functions such as sensory perception, generation of motor commands, spatial reasoning, conscious thought, and language. Disruptions to normal formation of this region of the brain are also associated with intellectual disability and seizures.

Scientists know that many of these brain structures are formed during the first 2 trimesters of pregnancy. As a result, experts suspect that environmental influences that may contribute to the development of ASDs are most likely those that occur to the mother early in her pregnancy. Furthermore, newer brain imaging techniques suggest that there are atypical connections between different parts of the brain of individuals with ASDs. Future research will help us to understand the nature of these abnormal connections and how they affect the function of the brain in a manner that leads to symptoms of an ASD.
Scientists have known for decades that genetics play an important role in the development of ASDs. Identical twins, who share the same genes, are significantly more likely to both have an ASD than fraternal twins, who share fewer genes. According to a large study published in 2011, a younger sibling of a child with an ASD has approximately 20 times the risk of developing an ASD as a child who does not have an affected sibling. And gender plays an important role too, as boys are more likely to have an ASD than girls. In the 2011 study, approximately 1 in 4 younger male siblings developed an ASD, whereas 1 in 11 younger female siblings were diagnosed with an ASD by the age of 3 years.

Experts know too that there are many genetic abnormalities involved in ASDs, not just a single abnormality (as there is in, say, sickle cell anemia); none of these are in all children with an ASD. Some children with an ASD have genetic mutations—permanent changes in DNA—that they did not inherit from their parents. Some children have changes in the structure of their chromosomes. Other children with ASDs have deletions and duplications of genetic material. These genetic abnormalities—rare genetic mutations, chromosomal abnormalities, and deletions and duplications of genetic material—account for at least 10% of all cases of ASD. Down syndrome, for example, is caused by having an extra copy of chromosome 21, and children with this condition have a very high prevalence—almost 10%—of autism. But each genetic abnormality by itself is rare and occurs in no more than 1% to 2% of all ASD cases.

Because each of the many genetic factors that may increase the risk for an ASD account for such a small percentage of all cases, scientists suspect that there are other factors that have yet to be discovered at play. It’s quite possible that ASDs, as with other conditions, are the result of a variety of interactions between genes and factors in the environment. Ongoing research will continue to identify genetic and environmental factors involved in the underlying causes of ASDs.
Genetic Disorders and Autism Spectrum Disorders

Some people diagnosed with an ASD have known genetic mutations that result in a specific genetic disorder. Not every child with one of these disorders has an ASD, but having one of these genetic disorders certainly raises a child’s risk for developing ASD. A partial list of the many genetic disorders associated with ASDs follows.

Fragile X Syndrome

Fragile X syndrome is a genetic disorder that affects the X chromosome passed down on the mother’s side. It is the most common known genetic cause of ASDs and intellectual disability in boys. Because boys have only one X chromosome, they are affected more severely than girls. Boys with FXS may have distinctive physical features that may include having an unusually large head, ears, and testicles (after puberty). Children with FXS may also have weak muscles and loose joints. Fragile X syndrome is the most common cause of inherited cognitive impairment, which can range from learning disabilities to more severe intellectual disabilities. As many as 30% to 50% of people with FXS will have some characteristics of ASDs.

If your child has an ASD and intellectual disability or global developmental delays—meaning your child isn’t reaching all the developmental milestones that are expected at different ages—or there is a history of intellectual disability on the mother’s side of the family, testing for FXS should be done.

Rett Syndrome

Rett syndrome occurs primarily in girls and usually appears sometime in the first 2 years of life after a period of normal development. Girls with Rett syndrome typically lose control of hand skills and develop hand-writhing movements. They also develop difficulties walking, slowed head growth, seizures, and trouble with social skills.

Thanks to DNA sequencing, Rett syndrome can be diagnosed in more than 90% of cases.
Tuberous Sclerosis Complex

Children who have tuberous sclerosis complex have lesions in the brain, skin, and other organs. The condition is often associated with recurrent seizures. The link to ASDs in tuberous sclerosis complex is high—as many as 25% of children with this condition have an ASD.

Angelman Syndrome

Angelman syndrome affects 1 in 15,000 children. Children who have this disorder have intellectual disability, an unsteady gait, atypical laughter, seizures, and distinctive facial features. The condition is often overlooked as a cause of ASDs, intellectual disability, or cerebral palsy. Certain genetic tests can identify more than 80% of cases.

Phenylketonuria

Universal newborn screening has almost eliminated this once common cause of intellectual disability and ASDs. Phenylketonuria is a metabolic disorder that affects the body’s ability to process a specific chemical called phenylalanine, which is found in many foods. When the chemical builds up in the body, it can become toxic to the developing brain. Restricting foods that contain phenylalanine in early infancy can prevent disabilities.

Smith-Lemli-Opitz Syndrome

Smith-Lemli-Opitz syndrome (SLOS) is a rare disorder caused by a defect in the way cholesterol is manufactured. It affects 1 in 20,000 children. Children with this condition often have multiple congenital malformations, such as cleft palate, extra toes, and genital deformities. However, some children with mild SLOS have an ASD and mild webbing of their toes. They may experience failure to thrive and feeding problems. Most children with this condition are on the autism spectrum.

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<th>GENETIC TESTS</th>
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<td>New genetic tests called microarray allow geneticists to see extremely tiny changes in DNA. Over time these tests are becoming more sensitive and one day may give us a better understanding of how changes in DNA cause autism spectrum disorders.</td>
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Could Epigenetics Have the Answer?

Epigenetics merges nature and nurture. It is the study of how the environment changes the way genes work. Epigenetic changes occur on the surface of genes and may be caused by exposure to environmental factors in the foods you eat and stress you experience. Scientists believe it’s possible that these epigenetic changes are passed on from one generation to the next.

Most often, these epigenetic changes result from methyl groups, chemical entities that latch on to DNA and then silence or activate the gene. Some experts believe that epigenetics may eventually help researchers pinpoint the potential environmental causes of autism, as well as many other diseases (for example, cancer, Alzheimer, obesity), and lead to the development of new therapies for treatment. Epigenetic factors are already known to cause certain genetic disorders such as Angelman syndrome (see page 29).

Among recent findings using epigenetics is a study of oxytocin. Experts have long suspected that people with ASDs do not have as much oxytocin (the bonding hormone responsible for social behavior such as recognizing loved ones, building trust in others, and alleviating anxiety) as those without ASDs. Recent research from scientists at Duke University has shown that the problem may actually lie with oxytocin receptors, which allow the hormone to bind to neurons and take effect. The researchers found that people with an ASD have more methyl groups on the oxytocin receptor gene than people without an ASD. When these methyl groups become part of the genetic code, it is believed that the gene is “turned off,” which would explain some of the social difficulties in people with ASDs. Controlling methylation of these genes may possibly lead to new treatments, but more research is still needed before that can occur.

Environmental Causes

No one knows exactly what it is in our environment that may lead to an ASD, but there have been no shortage of theories as to a probable cause. Some people, for instance, point to vaccines, especially if their child displayed no signs of ASDs until after he received the vaccine, despite evidence to the contrary that has shown no link.
Three main theories that have been raised about vaccines and autism are

1. The measles-mumps-rubella (MMR) vaccine causes autism by damaging the lining of the intestines.

2. Thimerosal, a mercury-containing preservative in some vaccines, causes autism by damaging the nervous system.

3. Receiving too many vaccines at once leads to autism by affecting the immune system.

At this writing, there have been 29 studies since 1998 that have looked at the MMR vaccine and ASDs, and all have come to the same conclusion: there is no evidence that the MMR vaccine causes autism. For example, one study compared all children in Denmark (535,544) who got the MMR vaccine with children who didn't get the vaccine and found that the risk for ASDs was no different for children who received the MMR vaccine than for those children who did not.

