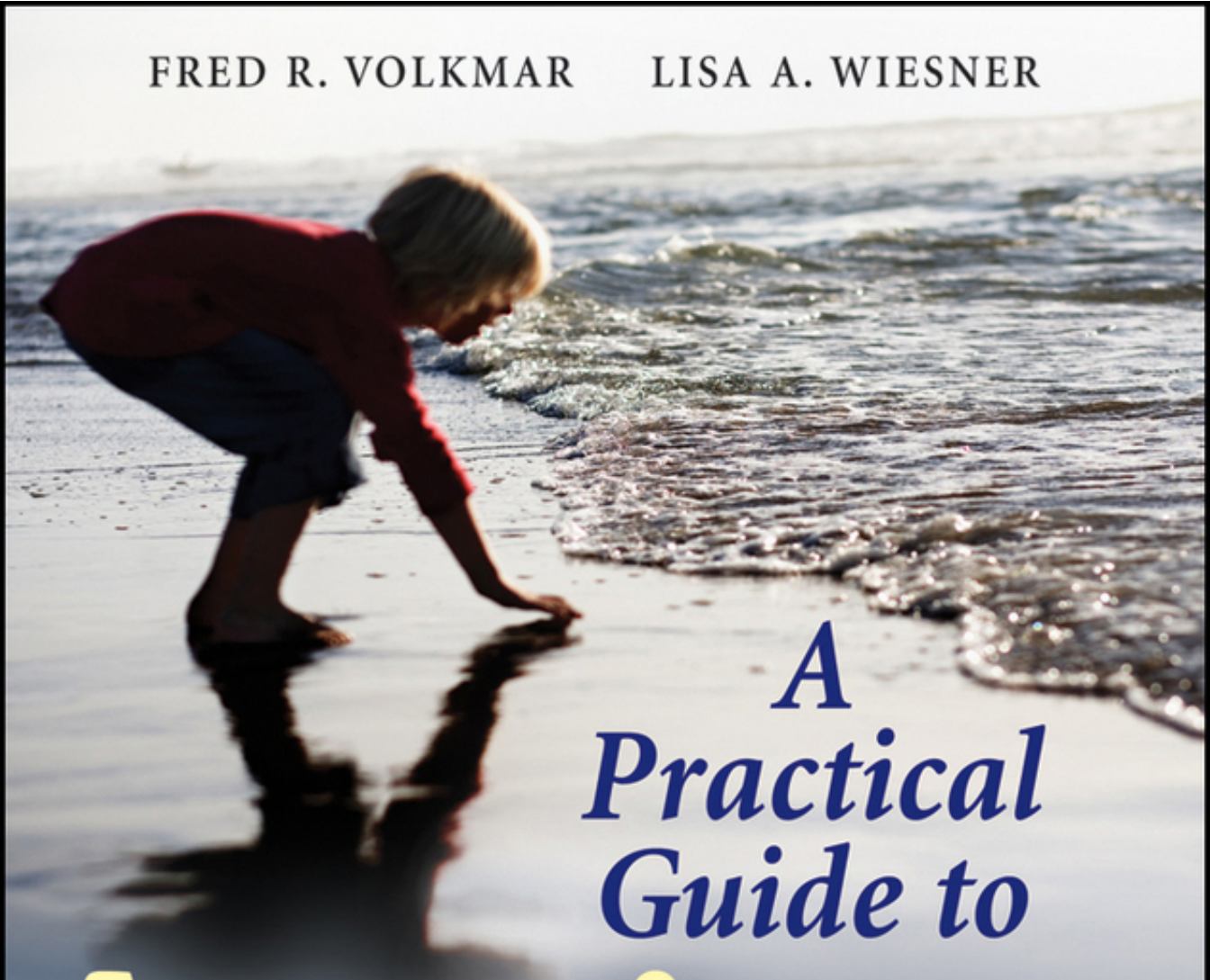
FRED R. VOLKMAR LISA A. WIESNER



A Practical Guide to Autism

What Every Parent, Family Member, and Teacher Needs to Know

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Fred R. Volkmar

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To Lucy and Emily,
who have taught us about being parents
and to the many children and their parents
who have taught us about being doctors

Contents

Foreword

Autism is in the public spotlight now more than ever as new research and information appears almost daily. Although in many ways this is a positive development it also presents challenges to families and practitioners who want to keep up with the latest developments and are left to sift through new information by themselves to see what is credible and relevant for them. Each of us needs a personal research assistant who can determine which information we need to pay attention to and let us know how it might affect our daily work and the children we are living with or serve.

Since we each don't have our own research assistants on staff, I am delighted to recommend this wonderful book by Fred Volkmar and Lisa Wiesner. Both of these talented professional leaders have combined their scientific skills and understanding of the field with great practical experience and ideas about how research can be translated into clinical practice. The result is a book that provides the best and most comprehensive information about recent scientific developments and a splendid practical guide for how they are being implemented and what we are learning in the process. The issues are presented in all of their complexity but translated into language that is clear, direct, and easy to follow. The format also lends itself to understanding the complex issues and their implications through excellent charts, question and answer sections, and chapters that vary from describing diagnostic issues to stating very specifically how to expand and evaluate the services one is receiving. The comprehensive references and lists of additional resources also add greatly to the overall package.

As a professional dedicated to understanding scientific advances and helping families and teachers to utilize them most effectively, I am very pleased to have an ally like this book available. I am very grateful to the authors for providing a very credible, practical, and relevant addition to our field to help the many advocates and family practitioners to better understand the exciting new developments and how they can be implemented in our day to day work. Those taking

the time to read through this superb volume will find it time well spent that pays back dividends in many different ways.

Gary B. Mesibov, Ph.D.
Professor and Director of TEACCH
Treatment and Education of Autistic and Related Communication Handicapped Children
University of North Carolina at Chapel Hill

Preface

We have written this book to provide parents and teachers with information that we hope will help them get the best possible care for their child or the children under their care. The two of us approach this from slightly different perspectives. One of us, Fred Volkmar, is a child psychiatrist whose main area of clinical work and research is in autism. The other, Lisa Wiesner, is a pediatrician who has seen children with autism and other disabilities in her pediatric practice. In addition to bringing our professional perspectives to the book, the two of us are married and parents of two children. We hope that this book will provide parents and teachers of individuals with autism and related disorders some practical and useful information.

Our aim is to provide an understandable guide to what we feel are the most important things for parents and teachers to know. We try, as much as possible, to refer readers to relevant materials—both books, chapters, and research papers. We have tried, as much as we can, to stick with resources that are readily available and written in ways that parents as well as professionals can understand. It is, in some ways, gratifying to be able to say that we are by no means exhaustive in this regard. There are many excellent resources not included here, although we've tried to give a reasonable sample of the best things available.

We have tried to think very broadly about the kinds of information that parents and teachers need to know. Accordingly, we have chapters focused on a wide range of topics. Given our interest in the medical as well as behavioral and educational aspects of autism, we've included chapters on medical issues, safety, and medications, as well as what would be the more usual chapters in a book of this kind.