Thimerosal is an organic mercury-containing antibacterial compound that was in US vaccines until 1999. There have been 11 studies since 1998 that have looked at thimerosal and ASDs, and all have come to the same conclusion: there is no evidence that children who received vaccines with thimerosal have a greater risk of autism than those who didn't. For example, a recent study comparing 200 children with ASDs with 700 children without ASDs showed that there were no differences in the amount of thimerosal that each group received from vaccines. Another study showed that groups of school-aged children exposed to smaller or larger amounts of thimerosal performed similarly on a series of psychologic tests. In addition, the rate that ASDs are diagnosed has continued to rise even though thimerosal has been removed from almost all vaccines for more than 10 years.

These days, a common concern is that getting a number of vaccines at the same time might somehow weaken the immune system, triggering the development of ASDs. Because of this belief, some have suggested that parents should space out vaccines by using an alternative vaccine schedule. There are a number of reasons why this is not recommended. First, receiving multiple vaccines at the same time is safe. Today's more refined vaccines even when given together cause less stress to the immune system than vaccines of the past. This means that even though children receive more vaccines at the same time, the challenge to the immune
system is less than in years past. Secondly, there is evidence that delaying vaccines does not make a difference in development. A recent study comparing school-aged children who received their vaccines on time with those who did not found that timely vaccination was associated with better performance on numerous tests of language and intelligence. Less-vaccinated children did not do better on any of the tests. For parents who are concerned that children receive too many vaccines too soon or who believe delaying immunizations is beneficial, this study provides reassurance that timely vaccination during infancy has no adverse effect on long-term neuropsychologic outcomes. Lastly, it’s important to understand that the vaccination schedule created by the CDC was designed to protect children when they are most vulnerable to disease. Delaying vaccines or not vaccinating your child puts him at risk for vaccine-preventable diseases. Even those who are fully vaccinated increase their chances of contracting a vaccine-preventable disease if they live in a community where many people are under-immunized. One study found that for every 1% increase in proportion of school-aged children who were under-immunized, the risk of pertussis infection among fully vaccinated children doubled.

Skipping or delaying vaccines also increases the risk for other children in your neighborhood. For example, many communities in the United States are currently experiencing outbreaks of measles because there are too many unvaccinated children. And unvaccinated children help the virus move more easily from person to person. If you have concerns about vaccines, be sure to talk with your child’s pediatrician.

As of this writing, more than 40 studies since 1998 have been published in peer-reviewed literature, and all have come to the same conclusion: vaccines do not cause autism. Approximately 1% of the population has the condition, yet researchers all over the globe have examined the records of hundreds of thousands of children and could not find any evidence of a relationship between vaccines and autism. With such a large number of children studied, if there were a link between ASDs and vaccines, it would have shown up in one of these studies. For a listing of studies, visit www.HealthyChildren.org/vaccinestudies.

What scientists do know is that ASDs may often result from complex interactions between environmental exposures and a person’s genes. So while you may be born with a genetic predisposition for ASDs, it requires an environmental exposure or event—which may even occur in the womb—for that gene to be expressed.
Some experts suspect that chemicals in our environment are involved in causing ASDs. Toxins like inorganic mercury have been touted as likely suspects, as have many other heavy metals, pesticides, and substances in plastics. The human-created class of chemicals known as polychlorinated biphenyls is also suspected as a culprit. But so far, there has not been enough research to prove these suspicions.

Experts do know that exposure to certain drugs during pregnancy can contribute to the development of ASDs. Two drugs in particular deserve mention. One of them is valproate, an anticonvulsant used to treat bipolar disorder and epilepsy. The other is thalidomide, a drug now used to treat multiple myeloma, a type of bone marrow and blood cancer. Thalidomide had been used to treat morning sickness in pregnant women in the late 1950s and early 1960s but was subsequently banned for causing birth defects. These medications are believed to affect the development of the fetus’ brain in the early trimesters of pregnancy, a time when developmental abnormalities in the brain are most likely to occur.

Infections are often cited as a probable culprit too. But so far, only one infection—rubella—has been associated with autism. Rubella, also known as German measles, causes a rash, fever, and muscle and joint pain. Congenital rubella develops in a fetus when a mother is exposed to the rubella virus early in her pregnancy. Babies born with this disorder often have many birth defects in addition to severe developmental delays. They may also have symptoms of ASDs. In recent years, the disorder has become less of a concern since the rubella vaccine was introduced, which is ironically a component of the controversial MMR vaccine.

Recent studies suggest that babies exposed to large amounts of alcohol in the womb can develop ASDs as well as a spectrum of other neurodevelopmental disorders. These babies have fetal alcohol syndrome or alcohol-related neurodevelopmental disorder. They often have growth deficiencies that lead to short stature, small head circumference, decreased muscle tone, facial abnormalities, cardiac defects, and delayed development. Children with fetal alcohol syndrome may be also diagnosed with ASDs, though the link between autism and prenatal exposure to alcohol requires further research.
Family Health and History

The health of mom and dad always plays a major role in a child's health, so it's no surprise that experts have probed for links between family health and ASDs. In particular, scientists have zeroed in on the mother's history of autoimmune diseases, illnesses in which the body's immune system attacks itself. In particular, research has shown that children whose mothers have rheumatoid arthritis or celiac disease are more likely to have ASDs. Rheumatoid arthritis is a disease in which the body attacks the lining of the joints, causing inflammation and pain. In celiac disease, the body cannot tolerate gluten, a protein in wheat, rye, and barley. When someone with celiac disease eats gluten, the immune system launches an attack on the villi, small protrusions that line the small intestine.

The risk for an ASD also goes up if there is a family history of type 1 diabetes, another autoimmune illness. In people with type 1 diabetes, the immune system attacks cells in the pancreas and destroys its ability to make insulin, a hormone that's essential for turning the foods we eat, particularly carbohydrates, into energy. So while autism is not an autoimmune disorder, it's possible that the genes involved in causing autoimmune disease may also be playing a role in the development of ASDs. In women with rheumatoid arthritis, there is evidence that they may produce antibodies during pregnancy that affect fetal brain development.

Another factor in early pregnancy that appears to increase the risk for ASDs is advanced maternal and paternal age. Studies have shown that older parents were more likely to have a child with an ASD than younger parents. One study found that for mothers aged 20 to 39 years, every 10-year increase in their age raised the risk of them having a child with an ASD by 38%.

No one knows exactly why older women and men are more likely to have children with an ASD, but experts suspect that a woman's hormonal changes at an older age could affect fetal brain development. In addition, they suspect that the use of reproductive technologies may play a role. Simply being older causes more age-related changes in a woman's genes and adds to the cumulative effects of exposures to environmental toxins, both of which may affect the fetus. Older men have a great number of spontaneous genetic mutations in their sperm as they age, which may contribute to higher risk for an ASD. But more research is needed to know exactly how advanced parental age affects risk for ASDs.
The amount of time between pregnancies—also known as the inter-pregnancy interval—is also a risk factor. One study that examined birth records of second-born children found that those who were conceived within 12 months of the birth of their older sibling were more than 3 times more likely to be diagnosed with ASDs. Children conceived 12 to 23 months after an older sibling were almost 2 times more likely to have been diagnosed with ASDs.
Ongoing Research

Solving the mystery of what causes ASDs has spawned intense research efforts around the world. Some of the major research projects now underway are:

- The Centers for Autism and Developmental Disabilities Research and Epidemiology is being directed by the CDC in California, Colorado, Maryland, North Carolina, Pennsylvania, and Georgia. Each center is working on aspects of a project called Study to Explore Early Development and looking for causes and risk factors of ASDs. In California, for instance, the focus is on identifying biological traits that will help with early identification of children with ASDs and in investigating potential environmental risk factors. For more information, see www.cdc.gov/ncbddd/autism/caddre.html.

- Early Autism Risk Longitudinal Investigation (EARLI) is following more than 1,200 mothers from pregnancy through the first 3 years of their babies’ lives to examine potential environmental risk factors that may be involved in ASDs. The EARLI study will look at the DNA profiles of all family members and test the hypothesis that ASDs have a genetic and an epigenetic basis. Research is being done at multiple sites including Johns Hopkins, Drexel University, Children's Hospital of Philadelphia/University of Pennsylvania, and Kaiser Permanente in northern California. For more information, see www.earlistudy.org.

- The Centers for Children's Environmental Health and Disease Prevention (CCEHDP), which is supported by the National Institute of Environmental Health Sciences and Environmental Protection Agency, is examining the effect of exposure to chemicals such as lead, mercury, and pesticides on neurodevelopmental disorders such as ASDs, attention-deficit/hyperactivity disorder, and developmental delays. The research is being done at several sites including the University of California, Davis (UC Davis), and the University of Medicine & Dentistry of New Jersey. For more information, see www.niehs.nih.gov/research/supported/centers/prevention.
At UC Davis, the CCEHDP has established the first large epidemiologic study of ASDs called Childhood Autism Risks from Genetics and the Environment. This study, which is also being conducted at the University of California, Los Angeles, will involve up to 2,000 children with ASDs and other developmental delays or intellectual disabilities as well as children with typical or expected development. It will examine various aspects of their lives before and after birth, such as environmental exposures, medical history, and diet, as well as physiologic factors that may affect brain development such as specific genes, lipids like cholesterol, and molecules involved in the immune and nervous systems. The study will involve children born in California who are between the ages of 24 and 54 months.

The National Children's Study is examining the effects of genetics and the environment—including air, water, and diet, as well as cultural and community influences and family dynamics—on the health of children across the country, from birth to age 21 years. The study also involves pregnant women and women who may become pregnant. Findings will be used to help improve the health of children. For more information, see www.nationalchildrensstudy.gov.

Understanding the causes of ASDs is critical to improving prognosis for children diagnosed with this disorder. It will also pave the way to more effective treatments. Ultimately, knowing the cause of ASDs may even help us find ways to prevent this baffling disorder.

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**I have a daughter who has an autism spectrum disorder (ASD) and a son who does not. Could my son carry a gene for ASDs that he’ll pass on to his offspring?**

Experts know that if one child in the family has an ASD, that child’s siblings are at greater risk for developing ASDs too. If your son is healthy, the odds that he will pass on a gene for ASDs is low. However, his odds may be slightly higher than someone else who does not have a sibling with an ASD. The genetics underlying ASDs are complex, and it may be helpful for parents to seek counseling from a geneticist or genetic counselor to better understand the risk of recurrence.
Autism Champion: Alison Singer

Alison Singer was a successful television executive at NBC when her daughter Jodie was diagnosed with autism in 2000. The diagnosis changed Alison's life forever. She left television and became acting CEO of Autism Speaks when it was launched in 2005.

In 2009, she founded her own organization, the Autism Science Foundation. The group raises money to support ASD research, brings together parents and scientists to share information, and trains scientists to work with the media. It also brings the latest science to people at the forefront of ASD—parents and educators.

Getting the science out is Alison's way of combating the myths that continue to linger about vaccines and the unproven remedies that are often touted as treatments. Even now, Alison gets frequent e-mails offering an alleged cure. “These quacks prey on the desperation of parents who are willing to try anything they think might possibly help their child,” she says. “Believe me, I get it. We love our children so much and want them to improve and so we are willing to try anything. That's why it's so important to do the science to test interventions. We need to know what works and what doesn't. These are not quick fixes; they take time and lots of effort, but they really do help.”

Jodie is now 14 years old. Time has taught Alison that with the right therapies, children with ASDs can make significant improvement. “The day they're diagnosed is the bottom,” says Alison, who also has a younger daughter who does not have an ASD. “It's the worst day. From then on, the kids will continue to gain skills. They will make gains that astonish you, even if it's sometimes 2 steps forward and 1 step back. Those steps back are frustrating, but I have learned to appreciate the steps forward more than I ever imagined was possible.”

Her advice to parents? Talk to other parents in your community. “Services are always delivered locally,” she says. “Your best source of information will often be other parents in your school district.”
CHAPTER 3

How Do I Know if My Child Has an Autism Spectrum Disorder?
Carly was concerned. At 17 months, her son Asher wasn’t talking yet, and when she called his name, he would rarely respond. At the 18-month visit, Asher’s pediatrician listened to her concerns and performed an autism screening test. He informed her that he also shared her concerns about Asher’s behavior and development. Asher was referred to a specialty clinic and subsequently diagnosed with an autism spectrum disorder (ASD). While the diagnosis was a shock to his family, they were ultimately thankful to have their concerns validated. The diagnostic evaluation also allowed them to learn more about Asher and how to help him. Today, at the age of 7, Asher still faces challenges with social skills and communication but is making steady progress, much to the delight of his family.

Getting a diagnosis for a child with an ASD often isn’t easy. Unlike some conditions, such as diabetes or celiac disease, ASDs aren’t diagnosed with a blood test. There are currently no x-rays or scans that can detect ASDs. Instead, diagnosis is made based on caregivers’ description of the child’s development and by careful observations of characteristic behaviors by providers who have expertise with ASDs. In some cases, the path to a diagnosis begins with something as simple as a parent’s hunch or a sense that something isn’t quite right.

Diagnosing an ASD is difficult for many reasons. For one, every single case is different. While children on the autism spectrum share similar characteristics, exactly how those traits play out will vary from one child to the next. The severity of autism varies considerably too. For example, some people with ASDs have very mild forms, display virtually no speech problems, and are capable of independently meeting their needs as adults. Some may even be considered gifted and exceptionally bright. Others have severe forms of the disorder, with significant disability, and may have a lifelong dependence on others to meet their needs. Still others have genetic disorders or medically complex conditions requiring medical stabilization before a child’s developmental status can be accurately determined.

One thing experts do know now is that early diagnosis and treatment of an ASD is very important in determining how well a child lives with it. Herein lies the challenge. While most signs of ASDs are apparent by the time a child is 3 years old, many children are not diagnosed until they are older.
Age of diagnosis may depend on where children fall on the spectrum as well as other socio-economic factors. One study found that the average age of diagnosis in children with ASDs was 3.1 years, while those with pervasive development disorder—not otherwise specified (PDD-NOS) was 3.9 years. Among children with Asperger syndrome, the average age of diagnosis was 7.2 years. The study also uncovered some possible explanations as to why some children are diagnosed sooner and others are diagnosed later. Children were typically diagnosed later if they lived in a rural setting, came from households with lower incomes, and had consulted 4 or more different primary care physicians. Symptoms make a difference too; children who had severe language delays were diagnosed an average of 1.2 years earlier than those who did not. Those who demonstrated hand flapping, toe walking, and unusual play over a period were diagnosed at a younger age, while those who were oversensitive to pain or had a hearing impairment tended to be diagnosed later.

Children with ASDs who have intellectual disabilities (or global developmental delays) are usually diagnosed earlier than those who do not. Those who have regressive autism, in which signs of ASDs appear after a period of seemingly normal development, are also more likely to be diagnosed early. Boys are generally diagnosed at a younger age than girls. It’s possible that certain traits in girls with ASDs, such as shyness, may be more socially acceptable and therefore more easily overlooked.

Although diagnosing a child at a young age is important for getting the early intervention that is so critical to children with ASDs, a national study has shown that the median age of diagnosis was 6 years, and more than a quarter of children were not diagnosed until age 8. The good news is that the age of earliest diagnosed cases is dropping. According to the Centers for Disease Control and Prevention (CDC), the earliest cases were diagnosed between 49 and 66 months in 2002; by 2006, earliest cases were diagnosed between 41 and 60 months of age; and by 2008, the age of earliest case identification was between 36 and 59 months.

Early diagnosis requires a partnership between parents and pediatricians. Within this partnership you, as the parent, should feel comfortable bringing up any concerns you have about your child’s behavior or development—the way she plays, learns, speaks, and acts. Likewise, your child’s pediatrician’s role in the partnership is to listen and act on your concerns. During
your child’s visits, the pediatrician may ask specific questions or complete a questionnaire about your child’s development. Pediatricians take these steps because they understand the value of early diagnosis and intervention and know where to refer you if concerns are identified. This chapter will help you recognize the early signs of ASDs so that you can better partner with your child’s pediatrician to get your child the help she needs. The importance of this partnership cannot be stressed enough.