We are very much aware that, for parents, the rewards of raising a child with an autism spectrum disorder are just as great as for any other parents. However, the challenges can be more daunting because parents have to take the child's difficulties into account in almost all decisions made about his or her education and health care. This book reviews basic information about autism and related conditions and how these disorders are diagnosed and treated. Keep in mind that in this book we are trying to provide important *general* information that will help parents and teachers provide good care. This book can't (and won't)

substitute for having a good working relationship with the various professionals who can advise you about what is best for your child in particular. The information provided in this book should supplement but does not replace the need for the child and family to have an ongoing relationship with educators, health and mental health care providers, and legal professionals who know the child and specifics of the situation very well. Laws, for example, can vary significantly from state to state, and this knowledge may be important as parents and others think about long-term planning. Similarly, keep in mind that while we've made every effort to be accurate and up to date, knowledge changes over time, and with the increasing amount of research on autism, the pace of change has quickened considerably.

Over the past decade there have been important advances in understanding the syndrome and in treating its symptoms. We know now that we can recognize and reduce the negative effects of these conditions on the child's development and behavior. In considering any intervention, it is always important to weigh the risk against the possible benefit of the intervention. As the saying goes, ''The perfect is sometimes the enemy of the good.'' That is, sometimes it is better not to strive for perfection but for reasonable care and quality of life. As discussed in this book, many new treatments for autism also periodically become available. Sometimes these are well evaluated scientifically. Unfortunately, much of the time they are not. In a later chapter in this book, we will review some of these treatments and discuss how parents and teachers can make informed decisions about using them.

In each chapter we include questions from parents and our answers. We hope that these are a helpful way for you to learn from the experience of others. Throughout the book, we also include some tables and boxed material with additional information. The final sections of the book include a glossary as well as a list of additional resources. A reading list is included at the end of every chapter. Again, remember that in reading this or any book, it is important to interpret the information with the specific child in mind. In this effort, various professionals can be important allies.

We are grateful to a number of our colleagues who have reviewed parts of this book in our efforts to make it helpful to parents. We have profited from their wisdom and comments. They include, in alphabetical order: Karyn Bailey, Leah Booth, Rebecca Carman, Kasia Chawarska, Mark Durand, Tina Goldsmith, Debbie Hilibrand, Ami Klin, Kathy Koenig, Andrés Martin, Prisca Marvin, Nancy Moss, Rhea Paul, Michael Powers, Brian Reichow, Terrell Reichow, Penn Rhodeen, Celine Saulnier, Larry Scahill, Alison Singer, Tristam Smith, Kathy Tsatsanis, and Sally Zanger. We also thank our various colleagues who kindly allowed us to reprint, sometimes with modifications, materials from other sources, particularly the *Handbook of Autism*. Gary Mesibov merits special

thanks for his willingness to contribute a foreword. We also are grateful to our editor, Patricia Rossi, and the staff at Wiley for their unflagging support and help in making this book as parent friendly as possible. We also thank a dedicated staff of professionals who helped us assemble the final copy—Lori Klein and Rosemary Serra, and, in particular, Emily Deegan, who went above and beyond the call of duty in coping with the many demands of producing this book. Finally, we thank our children, who have taught us much about child development, and, of course, our patients and their families, who have taught us much about autism.

Fred Volkmar, MD
Lisa Wiesner, MD

CHAPTER 1

What Is Autism?

This chapter gives some background on **autism** and related **autism spectrum disorders (ASDs)**. The recognition of autism as a disorder is a relatively recent one, first described in 1943 but not ''officially'' used as a diagnosis until 1980. Other conditions such as Asperger's disorder were ''officially'' recognized even more recently. In this chapter we discuss these disorders and how our understanding of them has changed over the years. This is important for several reasons. One is that you may hear many different terms used to describe a child's difficulties. Second, because knowledge has changed over the years, there are some misconceptions about autism that you may encounter (particularly among people who haven't kept up with the field!). Finally, if you are looking at this book, you are probably wondering if a child you know has autism. We think it would be helpful for you to know something about autism!

Some Terms

The term ***pervasive developmental disorder*** **(PDD)** refers to the overarching group of conditions to which autism belongs. The term *PDD* refers to the *class* of disorder to which autism belongs—autism is a kind of PDD like apples are a kind of fruit. Within this class, several disorders are now officially recognized: Autism (also referred to as autistic disorder, infantile autism, or childhood autism), **Rett's disorder**, **childhood disintegrative disorder (CDD)** (also sometimes referred to as Heller's syndrome or disintegrative ''psychosis''), **Asperger's** disorder (also sometimes called Asperger's syndrome or autistic psychopathy), and, finally, **pervasive developmental disorder not otherwise specified (PDD-NOS)** (sometimes termed *atypical PDD* or *atypical autism*). The terms *PDD* and *PDD-NOS* are sometimes confusing. The term *PDD* technically refers to all these disorders—that is, to the entire group of conditions. The term *PDD-NOS* is a specific diagnosis included within the PDD category; it refers to a condition in which the child has some troubles suggestive of autism,

but these don't seem to fit the better defined diagnostic categories - it is essentially a term for conditions that are suggestive of autism but ''not quite'' autism. Paradoxically, this condition is probably the most common pervasive developmental disorder but is also the least studied. Although the term *ASD* is commonly used, it is not an ''official'' term but generally means the same thing as PDD, that is a disorder somewhere in the autism ''ballpark.'' There are official guidelines for the diagnosis of each condition (see Appendix 1).

What Is an "Official" Diagnosis?

The most frequently used system for diagnosis in the United States is the American Psychiatric Association's ***Diagnostic and Statistical Manual of Mental Disorders***. It now exists in its fourth edition and is often referred to as **DSM-IV**. The diagnoses in the DSM-IV and the code numbers assigned to these diagnoses are used for many different purposes, such as record keeping, public health information, and insurance reimbursement. The DSM shares code numbers with the international diagnostic system (*International Classification of Diseases*, 10th ed. [**ICD**-10]). Fortunately, at present, the DSM and ICD approaches to the diagnosis of autism and related conditions are essentially the same. These give guidelines to physicians and other health care providers about diagnoses. As you can imagine, when there is a simple blood test or guideline (e.g., as there is for diabetes), the diagnostic part of things is pretty straightforward. For other conditions, particularly those involving development and behavior, we are not yet at the stage of having simple blood tests, so these guidelines focus more on the history of the child's development and observation of the child's behaviors. These guidelines are intended to particularly help people (including health care professionals) who aren't experts about the specific conditions. As we will discuss later in the book (Chapter 3), there are also some other good approaches to screening for conditions like autism—often, these are based on the official guidelines.

PERVASIVE DEVELOPMENTAL DISORDERS (PDDS) (SOMETIMES REFERRED TO AS AUTISM SPECTRUM DISORDERS [ASDS])

Official Name	Other names
Autistic disorder	Childhood autism, infantile autism, early infantile autism
Rett's disorder	Rett's syndrome
Childhood disintegrative disorder	Heller's syndrome, disintegrative disorder, disintegrative psychosis
Asperger's disorder	Asperger's syndrome, autistic psychopathy, autistic personality disorder
Pervasive developmental disorder NOS	Atypical PDD, atypical personality, atypical autism

AUTISM

Leo Kanner and the First Description of Autism

The condition now known as **autistic disorder, childhood autism**, or **infantile autism** (all three names mean the same thing) was first described by Dr. Leo Kanner in 1943. Dr. Kanner, the first child psychiatrist in this country, reported on a group of 11 cases that appeared to exhibit what he called ''an inborn disturbance of affective contact.'' By this he meant that, in contrast to normal babies, these children came into the world without the usual interest in other people. For normally developing babies, people are the single most interesting things in the environment. Kanner believed that the difficulty for children with autism in dealing with the social world was congenital in nature; that is, the children were born with it. Dr. Kanner gave a careful description of the unusual behaviors these first cases exhibited.

For example, he mentioned that these children exhibited ''***resistance to change***.'' By this he meant that they literally were resistant to change, and also referred to this as ''**insistence on sameness**.'' For example, a child might require that the parents take the same route to school or church every time they went and become very upset if there were any deviation from this routine. They might panic if anything in their living room was out of place. They might be very rigid about what kinds of clothes they would wear or foods they would eat. The term *resist*ance *to change* also was used to refer to some of the unusual behaviors frequently seen in autism, for example, the apparently purposeless motor behaviors (stereotypies) such as body rocking, toe walking, and **hand flapping**. Dr. Kanner mentioned that when language developed at all, it was unusual. For example, the child with autism might fail to give the proper tone to his speech (i.e., might speak like a robot) or might echo language (**echolalia**) or confuse personal pronouns (**pronoun reversal**). For example, when asked if he wanted a cookie, the child might respond, ''Wanna cookie, wanna cookie, wanna cookie.'' Sometimes the language that was echoed was from the distant past (delayed echolalia). Sometimes it happened at once (immediate echolalia). Sometimes part of it was echoed but part had been changed (mitigated echolalia). In his original report, Kanner stated that there were two things essential for a diagnosis of autism: (1) the autism or social isolation and (2) the unusual behaviors and insistence on sameness.

As time passed, it became clear that language/communication problems were also important in the diagnosis (when you think about it, of course, language is an important aspect of social development!). Including these problems along with the early onset of the condition that Kanner mentioned, we have what continue to be the four hallmarks of autism: (1) impaired social development of a type quite different from that in normal children; (2) impaired language and

communication skills—again of a distinctive type; (3) resistance to change or insistence on sameness, as reflected in inflexible adherence to routines, motor mannerisms, stereotypies, and other behavioral oddities; and (4) an onset in the first years of life.

KANNER QUOTE

The outstanding, ''pathognomonic,'' fundamental disorder is in the children's *inability to relate themselves* in the ordinary way to people and situations from the beginning of life. Their parents referred to them as having always been ''self-sufficient''; ''like in a shell''; ''happiest when left alone''; ''acting as if people weren't there''; ''perfectly oblivious to everything about him''; ''giving the impression of silent wisdom''; ''failing to develop the usual amount of social awareness''; ''acting almost as if hypnotized.'' This is not, as in schizophrenic children or adults, a departure from an initially present relationship; it is not a ''withdrawal'' from formerly existing participation. There is from the start an *extreme autistic aloneness* that, whenever possible, disregards, ignores, shuts out anything that comes into the child from outside. Direct physical contact or such motion or noise as threatens to disrupt the aloneness is either treated ''as if it weren't there'' . . . resented painfully as a distressing interference.

. . . This insistence on sameness led several children to become greatly disturbed upon the sight of anything broken or incomplete. A great part of the day was spent in demanding not only the sameness of the wording of a request but also the sameness of the sequence of events.

. . . The dread of change and incompleteness seems to be a major factor in the explanation of the monotonous repetitiousness and the resulting *limitation in the variety of spontaneous activity*. A situation, a performance, a sentence is not regarded as complete if it is not made up of exactly the same elements that were present at the time the child was first confronted with it. If the slighted ingredient is altered or removed the total situation is no longer the same. . . .

From Leo Kanner, Autistic disturbances of affective contact, *Nervous Child, 2*, 217–250, 1943.

Some Early Mistakes About Autism

While Kanner's description remains a ''classic,'' it was not, of course, the last word on the subject. Some aspects of his original report inadvertently served to mislead people. Some of these mistaken first impressions took many years to clarify. For example, Kanner originally thought that children with autism

probably had normal **intelligence**. He thought this because they did rather well on some parts of **intelligence (IQ)** tests. On other parts, however, they did quite poorly or refused to cooperate at all. Kanner assumed that, if they did as well on all parts of the IQ test as they did on the one or two parts on which they seemed to do well, the child would not be retarded. Unfortunately, it turns out that often cognitive or intellectual skills are difficult to assess, in large part because they are very scattered. Put another way, children with autism often do some things well, such as solving puzzles, but they may have tremendous difficulty with more language-related tasks. The degree of discrepancy among different skill areas is very unusual in the typically developing population but very frequent in autism. We now appreciate that many, maybe about half, of children with strictly defined autism function in the range of **mental retardation (MR)** or **intellectual disability**[1] when you combine all of their sometimes quite variable scores. However, the pattern of performance in autism is very unusual and quite different from that usually seen in mental retardation without autism. You will see examples of this in Chapter 7, 8 and 9. Similarly, since the different abilities that go into estimating one's intelligence are often so different in autism, the use of a single score can be rather misleading; for example, sometimes a child with autism may have average or above-average abilities when it comes to tasks that are not verbal, whereas the same child's ability with verbal tasks can be very significantly delayed. In such cases, which score is the right one? Both are, in some sense, but this means that you have to understand this and avoid using a single score to represent how the child functions. Sometimes schools or agencies will want to use a single overall score to describe the child's cognitive abilities, but in fact the single score may be very misleading.

Fairly frequently (maybe 10% of the time), children with autism have some unusual ability, for example, to draw (see Figure 1.1), play music, or memorize things, or sometimes calculate days of the week for events in the past or future (calendar calculation). These abilities are usually isolated (the otherwise wonderful portrayal of the man with autism in the movie *Rain Man* is a bit misleading in this respect). These individuals, now usually referred to as *autistic savants*,

[1]Note that in some countries, such as England, people have started to refer to mental retardation as a learning disability; in the United States, the term *learning disability* generally refers to a very specific problem in learning (e.g., in reading). We will consistently use the term *intellectual disability* in this book to refer to the combination of significantly subaverage IQ (below 70) and similarly delayed adaptive skills as defined in the DSM-IV as mental retardation. We also use the term *developmentally delayed* to refer to children, especially young children, who seem to be at high risk for having intellectual disability/mental retardation.