**Begin Early in Infancy**

Like adults, all babies are unique. Some start babbling early on, while others are late talkers. Some start crawling at a young age; others seem to take longer to start moving about. Even within families, parents often marvel at how differently their children grow and develop. But experts are increasingly convinced that early signs of ASDs are evident even during the first few years of life.

While it may be challenging to diagnose an ASD in a child younger than 2 years, it is important for you as a parent to monitor your child’s development carefully so that you can identify any concerns as soon as possible. For instance, by the end of their third month, most babies have started to smile and show pleasure in playing with others and are gradually becoming more communicative with their expressions and body movement. They’re usually able to raise their head and chest when they’re on their tummies and stretch out their legs and kick. Place their tiny feet on a firm surface and they will push down. Most can open and shut their hands and bring their hand to their mouth. They usually can reach for dangling objects and are starting to take hold of toys.

Meanwhile, they may be watching you intently and following moving objects. They can often recognize familiar objects and people from a distance. They may smile at the sound of your voice and turn their heads in the direction of sound. Some may be cooing (making vowel sounds) and imitating the sounds you make.

In babies who may have developmental disabilities such as ASDs, some of these milestones may be delayed or absent. Babies with ASDs may only rarely respond to loud noises, smile at others, or reach for objects. Some may seldom take note of their hands or follow moving
objects. They may only occasionally vocalize, pay attention to new faces, or support their head well.

Of course, even perfectly healthy babies may not achieve these milestones by the end of 3 months of age either. Some babies simply develop a little more slowly. If your baby does not meet all of these milestones on time, it does not necessarily mean she has an ASD or another developmental disability. While ASDs and other developmental disabilities are not typically diagnosed during infancy, children who are late achieving developmental milestones benefit from treatment. If your child does display signs of a developmental delay, you can contact an early intervention or Part C services program, which is geared to help infants and toddlers from birth through age 3 who may be at risk for a developmental disorder. We’ll discuss those in greater detail in Chapter 5. Most important of all, watching for these signs will make you aware of a potential problem, so you can bring it to your pediatrician’s attention. If these delays persist or new ones develop, you and your child’s pediatrician can intervene at an earlier age to help your child reach her full potential.

What to Look for in Autism Spectrum Disorders

When your child has a cold, you expect a runny nose, some coughing, and perhaps a low-grade fever. When your child has eczema, you know his skin will itch and develop a rash. But when your child has an ASD, it’s a lot harder to know what to expect, especially given how different the condition reveals itself in each child. But there are some major characteristics that are common to most children with ASDs.

Unusual Language Development

Language development varies widely among children with ASDs. Some are early talkers and never seem to run out of things to say. Others are naturally quiet and start speaking much later. Speech typically begins with producing vowel sounds, or cooing, in the first few months of life. By 6 months of age babies can combine consonants and vowels, called babbling, making simple sounds like “da” or “ba.” Babbling gradually evolves as your baby starts to link these sounds (“da da”) and introduce new ones such as “pa.”
Between 4 and 6 months the typically developing child will display a back-and-forth pattern of speaking that alternates between cooing or babbling and silence. For instance, babies often vocalize to themselves when they first wake up, only to fall silent when mom enters the room, as if waiting to hear what she has to say. When mom leaves to retrieve diapers, they may start vocalizing again. When babies vocalize in this manner, it is possible for caregivers to sustain a “conversation” with them in which turn-taking occurs, adults speaking in regular sentences and babies cooing and babbling. These back-and-forth vocalizations, together with eye contact and shared emotions (elements of nonverbal or body language), set the stage for later conversations using real words. Over time, a baby’s sounds become more distinct and start to sound like words. Eventually, the typical child will begin to form short sentences.

Most parents are eager to hear their children utter their first words. A child’s first utterances often inspire awe and excitement. So it’s not surprising that when these events do not occur, parents are apt to take notice and bring it up with their child’s pediatrician. Language delays are often the first signs noticed by parents and doctors that raise concern that a child may have an ASD. They’re often the first indication to a pediatrician that a child needs evaluation.

Language differences characterize all forms of ASDs, to varying degrees. In some children with ASDs, language skills may be absent or delayed. Other children, like those diagnosed with Asperger syndrome, may possess advanced speaking skills but struggle having a back-and-forth conversation because they have the need to speak only about a preferred topic. This type of language challenge common to children with Asperger syndrome reflects qualitative differences in language development that distinguish their language skills from those of typically developing peers. Specifically, these differences are in the area of pragmatic language—using language for social communication.

Pragmatic language involves skills such as picking up on body language, maintaining eye contact, understanding implied meaning, using normal voice inflection and volume when speaking, maintaining the topic of conversation, and recognizing the interest level of others in what is being discussed. Such differences may not be obvious until preschool when interacting with peers. Whereas many children with ASDs have language delays, such as those with autistic disorder and PDD-NOS, all children on the autism spectrum have challenges with pragmatic language.
Most language delays are evident by the time a child is 18 months old. It is most apparent if you notice that your child is not showing the desire to communicate or express himself with gestures such as pointing. Children who have milder forms of autism will usually develop speech, but their language may be odd and lack purpose. For example, they may say words that seem to have no intent and that may be taken from television programs or movies. The early speech patterns of children with ASDs may have some distinctly unusual patterns.

Echolalia

Echolalia is the repetition of another person’s speech. It may be immediate, meaning the child will repeat what he hears right after he hears it, or delayed, meaning the repeated phrase will pop up hours, days, or even weeks later.

Keep in mind, though, that echolalia can occur in children who do not have ASDs too. The difference, however, is that in children who do not have ASDs, echolalia tends to be of the immediate kind and then completely disappears from the child’s vocabulary. In children who have ASDs, echolalia may last throughout their lives. And the degree of echolalia significantly affects their ability to communicate effectively with others.

They also tend to display a mix of immediate and delayed echolalia and are more likely to repeat larger chunks of material. For example, rather than repeat the slogan of a television commercial, they may recite the entire commercial and do so for long periods, even while others are trying to communicate with them.

At first, echolalia may create the impression that a child with an ASD is verbally gifted. His vocabulary, grammar, and syntax may make him sound sophisticated for his age. Some kids may even display remarkable skills at labeling colors, shapes, letters, and numbers. But in the child with an ASD, the voice may be delivered in a monotone fashion or other type of peculiar intonation. A closer listen often reveals delayed or absent receptive language, which means difficulty understanding what is spoken to him. Most children with typical development are able to follow simple one-step commands by the time they are 12 to 15 months old. If you ask a child with an ASD to get a toy, he is less likely to respond. If you ask him to identify a familiar object, such as a sippy cup or shoe, from amongst several items, he is often unable.
Pop-up Words
Some children with ASDs will say a word without any provocation and seemingly no reason. Delivery is entirely spontaneous and often inconsistent. For instance, a child may be playing with a ball when he starts saying “Dog,” when there is no dog—real, stuffed, or in pictures—nearby, nor has a dog been recently seen. In some children, the pop-up word will be spoken during times of stress, such as the child in the dentist’s office who says “Bye-bye” when the dentist attempts to place him in the procedure chair. Pop-up words can last for days or weeks and then disappear.

Giant Words
Children with ASDs sometimes say phrases that link together several words, such as “What is it? I don’t know.” These phrases are spoken without true meaning, and the children are unable to combine words into sentences that have any real meaning.

Some children appear to master all the language skills appropriate for their age, only to have them diminish between 15 and 24 months of age, often at 18 to 21 months. These children may have regressive autism. When a child has regression, he may lose verbal skills as well as communicative gestures. The loss may be sudden or gradual. For many parents, the loss of language skills is often a red flag that something is amiss.

For more information about how your infant and young toddler should be communicating with you, see “Milestones During the First 2 Years” on page 48.