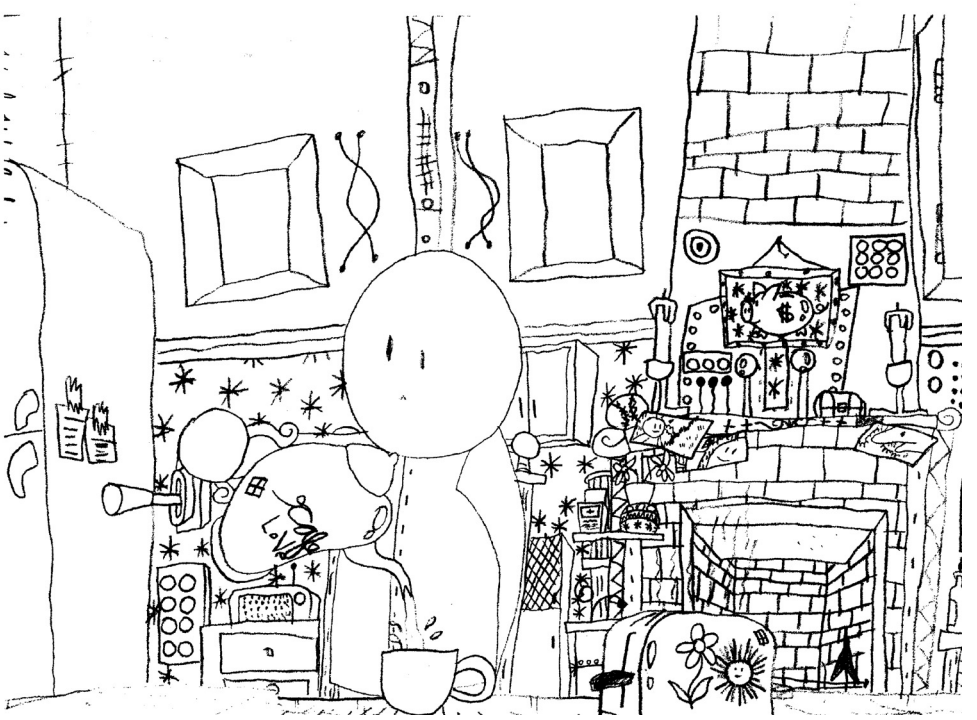

FIGURE 1.1 **"Bim gets breakfast in the love kitchen." Drawing by a child with autism. Bim is the child's made-up cartoon character. We thank the child and his parents for permission to reproduce. Reprinted, with permission, from F. Volkmar and D. Pauls, Autism. *The Lancet, 2362*, 1134, 2003. Reprinted with permission.**

sometimes lose their abilities as they get older. But it was just this kind of remarkable ability that led people to minimize the child's areas of difficulties.

FALSE LEADS FOR RESEARCH

- Impression of normal levels of intelligence because children did well on some parts of IQ tests:
  Implication: Bad performance due to lack of motivation of child (rather than variabilty in skills)
  Subsequent research: Significant scatter in abilities is often present, marked discrepancies between skills areas (e.g., verbal and nonverbal IQ) are common.
- Autism a form of schizophrenia:

Impression: Confusion with schizophrenia given the use of the word *autism* (earlier used to describe self-centered thinking in schizophrenia).
Implication: Autism might be the earliest manifestation of schizophrenia.
Subsequent research: Autism and schizophrenia are not related; rarely (no more than expected by chance) children with autism develop schizophrenia.

- Increased rate in more families with higher levels of education in Kanner's original paper:
  Implication: Effects of experience.
  Subsequent research: There is no increase in autism among parents with more education (more educated parents likely to get to the one child psychiatrist in the country).
- No associated medical conditions (children had an attractive appearance):
  Implication: Exclusion of ''organic'' cases (if medical condition present) from having autism.
  Subsequent research: High rates of seizures, higher than expected rates of some disorders—especially some genetic disorders.

Another source of confusion came because Dr. Kanner originally suggested that autism was not associated with other medical conditions. We now know this is not true. Over the years, hundreds of conditions have been reported to be related to autism; it now seems that really only a few are especially frequent with autism. For example, we now know that sometimes autism is seen with conditions like **fragile X syndrome** or **tuberous sclerosis** (both of which will be discussed in Chapter 10).

When we look at all the different medical conditions that might be involved in causing or contributing to the child's autism, probably no more than 10% of autistic individuals have them. Most importantly, as children with autism were followed over time, it became apparent that 20–25% of them would develop **seizures (epilepsy)**, as we discuss in Chapter 12.

Dr. Kanner originally guessed that autism was a very distinctive condition, and we now know that this is true. At the same time, he used the word *autism*—a word that previously had been used to describe the unusual, self-centered, and self-contained thinking seen in a major mental disorder called **schizophrenia**. Thus, his use of the word *autism* suggested to many that perhaps autism was the earliest form of schizophrenia. It took many years for this to be clarified. We now know that autism and schizophrenia are not related. Very occasionally, but not more than would be expected by chance, individuals with autism may, as adolescents or adults, develop an illness like schizophrenia. Autism differs from schizophrenia, however, in many different ways, including its clinical features, course, associated difficulties, and family history.

Finally, Kanner mentioned that in 10 of 11 families, the parent or parents were highly educated and successful. It also appeared that parents and children interacted somewhat unusually at times. This led to the idea, particularly in the 1950s, that highly successful parents somehow ignored or otherwise ill treated their child to cause autism and that, as a result, autistic children might be well served by isolating them from their families. This view was taken by a man named Bruno Bettelheim at his school at the University of Chicago. It is now very clear that this is not true. Instead, it is clear that Kanner's original sample was a highly selected one; that is, individuals who were very educated and successful in the 1940s would be just the kinds of people who could find the one person and only child psychiatrist in the country who was doing research on the kinds of problems their children had. It also became clear that unusual aspects of parent–child interaction were just as likely to come from the child, rather than the parent. In contrast to the 1950s, where often the emphasis was on putting the child in an institution, we now believe that children with autism are best served by remaining in their families and communities and that other children, parents, and family members are their best and strongest advocates.

Services for Children with Autism

Until the passage of the Education for All Handicapped Children Act in 1975, parents of children with autism often were at a loss as to how to educate them. Research began to suggest that structured educational programs were more effective than unstructured ones—that is, programs in which the adult had an agenda for teaching the child were better than ones in which the child was left to her own devices to learn. Before 1975, parents often were told by schools that there was no way their child could be educated. Often, parents were advised to place their child in a residential or large state institution where the child got little in the way of intervention. Indeed only a small proportion of children with autism were educted in public schools before passage of this law.