Social Skill Deficits
Human beings are hardwired for socializing. We want to share our lives with other people, so we gather for meals, throw parties, and meet for coffee. The drive to be social starts in infancy when babies gaze adoringly at their parents, coo at the sound of their voices, and later point at objects they want them to see. In children with ASDs, that desire for connectedness is diminished or absent. Children with ASDs may be content to be left alone and are less likely to seek out others for interaction. The lack of social reciprocity may seem to emerge in toddlerhood, but experts now know that more basic social skill deficits can be apparent even
Chapter 3 • How Do I Know if My Child Has an Autism Spectrum Disorder?

MILESTONES DURING THE FIRST 2 YEARS

Long before your baby utters her first word, she has already started communicating with you, using smiles, looks, movements, and sounds. Children develop at different rates, but they usually are able to do certain things at certain ages. Following are general developmental milestones. Keep in mind that they are only guidelines. If you have any questions about your baby’s development, ask your child’s pediatrician—the sooner the better. Even when there are delays, early intervention can make a significant difference.

By 1 year, most babies will

• Look for and be able to find where a sound is coming from.
• Respond to their name most of the time when you call it.
• Wave goodbye.
• Look where you point when you say, “Look at the ________.”
• Babble with intonation (voice rises and falls as if they are speaking in sentences).
• Take turns “talking” with you—listen and pay attention to you when you speak and then resume babbling when you stop.
• Say “da-da” to dad and “ma-ma” to mom.
• Say at least 1 word.
• Point to items they want that are out of reach or make sounds while pointing.

Between 1 and 2 years, most toddlers will

• Follow simple commands, first when the adult speaks and gestures, and then later with words alone.
• Get objects from another room when asked.
• Point to a few body parts when asked.
• Point to interesting objects or events to get you to look at them too.
• Bring things to you to show you.
• Point to objects so you will name them.
• Name a few common objects and pictures when asked.
• Enjoy pretending (for example, pretend cooking). They will use gestures and words with you or with a favorite stuffed animal or doll.
• Learn about 1 new word per week between 1½ and 2 years.
earlier. Crucial building blocks of more advanced social skills include joint attention, social orienting, and pretend play.

Joint Attention

A toddler does not look at Elmo on the television despite his father’s pointing and saying, “Look!” A young child finishes a drawing of his mother but does not bring it over to show her. A school-aged child rarely shares what happens at school with his parents despite their repeated requests. These children with ASDs all demonstrate deficits in joint attention. Joint attention is engaging another’s attention to objects, events, or other persons simply for the enjoyment of sharing an experience. Like all developmental milestones, it is mastered in steps that occur at predictable ages throughout childhood.

Joint attention starts early when a typically developing baby recognizes a parent or familiar caregiver’s voice, smiles, and reacts with happy smiles of his own. At about 8 months of age, the baby will follow your gaze when it shifts away to see what you are looking at. Sometime between 10 and 12 months of age, when you point in the direction of an interesting object or event and say, “Look!” your baby will respond by turning his head to see what’s intriguing you. Your baby will then turn his gaze back to you to affirm that he saw what you were indicating.

In children who have ASDs, this type of experience sharing may not develop at the same rate. Babies with ASDs are less likely to look in your direction or show interest in engaging

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<th>MILESTONES DURING THE FIRST 2 YEARS, CONTINUED</th>
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<td><strong>By 2 years of age, most toddlers will</strong></td>
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<tr>
<td>• Point to many body parts and common objects.</td>
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<td>• Point to some pictures in books.</td>
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<tr>
<td>• Follow 1-step commands without a gesture like “Put your cup on the table.”</td>
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<td>• Be able to say about 50 to 100 words.</td>
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<tr>
<td>• Say several 2-word sentences and phrases like “Daddy go,” “Doll mine,” and “All gone.”</td>
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<tr>
<td>• Be understood by others (or by adults) about half of the time.</td>
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earlier. Crucial building blocks of more advanced social skills include joint attention, social orienting, and pretend play.
you. Saying the baby’s name loudly or touching him on the shoulder may not get his attention. Even if it does, a child with an ASD is not as likely to look back at you to share in what you’ve both just looked at.

The difficulty with social engagement continues into toddlerhood. At about 12 months, the typically developing child will begin trying to assert himself socially by asking for an object that is out of reach. He may do this by making simple sounds like “uh” or pointing with an index finger. This is called imperative pointing, or pointing to request. By about 14 to 16 months of age, the child with typical development will point at objects he likes simply to comment to you that it is pleasing to him and to share that experience with you. This is called declarative pointing, or pointing to show. He will alternate between looking at you and at the object or event that has captured his interest. This is the full expression of joint attention. Consistently demonstrating joint attention, experts say, reliably predicts whether a child will develop functional language within a year.

Delayed or lack of joint attention is one of the most specific early signs of ASD. Compared to children with typical development, children with ASDs are less likely to point to or comment about objects or events. When they do point, they may show little enthusiasm and make little effort to connect with you while pointing. Although some children with ASDs may point to shapes, objects, and colors they have learned, it is more often to label than to share an experience. Despite these challenges, numerous studies have shown that with early intervention, children with ASDs can improve their joint attention.

**Social Orienting**

The first time a baby turns her head at the sound of her name is an exciting moment for most parents. Developmentally, it’s also an important social skill milestone, known as social orienting. This milestone is usually achieved by the time a child is 8 to 10 months old.

Children who have ASDs may not acknowledge a caregiver’s bid for their attention. It may take calling a child’s name louder and louder to finally get her attention. Failure to respond to their name is one of the most common early signs of ASD in young children but is sometimes overlooked. While it is important to consider the possibility of a hearing problem,
most children with ASDs hear and respond to environmental sounds (for example, the doorbell) but seem to respond less to the human voice. With time and intervention, most children with ASDs will improve their ability to respond to their name.

**Pretend Play Skills and Friendships**

As you might imagine, you will notice differences in the way children with ASDs play with toys and with other children. Typically developing children will begin playing by grasping objects, a skill that develops around 4 months of age. They may start mouthing objects, and at 8 to 10 months of age may start banging toys together or against the floor or table, or tossing them around. This stage of play is known as the sensory-motor stage. Around 1 year, they become more aware of how toys are meant to be handled and may start playing with them more appropriately. For instance, instead of banging blocks on the floor, they may now stack the blocks. Soon after, pretend play emerges, and they may use toy bottles to feed a baby doll or a toy telephone to chat with grandma. Pretend play gradually becomes more sophisticated, and simple objects may be used to represent other, more complex ones. Bananas, for instance, may become telephones, and wooden blocks may be used as cars.

Compared with typical children, children with ASDs engage in significantly less pretend play before the age of 2. They may have very little interest in toys, preferring instead to play with everyday objects like string, pens, and rocks. If they do develop an interest in toys, they may tend to play with parts of the toy and not the whole toy itself. So rather than push a toy truck along the floor, a boy with an ASD might pick it up and focus on spinning the wheels or opening and closing the doors. Children with ASDs who have normal nonverbal intelligence may be especially skilled at putting things together, such as stacking cups and assembling puzzles, and may later on become masterful at computer games, or what is called constructive play. Some children with ASDs may also insist on repeatedly lining up objects, which is known as ritualistic play. These types of play do not involve imitation, observation, or other people and are better suited for children who do better at play that involves trial and error problem-solving skills.

What can be deceiving is that some children with ASDs enjoy roughhousing. They may like it when dad tosses them in the air or tickles them on the floor. Many children who have ASDs
enjoy the sensory-motor aspects of this type of play. Though it may appear to be “typical” behavior, it is often the sensory aspect of this kind of play that a child with an ASD prefers as opposed to the social engagement. In other words, they are seeking out the sensory stimulation that occurs when they are tossed or tickled, not necessarily the companionship of the other person.

To the unsuspecting parent, a child with an ASD may appear easy to manage because he is content to play by himself for hours without seeking out mom or dad. But a closer look at the child’s style of play will reveal that the play is sensory-motor, constructive, and ritualistic, and does not involve other people. Later on in life, the child with an ASD often struggles to interact with peers and cooperate in groups with social rules. As a result, children with ASDs are often the victims of bullying and left out of most social circles.