Now schools in the United States are mandated to provide a free and appropriate education for all individuals with disabilities. This is a radically different approach. As programs have become increasingly sophisticated, schools have done an increasingly better job of providing education for children with autism. This means that schools often are now the major place for **intervention** for children with autism. As a result, it appears that more children are being identified in schools and receiving services and, importantly, it also seems that, as a group, children with autism are doing better. As we'll talk about in Chapter 9 many are now able to go to college.

Asperger's Disorder

In understanding Asperger's disorder, it is important to know where the concept came from in the first place, how it has been used over the years to refer to very different kinds of problems in children, and how it is used now. Hans Asperger was a medical student working at the University of Vienna during World War II. He had to write a paper on some aspect of research, and he chose to write his paper on boys who had trouble forming groups. These boys had marked social problems, but their language and communication was, in some ways, very good. Asperger described them as being rather pedantic ''little professors'' who tended to intellectualize everything. Asperger also mentioned that they had unusual interests. For example, the child would know all the train or bus schedules into and out of Vienna. These unusual and what are termed **circumscribed interests** continue to be an important feature of the condition. They are unusual in that they are indeed highly circumscribed but, more importantly, they interfere with other aspects of the child's life. This is what changes something from being a personality quirk into a disorder that merits intervention. In addition, Asperger mentioned that the boys were clumsy and awkward in terms of motor skills. He also mentioned that in several cases it appeared that other family members, particularly fathers, had similar kinds of problems.

ASPERGER'S DISORDER

- Asperger (1944) medical student in Vienna wrote his medical school thesis on boys who couldn't form groups.
- Described a series of boys with marked social and motor problems, unusual circumscribed interests (that interfered with getting skills in other areas), but good language and cognitive abilities. Family history was often positive for similar problems in fathers.
- Modifications in original description over time. Cases seen in girls, in lower IQ individuals, some individuals with language problems.

Asperger thought of the condition he described as something more like a personality trait, rather than a developmental disorder. He speculated that the condition was not usually recognized until after about age 3. Asperger originally chose a name that has been translated from the German as either *autistic psychopathy* or *autistic personality disorder* for the condition; that is, he used the word *autistic* in the same way that Leo Kanner, just a year before, had used the word *autism*. However, because of the war, neither Asperger nor Kanner knew of each other's work for some time. Asperger used the word *psychopathy* because he also noted

that these boys had difficulties with being compliant and had some behavior problems. In recent years, the practice has been to refer to this condition as *Asperger's disorder* or *Asperger's syndrome* (AS). Asperger, who lived for many years after describing this condition, saw many cases in his lifetime. And even until the end of his life, he felt that the condition was different from infantile autism.

Although Asperger had been working on his condition for many years following World War II, it received little recognition until the 1980s in English-speaking countries when Dr. Lorna Wing published a paper on it. She said that some aspects of Asperger's original report had to be modified. For example, she felt AS could be seen in girls and children with mild mental retardation. She also pointed out that the family histories could be more complicated than Asperger originally thought. As time went on, several different views of Asperger's disorder came into being. Some people were confused about the relationship of Asperger's disorder and autism; that is, whether Asperger's disorder was just the same thing as autism in smarter people. Another set of investigators and clinicians equated the term with adults with autism. Yet another set of clinicians would use the term *Asperger's disorder* interchangeably with the term *pervasive developmental disorder not otherwise specified* or *atypical pervasive developmental disorder* (this is a concept discussed subsequently in this chapter). Finally, some continued to use the term *Asperger's disorder* to refer to a specific set of symptoms that would deserve a special category in a book like the DSM-IV.

Additional problems arose because researchers outside the field of psychiatry also began to see very socially odd children who did not quite seem to have autism. A number of terms came into use that had some degree of overlap with AS. For example, some **neurologists** described something they called the *right hemisphere learning disability syndrome*; from within the speech/language literature came the concept of *semantic–pragmatic processing disorder*, and from psychology a profile of disabilities called the **nonverbal learning disability** (NLD) syndrome was described. Within the field of psychiatry itself, there have been some attempts to describe children with problems similar to those described by Asperger, notably the notion of "**schizoid personality**" as described by Sula Wolf and her colleagues (1995). It is perhaps not surprising, given all these factors, that there has been much controversy about Asperger's disorder.

As currently defined, AS shares some features with autism—notably, the social interaction problems—but early language and cognitive skills are relatively preserved. In contrast to autism, the child's difficulties usually are not recognized for some years, and usually the child has a very intense and all-absorbing interest. Figure 1.2 provides an autobiographical statement (name changed, of course) and drawing illustrating this 10-year-old boy's area of obsessive interest—in his case, time in the universal sense. He was interested in fitting together the various

My name is Robert Edwards. I am an intelligent, unsociable but adaptable person. I would like to dispel any untrue rumors about me. I cannot fly. I cannot use telekinesis. My brain is not large enough to destroy the entire world when unfolded. I did not teach my long-haired guinea pig, Chronos, to eat everything in sight (that is the nature of the long-haired guinea pig).

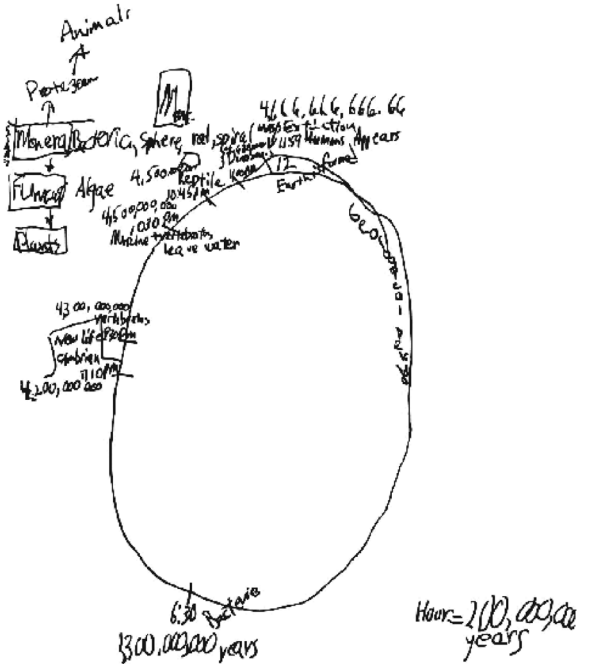

FIGURE 1.2 **Autobiographical statement and drawing of area of special interest, 10-year-old boy with Asperger's disorder.**

Volkmar et al. *American Journal of Psychiatry, 157*(2), 262–267. Reprinted with permission.

periods of recorded and prerecorded history and would spend his free time researching these issues and trying to talk to peers about them.

Sometimes it is difficult to distinguish AS from high-functioning autism. Regardless of which is the better term, the important thing here is to highlight that the child's difficulties are not simply willful bad behavior, but have come from a developmental problem—this is particularly true when a child has good verbal skills. Often, children with AS have a particular kind of learning disability, NLD where nonverbal skills can be quite impaired even when verbal skills are good. Documenting the child's profile of strengths and weaknesses can be very helpful to schools. One important treatment difference from more typical autism is that since children with AS have better verbal skills, we can sometimes use

language-based treatments such as very structured and problem-oriented **psychotherapy** and counseling. If verbal skills are much better than nonverbal ones, we can also try to use this in teaching. Other implications of AS may have to do with other aspects of planning, for example, vocational planning. Patients with AS may not have good motor skills, and it is important to realize this in helping them plan for adult work.

NONVERBAL LEARNING DISABILITY (NLD)

- A *profile* (pattern of strengths and weaknesses) on psychological testing.
- Both *assets* AND *deficits* are present.

Strengths	Weaknesses
Auditory perception	Tactile perception
Rote verbal capacities	Motor coordination
Verbal memory skills	Visual-spatial skills
Verbal output	Nonverbal problem solving

- NLD has a deleterious impact on the person's capacity for socialization.
- NLD is not an official diagnosis.
- NLD does seem to be frequently associated with Asperger's disorder and sometimes PDD-NOS but NOT with autism (in autism a different profile is usually seen with nonverbal skills being a relative strength for the child, not an area of weakness).
- Important implications of NLD profile for research on brain mechanisms.

Childhood Disintegrative Disorder

Although it is, fortunately, rather rare, CDD is important for several reasons. It first was described almost 100 years ago by a specialist in special education, Theodore Heller, who was working in Vienna. He noticed that several children had developed normally for some years and then had a marked and profound loss of skills. They did not regain skills to previous levels. Originally, Heller termed this condition ***dementia infantilis***. Subsequently, it has been called other things: **disintegrative psychosis**, **Heller's syndrome**, or now childhood disintegrative disorder. The term *disintegrative psychosis* captured the child's loss of skills, but the word **psychosis** implied some loss of reality testing, which we no longer believe exists. The term *childhood disintegrative disorder* has the advantage of describing the condition without prejudging its cause. This condition clearly is quite rare, although it is also the case that many times children with the

condition probably have not been adequately diagnosed or studied. Consistent with what Heller said in the first place, children with this condition develop normally for several years of life. Typically, they talk on time, walk on time, acquire the capacity to speak in sentences, are normally socially related, and have achieved bladder and bowel control. Usually between the ages of 3 and 4 years, the child experiences a marked and enduring regression in skills. Many behaviors that resemble those in autism develop, such as the motor mannerisms (stereotypies) and the profound lack of interest in other people. One of the interesting questions for present research is whether children with autism who have a major regression in their **development** are exhibiting something like this condition. We talk more about these issues and regression in Chapter 13.

CHILDHOOD DISINTEGRATIVE DISORDER (CDD)

- First described by Theodore Heller in 1908.
- Child has a period of normal development.
  Usually 3–4 years, normal language and self-care skills.
  By definition, the child has the capacity for speech.
- Either rapid or more gradual regression in multiple areas.
  Child comes to exhibit many features of autism.
- Sometimes a brain-based disorder is found that accounts for the regression.
- Usually minimal recovery (outcome in general is worse than autism).
- Condition is rare but of much interest given potential for finding a specific cause.

Rett's Disorder

In 1966, a Viennese physician, Andreas Rett, described a group of girls with an unusual history. They were apparently normal at birth and developed normally for the first months of life. However, usually within the first year or so of life, their head growth began to decrease in rate. In addition, they started to lose developmental skills they had acquired. As time went on, they lost purposeful hand movements, and various unusual symptoms began to develop. They seemed to lose interest in other people in the preschool years, which is why there was the potential to misdiagnose the girls as having autism. As they became somewhat older, the developmental losses became more progressive and quite different from those in autism. Unusual hand-washing or hand-wringing stereotypies developed. Purposeful hand movements were lost (see Figure 1.3).

FIGURE 1.3 **Stereotypic hand movements in Rett's syndrome**

Reprinted, with permission, from R. Van Acker, J. Loncola, and E. Y. Van Acker. Rett's syndrome: A pervasive developmental disorders. In F. Volkmar, R. Paul, A. Klin, & D. Cohen (Eds.), *Handbook of autism and pervasive developmental disorders*, (3rd ed., chap. 7, p. 127).Hoboken, NJ: Wiley, 2005.

Additionally, the girls developed other unusual respiratory symptoms, such as breath-holding spells or air swallowing (aerophagia). Seizure disorders sometimes developed as well. Problems in walking and in posture were seen and, over time, **scoliosis** (curvature of the spine) often developed. By adulthood, the girls had become severely retarded young women. However, their course was different from that seen in autism. The degree of problems in breathing, loss of hand movements and other motor difficulties, curvature of the spine, and so on suggested that this was a very distinct condition. We discuss Rett's disorder in much more detail in Chapter 12.

RETT'S DISORDER

- First described by Andreas Rett (1966).
- All cases were female in his original report.
- Early development was normal.

- Head growth slowed (relative to rest of body).
- Purposeful hand movements lost.
- Some ''autistic-like'' features which tended to lessen over time.
- Various associated problems.
  - Scoliosis (curvature of the spine) and movement problems.
  - Unusual breathing patterns/breath-holding spells.
- Characteristic course.
- A gene has recently been identified that seems to be responsible for Rett's in at least some cases.

Pervasive Developmental Disorder Not Otherwise Specified

PDD-NOS is the so-called subthreshold pervasive developmental disorder. That is, this is the category that is used when a child, adolescent, or adult exhibits some features of a PDD but does not meet all the criteria for a diagnosis of one of the very specifically defined PDDs. This diagnosis is problematic in that it is a matter of clinical judgment on the part of clinicians as to whether to use it. Probably not surprisingly, given the essentially ''nondefinition'' definition, the term is used very inconsistently. Furthermore, the nature of this definition means that it is hard for researchers to have funding to study the condition. Somewhat paradoxically, it almost certainly is the case that this condition is several times more common than autism, affecting perhaps one in several hundred children. When you hear on the radio or television that the rate of autism is 1 in 150 children, it actually is more accurate to say that the rate of autism *spectrum* disorder is 1 in 150; this number includes all of the disorders within the PDD class.

PDD-NOS

- Historic interest in children with some but not all features of autism (''autistic like'').
- By definition, the definition is a ''negative'' one—that is, children who do not meet criteria for autism, Asperger's, etc.
- The child must have problems in the social area of the type seen in autism and at least one in either communication/play or odd behavior.

- The relationship of PDD-NOS to autism remains unclear—is this best seen as part of a "broad autism phenotype" or something different from autism?
- There may be several subtypes of PDD-NOS—some children with PDD-NOS have major problems with attention, others with emotional overreactivity.