Repetitive and Unusual Behaviors
Children who have ASDs may display different behaviors and peculiar mannerisms. They may flap their hands, rock their bodies, or twirl their fingers, especially when they become excited. Some children walk on their toes, nod their heads, or sniff and lick nonfood items. These behaviors are called stereotypies, repetitive behaviors that outwardly serve no apparent purpose and yet are performed compulsively. Stereotypies are generally harmless but can, in some cases, interfere with the child doing something else or prevent the child from learning a new skill. Stereotypies may not be obvious until after the age of 2.

Although stereotypies are common in children with ASDs, they don’t just occur with autism. Children who have intellectual disabilities or global developmental delays may also demonstrate stereotypies. Even young children with typical development may sometimes flap their hands when they’re excited or go through periods of walking on their toes.

Restricted Interests
Most children don’t escape childhood without developing a strong bond to a beloved teddy bear, special blanket, or treasured doll. Children with ASDs, on the other hand, may not, preferring instead to latch on to a hard object such as a pen, a flashlight, or an action figure.
The attachment to that object is also more persistent, and they may insist more intensely on holding the object at all times.

Children who have Asperger syndrome may be less consumed with objects and more enamored with topics and facts. But the fierce interest in these topics is often stronger than it is in children with typical development. In some cases, the child with Asperger syndrome may have an encyclopedic storehouse of facts and information about the topic. Topics of interest are not necessarily unusual for small children. For instance, Ellen’s son Brian developed a strong interest in dinosaurs, which was no surprise because his father is a paleo-artist who has several paleontologists as friends. Besides, many young children are fascinated with dinosaurs. But Brian’s interest in dinosaurs has been all-consuming. He is often more than willing to discuss dinosaurs with others, to the exclusion of other subjects, even when his classmates show no interest. The intensity of his interest is common in children who have Asperger syndrome.

Other Common Features of Autism Spectrum Disorders

Language differences and deficits in social skills are the most prominent and defining characteristics of children with ASDs. But many children also have other difficulties.

Cognitive Challenges

Although cognitive deficits aren’t considered a core feature of ASDs, they are common among children who have these disorders. At one time, experts estimated that intellectual disability—the term varies depending on age and different assessment tools—applied to 90% of all children with an ASD. The latest data from the CDC indicate that these problems affect about 38% of children with ASDs. As you may recall, children who have Asperger syndrome do not have cognitive disabilities and have normal, or even above-normal, intellectual skills.

What many children with ASDs do have is unevenness in their skills and development. A child with an ASD may, for instance, be an exceptional math whiz but may struggle to read. Some children may also have incredible focus, memory, and mathematic skills, while others display notable musical and artistic talents. In rare instances, the child with an ASD
may have highly developed skills and talents that earn him the label of savant. A savant—as performed by Dustin Hoffman in the movie *Rain Man*—is a person with exceptional skills in a narrow area. For example, some savants may be capable of doing rapid calculations, memorizing large amounts of information, and mastering complex pieces of music with little practice. Savant abilities are somewhat rare (ranging from less than 1% up to 10%) in children and adults with ASDs.

### Sensory-Motor Symptoms

For children with ASDs, the sounds, sights, and textures that we experience on a daily basis can often be a challenging minefield to navigate. Some are hypersensitive, or overly bothered, by things in their environment. Others are hyposensitive and completely insensitive to sensations that others consider bothersome. But a child’s sensitivity to sights and sounds may not be consistent across the senses. For example, loud noises at a party may put a child with an ASD on edge, even though she’s totally oblivious to the sound of her mother’s voice calling her name. A child with an ASD might excessively inspect toys or other objects by gazing at them for a particularly long time or from different angles, while remaining uninterested in the rest of her surroundings.

Some children have tactile defensiveness, in which they’re overly sensitive to certain textures and surfaces, like the elastic in socks or labels in shirts. Some may resist hugs because they don’t like to be touched. They may also have oral aversions to certain textures in food. Children with ASDs may also show unusual sensory-seeking behaviors, such as a tendency to walk on their toes (even though they have full range of motion at their ankles), flap their hands, spin, rock back and forth, jump, or chew on objects.

Some children with ASDs have unusual motor skills. Some may appear to have advanced fine motor skills such as stringing beads, but most have trouble with gross motor skills like running, climbing, and jumping. Many also have trouble with coordination and motor planning, which involve thinking through a task and then doing the movements in the proper sequence. Children with Asperger syndrome, in particular, may be clumsy. Some children may appear hyperactive and show symptoms of attention-deficit/hyperactivity disorder (ADHD). Others may be withdrawn and hypoactive, and make little movement.
Common Health Problems

Children with ASDs often have other health and psychiatric conditions. These associated conditions may have profound effects on them. They can often affect children’s behavior, their ability to learn, and their overall health and well-being. Treating these conditions may help a child’s overall functioning, which is why talking to your child’s pediatrician about them is critical. In fact, a child may have one or several of these problems. Medications to help control some of these conditions will be discussed in Chapter 6. Here are some common problems in children with ASDs.

Seizures and Epilepsy

Children with ASDs are more likely than children with typical development to experience a seizure—sudden and excessive electrical discharges in the brain that can produce a variety of symptoms from unconsciousness and contractions of the muscles to undirected, uncontrolled, and unorganized movements. Seizures are more common in children with ASDs who have global developmental delay, intellectual disability, severe motor deficits, and a family history of epilepsy. During a seizure, a child may make jerky movements with his limbs, lose consciousness, or stare off into space. Seizures in children with ASDs are most common when the child is younger than 5 years and again during adolescence. Children with ASDs who are suspected to have seizures may require additional tests, including an electroencephalogram (EEG) or an imaging test of the brain, to confirm seizures and look for potential causes.

Gastrointestinal Disorders

Children with ASDs may be more likely to have gastrointestinal (GI) issues than typically developing children. Many children with ASDs experience chronic constipation, diarrhea, vomiting, and abdominal pain. Most GI disorders in children with or without ASDs are functional, meaning that there is not a specific cause within the GI tract that can be identified. This is especially true for some GI disorders such as constipation, which may be the result of a child’s selective eating habits and pickiness about food. Some GI disorders have an organic cause, meaning that there is a specific problem in the GI tract causing symptoms. This is true
for GI disorders such as celiac disease, an autoimmune condition triggered by gluten and related proteins. It is especially important to tell your child’s pediatrician about any weight loss, GI bleeding, prolonged or persistent vomiting, prolonged diarrhea, abdominal pain that is only in a small area of the abdomen, or fever because these may be symptoms of a more serious organic GI disorder.

Children with GI issues and ASDs may have difficulty informing their caregivers that they have abdominal pain. Instead, you may notice behaviors such as frequent clearing of the throat, screaming, whining, groaning, and sobbing for no apparent reason. Some children may display delayed echolalia and repeat a phrase they’ve heard in the past about their stomach or pain, such as, “Does your tummy hurt?”

Other children may grimace, grit their teeth, or wince. Some may mouth their clothing, lean their abdomen against furniture, or tap their fingers on their throat. Some children may eat, drink, and swallow more. Unusual postures such as the arching of the back, self-injurious behaviors, or an increase in repetitive behaviors may also be signs of GI distress.

Abdominal pain or discomfort can result in changes in a child’s overall well-being too. You may notice your child becomes irritable or may develop sleep problems. It is important to tell your child’s pediatrician about these symptoms or any of the other nonverbal behaviors common with GI conditions. After listening to you and examining your child, the pediatrician might choose to try a medicine to treat the most likely GI disorder (like constipation or gastroesophageal reflux) or do further testing for some of these conditions.

While ongoing research is exploring whether some children with ASDs have unique problems within the GI tract, the current way to treat GI disorders in children with ASDs is the same as for children without ASDs. This is because it is assumed that the problems within the GI tract are the same for both sets of children. There is no evidence at this point that children with ASDs have unique microscopic abnormalities in their intestines or overgrowth of yeast or other organisms that worsen behavior.

As of this printing, there also is no evidence that GI problems directly cause ASDs. One such theory was put forward in the 1990s. It claimed that changes in the GI tract (a “leaky gut”) caused by the measles-mumps-rubella vaccine given to 1-year-olds actually caused
Autism Spectrum Disorders: What Every Parent Needs to Know

ASDs. This study was later found to be significantly flawed and was retracted from the medical literature. Despite many theories of a GI basis for autism, there hasn’t been any proof of a specific link between a disordered GI system and symptoms of ASDs. (See Chapter 7 for more information on this issue.)

**Tics**
Some children with ASDs have tics—brief, involuntary movements and sounds that are also the defining symptoms of a neurologic condition called Tourette syndrome. The 2 conditions have a lot in common, including echolalia, obsessive-compulsive behaviors, and abnormal motor behaviors. There is some evidence to suggest that some of the same brain abnormalities in ASDs also exist in Tourette syndrome. In moderate to severe cases, medical treatment can be quite helpful.

**Sleep Disorders**
Studies show that between 40% and 80% of children with an ASD experience sleep problems. They may have trouble falling asleep, staying asleep, or waking up early. Severe sleep problems may affect a child’s quality of life, worsen his ability to pay attention, and cause him to be irritable and display more repetitive behaviors. Likewise, caregivers of children with sleep problems will likely have sleep interruptions as well, adding to a family’s overall stress level. Some children with ASDs appear to need less sleep than their typical peers. It is important to discuss sleep problems with your child’s pediatrician because they may be caused by other medical conditions (such as gastroesophageal reflux) that cause pain and lead to night awakenings. (See Chapter 6.)

**Attention-deficit/hyperactivity Disorder**
Many children with ASDs have difficulty staying on task and focusing and may be impulsive and hyperactive. Some children on the autism spectrum wind up being diagnosed with ADHD as well. Attention-deficit/hyperactivity disorder is a biological, brain-based condition that, left untreated, can lead to difficulties in school, low self-esteem, and problems
making friends. The condition is quite common and affects an estimated 6% to 9% of all school-aged children.

Children with ADHD have trouble filtering out irrelevant information. They struggle with prioritizing, organizing, and delaying gratification. In children who have ASDs, however, inattention may be related to self-directed thoughts or activities, such as the persistent repetition of a word, gesture, or act, rather than to minor distractions in the environment.

**Aggression and Self-injury**

Many children with ASDs have difficulty moderating the intensity of their emotions and controlling their impulses. Combined with the frustration of not being able to easily communicate their wants and needs, children with ASDs may exhibit aggressive behaviors and self-injury. On the other hand, a painful ear infection may cause a child to bang her head against the wall. Acting aggressively may also stem from stomach cramps caused by constipation. Aggressive behavior may be caused by an underlying psychiatric condition such as anxiety. With so many different causes, if your child is becoming aggressive toward others or herself, you should talk to your child’s pediatrician. Often it will be necessary for a number of professionals to work together to find out the cause of the aggression. Once the cause is known, there are many potential therapies that help.

**Anxiety Disorders**

Children with ASDs are prone to anxiety, which may show up as anything from feelings of nervousness to hyperactivity and other inappropriate behaviors such as screaming or aggressive acts. Because many children with ASDs are extremely rigid in their routines, unexpected changes can lead to an increase in anxiety and inappropriate behaviors. Anxiety may be more common in children whose families have a history of this condition. Children with ASDs who have challenges communicating may become anxious if they do not know how to respond or cope appropriately.

Children with ASDs who have anxiety can sometimes become obsessive in their behaviors. Many children become extremely rigid in their routines. They may want to move through their mornings in the exact same order every day and insist, for example, that their stuffed
animals be laid out in the same precise arrangement every day. When those rituals and routines are disrupted, they may have trouble adapting or have more intense or prolonged tantrums when caregivers try to transition them from one activity to another.

**Depression**

Children who have ASDs are more vulnerable to depression, a mood disorder that in children with typical development may lead to sadness, inactivity, and lack of interest in favorite activities. It may be more challenging to recognize depression in children with ASDs and other developmental disabilities. When considering depression, it may help to compare your child’s current state to how she “typically” acts, paying particular attention to crying spells, enjoyment of activities, interest in being around others, sleep patterns, appetite, and energy level. In children with ASDs and depression, the intensity, frequency, and duration of behaviors such as aggression and irritability may increase from typical levels. Often, there is a family history of depression.

**I Have Concerns; What Should I Do?**

The best thing you can do if you think your child might have an ASD is to bring up your concerns with your child’s pediatrician. By listening to your concerns and observing your child, your pediatrician can work with you to decide on the next step. If your pediatrician shares your concerns and recommends a more complete ASD evaluation, the process will help you learn more about how you can help your child reach his full potential. While getting an ASD diagnosis may be difficult for you and your family, receiving the diagnosis at a young age means you can start early with intervention therapies that will, in the long run, be the best for your child.

Carly, for instance, knew for months that something wasn’t right with her son Asher. She was devastated to get the diagnosis but immediately had help from an early intervention therapist who had been in the room when Asher was diagnosed. Carly took a couple of weeks to let the diagnosis sink in and to start figuring out what she needed to do. In the meantime, the therapist registered Asher for early intervention services, which Asher attended a few times before starting at a school for children with special needs 2 months later.
During every one of your child’s health supervision visits, your pediatrician may ask about any concerns you may have about your child’s behavior or development. Be sure to take these opportunities to talk about any concerns that you or other caregivers may have. Also, inform your pediatrician about any other family members who have ASDs or symptoms of ASDs. Your pediatrician will carefully observe your child and perform an examination. The frequent visits you have with your child’s pediatrician will allow for a complete view of your child’s overall development.

At your child’s 9-, 18-, and 24- or 30-month visits, your pediatrician may ask you to fill out a screening questionnaire about your child’s development. Some of these questionnaires will ask about all aspects of your child’s development. Others may ask about signs of ASDs. It’s important to know that these tools assist your pediatrician in identifying children at risk for developmental disabilities but are not used to diagnose any specific condition. If your child is found to be at risk, he will be referred for a comprehensive evaluation. It is during this evaluation that a specific developmental disorder may be diagnosed.

A comprehensive evaluation for ASDs may involve assessments by several professionals who ideally work as a team. Team members might include your child’s pediatrician, a developmental pediatrician, a psychologist, a psychiatrist, a neurologist, a speech-language

<table>
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<th>WORDS TO KNOW</th>
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<tr>
<td><strong>Developmental surveillance:</strong> the process your pediatrician uses to identify children who may be at risk for developmental disorders such as autism spectrum disorder (ASD). This involves listening to your concerns about your child’s development and behavior, making careful observations of your child during visits, and asking about other family members with developmental disabilities.</td>
</tr>
<tr>
<td><strong>Developmental screening:</strong> a process your pediatrician uses that involves parental questionnaires (standardized tools) about your child’s behavior and development to further clarify if a child is at risk for a developmental disability.</td>
</tr>
<tr>
<td><strong>Comprehensive evaluation:</strong> A multistep assessment of children who, through surveillance and screening, are found to be at risk for an ASD. It involves questioning caregivers, observing the child, performing a physical examination, and administering any tests that may assist in arriving at a specific diagnosis. Ideally, this is done by a team of professionals.</td>
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A case study in the October 2010 issue of the Journal of Developmental and Behavioral Pediatrics recounts the story of Jacob, a 22-month-old boy with no family history of autism. But his parents’ answers to 3 questions on a screening test raised concerns. They revealed to their pediatrician that Jacob did not pretend play, such as talking on the phone or taking care of a doll; did not respond to his name when they called; and sometimes stared at nothing or wandered for no purpose. On a different screening test of general development, the parents expressed concerns about Jacob’s limited speech. At almost 2 years of age, Jacob spoke only 2 words in Hebrew and 1 in English. Given the results on these 2 screening instruments, Jacob was referred for a diagnostic evaluation to look for developmental problems, autism being just one of them.