Although research on PDD-NOS is relatively sparse, some studies have appeared in recent years. Moreover, clinicians often have more experience from a clinical point of view with it because it seems to be more common than strictly defined autism. Children with PDD-NOS have problems in social interaction, but these are not as severe and pervasive as those in autism. These may include sometimes more overt and sometimes more subtle problems, for example, in initiating conversation, in playing with other children, in relating to parents or siblings. Unusual sensitivities are relatively common, although again usually not as severe as in autism. The term *PDD-NOS* is sometimes also used for children with very severe intellectual deficiency, who often have some features of autism, particularly stereotyped motor movements. With the exception of very retarded children, the outcome in PDD-NOS generally appears to be better than in most if not all of the other PDDs.

For a diagnosis of PDD-NOS to be made, the child should exhibit some problem in social interaction of the type usually seen in autism or other PDDs and at least some problem either in language and communication skills or in unusual behavioral responses to the environment and restricted interests. A single symptom of autism is, by itself, *not* sufficient for a diagnosis of PDD-NOS; rather, there have to be troubles in both the social area and either the language/communication or unusual behaviors category. As a practical matter, the diagnosis of PDD-NOS is used in several rather different situations. Sometimes very young children have many, but not all, of the features of autism. For example, at age 30 months the child may have marked social and communicative difficulties but does not exhibit the unusual behaviors usually associated with autism. This child might be given a diagnosis of PDD-NOS but then go on to develop unusual mannerisms or movements or other unusual responses to the environment; in this case, the diagnosis of autistic disorder would then be made. Occasionally, the term is used rather loosely; for example, someone may talk to you about PDD or mild autism when they mean PDD-NOS. In such situations, it is important to explicitly ask what is meant. Keep in mind that it is perfectly appropriate for a clinician to say that he or she isn't absolutely certain about the diagnosis particularly for younger children where the issue may become clarified only with certainty over time. What is important is getting appropriate services for the child.

How Common Are Autism and Related Conditions?

The first studies of the frequency or epidemiology of autism were conducted in the 1960s. Since that time, many studies have been conducted, mostly in Great Britain and countries other than the United States. Given what we know about autism, there is no reason to suppose that the frequency of autism is vastly different here, although there has been concern, as we'll discuss in a moment, that the frequency of autism may be increasing in this country. The lack of many good studies in the United States complicates the answer to this question, as we'll see shortly; it also makes it harder for educators and others to plan for the care of children with autism.

The various studies around the world have involved over 4 million children. Estimates of the rate of autism vary somewhat from study to study. If you lump all the studies together, a reasonable estimate of the rate is around 1.3 cases per 1,000 if you focus on autism strictly defined; if you broaden the group to include all individuals with a PDD or ASD, the number of cases is somewhere between 3 and 6 per 1,000 children.

Is the Rate of Autism Increasing?

There is some concern that the rate of autism may be increasing. However, we really don't know if this is true for several reasons. First, it is clear that awareness of autism has increased dramatically so that cases are more likely to be noticed. When one of us (FV) moved to New Haven in 1980 to work on autism at the Yale Child Study Center with Donald Cohen, people would ask what my research was about and when I said, ''Autism,'' they would frequently say something like, ''Isn't that wonderful—we need more artistic children''; people didn't even know what the word meant! Today, there are ads on radio and television from the Ad Council about autism and posted in the background of TV shows advertising autism-related groups. Another possible reason for an apparent (but not real) increase is changes in the diagnostic guidelines for autism—the current systems (both DSM and ICD) were designed to do a better job of detecting autism in more able children. Another problem has been the tendency to equate autism (strictly defined) with the much broader (and much less well defined) autism ''spectrum.'' Finally, there is an unusual problem with autism. Since the label often gets children more services than other labels, parents may push to get an autism label for educational purposes even if the child doesn't have autism strictly defined (this is a problem called **diagnostic substitution** and one of the reasons we have to be skeptical about state-reported data based on school services). This is a real problem since states, and sometimes regions within states,

vary widely (and wildly) in terms of how they provide services—in some states, only the label of autism really gets needed services. There is an excellent and very readable review of these issues in *Unstrange Minds*, a book by Roy Grinker, a professor who is also a parent (the book is listed in the reading list at the end of this chapter).

S D

It is clear that autism appears to be at least 3 to 5 times more frequent in boys. However, when girls have autism, they are more likely to have intellectual

TABLE 1.1 **DIFFERENTIAL DIAGNOSTIC FEATURES OF AUTISM AND NONAUTISTIC PERVASIVE DEVELOPMENTAL DISORDERS**

F	A D	A D	R D	C D D	P D DNOS
Age at recognition (months)	0–36	Usually >36	5–30	>24	Variable
Sex ratio	M>F	M>F	F (?M)	M>F	M>F
Loss of skills	Variable	Usually not	Marked	Marked	Usually not
Social skills	Very poor	Poor	Varies with age	Very poor	Variable
Communication skills	Usually poor	Fair	Very poor	Very poor	Fair to good
Circumscribed interests	Variable (mechanical)	Marked (facts)	NA	NA	Variable
Family history—similar problems	Sometimes	Frequent	Not usually	No	Sometimes
Seizure disorder	Common	Uncommon	Frequent	Common	Uncommon
Head growth decelerates	No	No	Yes	No	No
IQ range	Severe MR to normal	Mild MR to normal	Severe MR	Severe MR	Severe MR to normal
Outcome	Poor to good	Fair to good	Very poor	Very poor	Fair to good

M = male, F = female, MR = mental retardation, NA = not applicable.

SOURCE: Adapted, with permission, from F. R. Volkmar & D. Cohen, Nonautistic pervasive developmental disorders. In R. Michaels et al. (Eds.), *Psychiatry* (chap. 27.2, p. 4). Philadelphia, PA: Lippincott-Raven, 1985.

deficiency. We do not yet understand the basis for these differences. One theory is that, perhaps on a genetic basis, girls are generally somewhat less vulnerable to autism (hence the greater frequency in boys) and that for girls to have autism they must have greater genetic or central nervous system damage (hence the higher rate of intellectual deficiency in girls). Asperger's disorder and CDD are more common in males, and Rett's is usually found only in females. There does not appear to be as marked a sex difference in PDD-NOS.

R O D

Information on the frequency of other PDDs is not nearly as good as that for autism. Fortunately, both Rett's disorder and CDD are much less common than autism. AS estimates have ranged widely, from 1 in 500 children to 1 in 10,000; the condition is clearly less common if one uses a strict definition for it.

PDD-NOS is almost certainly the most common form of pervasive developmental disorder. Some have estimated the frequency of PDD-NOS as frequent as 1 in about 200, but again solid research data are lacking. Clearly, at least one in several hundred have some form of serious social disability consistent with autism or a related condition; this means that these conditions are a major public health problem. Table 1.1 summarizes the similarities and differences of the various PDDs.