Regardless of exactly who is involved, your child’s evaluation should include a health history, a physical examination, careful observation, and a hearing test. In addition, other team members might do more formal evaluations of your child’s language and cognition as well as administer other ASD-specific tests. Still other tests may be recommended if it seems that your child’s autism is associated with a medical condition such as those listed in Chapter 2. (Table 3-2 on page 63 lists screening tools that pediatricians may use to help refer children for ASDs.)

Even with so many experts and diagnostic tools available, accurately diagnosing a child with an ASD remains a challenge. Because there is not yet a clear biological marker that can be detected in the blood or seen on digital imaging to identify children with ASDs, a lot of factors may complicate an accurate diagnosis. Some of the criteria used to diagnose ASDs are not easily applied to very young children, especially those younger than 2 years. Also, it is not uncommon for families to receive different diagnoses from different evaluators. In addition, it is difficult in some parts of the country to have access to a team of health care professionals with the skills and expertise to diagnose ASDs.
TABLE 3-1. INTERDISCIPLINARY ASSESSMENT TEAM FOR CHILDREN WITH AUTISM SPECTRUM DISORDERS

<table>
<thead>
<tr>
<th>Team Member</th>
<th>Role</th>
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<tbody>
<tr>
<td>Audiologist</td>
<td>Evaluates for hearing loss as etiology for developmental delay</td>
</tr>
<tr>
<td>Developmental pediatricist, child neurologist, physician</td>
<td>Performs medical evaluation</td>
</tr>
<tr>
<td></td>
<td>Identifies and treats associated conditions</td>
</tr>
<tr>
<td>Geneticist and genetic counselor</td>
<td>Performs evaluation when an underlying medical condition or genetic syndrome is suggested by family history, examination, or clinical course</td>
</tr>
<tr>
<td></td>
<td>Counsels family on recurrence risk</td>
</tr>
<tr>
<td>Psychiatrist</td>
<td>Evaluates and treats associated psychiatric conditions and maladaptive behaviors</td>
</tr>
<tr>
<td>Psychologist</td>
<td>Administers cognitive or developmental testing</td>
</tr>
<tr>
<td></td>
<td>Administers diagnostic tools</td>
</tr>
<tr>
<td></td>
<td>Identifies associated psychiatric conditions and develops behavioral treatment plan</td>
</tr>
<tr>
<td>Occupational therapist</td>
<td>Evaluates for fine and gross motor deficits</td>
</tr>
<tr>
<td></td>
<td>Evaluates for sensory processing deficits</td>
</tr>
<tr>
<td></td>
<td>Develops plan for treatment</td>
</tr>
<tr>
<td>Social worker</td>
<td>Identifies family needs</td>
</tr>
<tr>
<td></td>
<td>Refers family to formal and informal support agencies and organizations</td>
</tr>
<tr>
<td>Speech-language pathologist</td>
<td>Evaluates for expressive, receptive, and pragmatic language deficits</td>
</tr>
<tr>
<td></td>
<td>Develops plan for treatment</td>
</tr>
</tbody>
</table>

Note: To facilitate recollection of developmental milestones and behavior, parents should review baby books, records, and video recordings of their child’s early years before attending a diagnostic evaluation.

When the Diagnosis Is Autism Spectrum Disorder

It can be difficult to learn that your child has a lifelong developmental disability. Naturally, you as a parent, other caregivers, and extended family need to grieve about this. You will undoubtedly worry about what the future holds. Keep in mind during these difficult times that most children with ASDs will make significant progress in overall function. Some children with ASDs can do exceptionally well and may even remain in a regular education classroom. Many will have meaningful relationships with family and peers and achieve a good level of independence as adults.

It is important to remember that while an ASD diagnosis may change what you thought your parenting experience would be, we now know that children with ASDs and other developmental disabilities can achieve so much more in life as long as they are given appropriate support and opportunities. Even parents like Carly, who was initially devastated to learn her son had autism, realize now that getting a diagnosis will help them better understand

**TABLE 3-2. SELECTED AUTISM SPECTRUM DISORDER SCREENING QUESTIONNAIRES BY AGE**

<table>
<thead>
<tr>
<th>Screening Tool</th>
<th>Ages</th>
</tr>
</thead>
<tbody>
<tr>
<td>Communication and Symbolic Behavior Scales Developmental Profile Infant-Toddler Checklist (CSBS-DP-ITC)</td>
<td>6–24 months</td>
</tr>
<tr>
<td>Modified Checklist for Autism in Toddlers (M-CHAT)</td>
<td>16–48 months</td>
</tr>
<tr>
<td>Social Communication Questionnaire (SCQ)</td>
<td>For child 4 years or older (who has developmental skills greater than or equal to 2-year-old)</td>
</tr>
<tr>
<td>Childhood Asperger Syndrome Test (CAST)</td>
<td>4–11 years old</td>
</tr>
<tr>
<td>Krug Asperger's Disorder Index (KADI)</td>
<td>6–21 years old</td>
</tr>
<tr>
<td>Autism Spectrum Screening Questionnaire (ASSQ)</td>
<td>7–16 years old</td>
</tr>
<tr>
<td>Autism Spectrum Quotient (AQ)—Adolescent Version</td>
<td>11–16 years old</td>
</tr>
</tbody>
</table>
their children and allow them to move forward with finding the right services for them. In the coming chapters we will describe how you can help your child access the support and opportunities that will allow her to reach her full potential.

Until recently, our 20-month-old son was always chatty and seemed to be on his way to saying some words. But my husband and I have noticed lately that he isn’t speaking as much or doing as much pointing or gesturing as he did just weeks ago. We recently moved to a new house and my husband started a new job with different hours, so he sees less of our son. Could the changes in environment be affecting our son’s communication skills? We’re worried.

It’s tempting to blame the slowdown in your son’s language skills on the move or not seeing as much of his father. But if your child is experiencing noticeable changes in his ability to communicate, you need to bring this to your pediatrician’s attention. Your son is at an age when setbacks in language skills may be a sign of autism. Approximately 25% to 30% of children with ASDs appear to be developing normally and then lose some or all of their language and social skills. Discuss your concerns with your child’s pediatrician. Getting prompt attention, even without a definitive diagnosis, will allow you to learn how to help your son and gain access to early intervention, which will help him reach his full potential.
**Autism Champion: Catherine Lord, PhD**

Catherine Lord, PhD, was an undergraduate when she took a psychology class at the University of California, Los Angeles, with Dr O. Ivar Lovaas, the psychologist who helped develop the applied behavior analysis therapy for ASDs. “It was just at the time when he was taking on the challenge of autism as a way to test a theory that operant conditioning could teach anyone anything,” Dr Lord recalls.

Dr Lord participated in a project involving teaching children with ASDs to speak. “I worked with 2 children who were so different from each other who also had amazing similarities,” she says. “I think that is what captivated me originally. I was also fascinated by the links that people with autism make between ideas and the things they see, even when they cannot easily communicate about them.”

Today, Dr Lord is the director of the Center for Autism and the Developing Brain at New York-Presbyterian Hospital, Weill Cornell Medical College, and Columbia University Medical Center. She is credited with devising the Autism Diagnostic Observation Schedule (ADOS), a standardized assessment of communication, social interaction, and play for diagnosing individuals with ASDs. She is also a coauthor of the Autism Diagnostic Interview, which was recently revised (ADI-R), for clinicians to use in interviews with caregivers about a child’s early development, communication, social interaction, and patterns of behavior.

The goal, she says, was to create a way to compare children from one center to the next. “We realized that the process by which clinicians made diagnoses was quite different at each center,” Dr Lord says. “Even the criteria for diagnosis were different. We wanted to have information about various symptoms of autism so that we could describe participants in a way that anyone could interpret.”

The ADI-R and ADOS have been important in providing standardized methods for research on the genetics and neurobiology of ASD. Both instruments have allowed clinicians all over the world to have valid and reliable tools for identifying and specifying the behaviors that we now know as ASDs.