Summary

This chapter has given some background information on autism and other PDDs. Relatively speaking, these are all fairly new diagnostic concepts, and in some ways it is surprising that we know as much as we already do about them. All these conditions share impairment in social interaction as a major feature although in Rett's the social problems are most notably early in life and then dramatically lessen. They differ from each other in various ways. The most well known of these disorders, autism, is seen in between 1 in 800 to 1 in 1,000 children. With the exception of PDD-NOS, the other conditions, such as Rett's disorder, CDD, and Asperger's disorder, are probably less common. We know that autism is often, but not always, associated with intellectual deficiency and is more common in boys than in girls. We also know that autism is frequently associated with evidence of brain impairment, such as seizure disorders, and parents (and doctors) should be alert to the possibility of a child's developing seizures. Fragile X is clearly seen in some children with autism, and routine testing for this condition makes sense. It makes sense to be particularly thorough when first evaluating a child for possible autism and in situations where the presentation is unusual and something ''does not quite fit.''

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QUESTIONS AND ANSWERS

1. **Is there a ''typical'' child with autism?**

   We have information, particularly from epidemiological studies, that tells us about *groups* of children with autism. In this sense, we have information on the typical or ''average'' child with autism, but it is important to realize that this is not a real individual; rather, it gives a sense of what can be. It is the *range* of what we see in autism, which is very unusual. As a diagnostic term, *autism* can be applied to the angelic-appearing, mute 2-year-old who is sitting in a corner playing with a piece of string; it can also apply to a college graduate who does computer ordering for a small company. If we had a large group of people with autism in a room together, we most likely would be struck initially by the differences and not the similarities. If, however, we spent more time, the similarities would begin to be seen. These would include major problems in negotiating through the social world, in communicating with others, and in responding to the nonsocial environment. These three commonalities are what are seen in every person with autism.

2. **Did autism exist before Leo Kanner described it?**

   Undoubtedly, there were cases of autism before Kanner first described the condition. For example, some people have argued that cases of

so-called ''wild'' or ''feral'' children like Victor the Wild Boy, who was reported in France in the early 1800s, were really children with autism. It is possible that before the improvements in childhood mortality in the 20th century and before the increased concern with children, cases of autism had not been noticed. It was Kanner's genius to be a very careful observer and become aware of what we now know as autism.

3. **Does the diagnosis really matter?**

   This is a good question—the answer is yes and a qualified no. The complication is that diagnosis is used for many different things—for research, for teaching people about commonalities in illnesses and disorders, for communicating rapidly, and for getting educational services. All these are legitimate goals, and the needs for diagnosis will vary somewhat depending on what the goal is. Essentially, the diagnosis grounds us in the general territory we are dealing with, but, of course, in terms of coming up with a program or treatment, we need to take the needs of the individual child into account. There are some people who would advocate against giving a diagnosis to avoid giving premature or stigmatizing labels; however, it is just these labels that sometimes help a child get services. For purposes of research, again depending on what is being studied, often rather strict diagnostic labels are needed, that is, to avoid what would be a confounding or complicating issue in interpreting the results of a research study.

4. **Is the frequency of autism increasing?**

   The short answer is that we don't really know for sure. It is the case that *schools* and *departments of education* have seen increased numbers of children with autism presenting for special services. One problem, however, is that labels for educational purposes may be more concerned with getting services than with precise diagnosis. Many educators understandably (from the point of view of giving services) lump autism and all related disorders together—this means that some of the ''data'' cited as indicating an increase in autism is really about the broader spectrum of autism and related conditions. Often, when one hears about the explosion of ''autism,'' one is really hearing about this. Another problem is that methods of diagnosis have changed over time and there has been a real (and in many ways successful) effort to expand the awareness of teachers, health care professionals, day care providers, and others about autism; that is, part of the reported increase may be more apparent than real in that the cases were already there but had been overlooked. Finally, the lack of good epidemiological data on autism, particularly in this country, makes it very difficult to answer the question in a way we could feel confident in. All this being said, it is concerning that estimates of autism around the

world seem to have increased over time—it remains to be seen how much this is a ''real'' increase.

5. **What are the differences between girls and boys with autism?**

   There are several differences. In the first place, boys are much more likely to have autism than girls (about 3 to 5 times more likely). But when girls have autism, they tend to have more severe cognitive problems. It is probably the case that, for whatever reason, girls are less vulnerable than boys and, accordingly, for a girl to get autism a bigger genetic ''hit'' is needed (presumably accounting for the greater degree of cognitive impairment). In Asperger's disorder (and in individuals with autism who function in the normal cognitive range), the ratio of boys to girls is much higher. In Rett's disorder, females are most commonly affected.

6. **Is childhood disintegrative disorder the same thing as disintegrative psychosis? If so, does that mean it is like schizophrenia?**

   An old term for childhood disintegrative disorder was *disintegrative psychosis*. This term came to be used at a time when science wasn't as advanced as it is today; the word *psychosis* was used in a very broad way. Today, the term *psychosis* has a very specific meaning; it implies a loss of reality testing and the presence of problems in thinking, such as delusions and hallucinations. Children with CDD do not exhibit these.

7. **Someone told me that Asperger's disorder is the same as autism. Is this true?**

   There is much disagreement about the relationship of the two disorders. Clearly, there is *some* relationship in terms of severe social difficulties. Several different problems complicate this issue. One problem is that the term *Asperger's disorder* has come to be used for many different things; another is that various terms for disorders have come through other sources and these concepts overlap (at least in part) with Asperger's. Terms like semantic–pragmatic processing disorder, right hemisphere syndrome, semantic–pragmatic disorder have all been used—along with nonverbal learning disability. Our own view, and that of the DSM, is that these are separate disorders. Major differences have to do with the fact that language is so very good (in some ways) in Asperger's. Also, in Asperger's, unusual preoccupations with fact-based knowledge about some topic is usually present. Finally, in contrast to autism, parents of children with Asperger's disorder tend not to be worried until the child enters preschool.

8. **Can Rett's coexist with autism?**

   No; by definition the two disorders are distinctive. There is a relatively brief ''autistic-like'' phase in Rett's (usually in the preschool years), but after that the conditions are quite different.

9. **My child has been diagnosed with PDD-NOS but the school has given him the label of autism. Is this okay?**

   Labels used by schools often differ somewhat from those used by medical professionals. These labels also vary considerably from state to state and sometimes within states! Often, for purposes of getting appropriate services, the label *autism* is used very broadly, so in this case it may be perfectly fine for your child. Keep in mind that you need to evaluate this in the context of your child's particular needs and that you can always ask to discuss the label and change or drop it.

10. **My daughter had a diagnosis of PDD-NOS when she was younger. Now she is 10 years old and has been mainstreamed for 2 years. She no longer needs special services. Can we now drop the label that used to get her special services that she no longer needs?**

    Yes, you can indeed drop the label. If it turns out, for whatever reason, that in the future she needs some special services, you can revisit this issue with the school.

11. **One of the teacher's in my child's Sunday school made some comment to me about parents causing autism. She said she was taught that "refrigerator mothers" did this. Is there any truth to this?**

    No, there is no truth to it. In the 1950s, there was some thought that perhaps parental care might cause autism, but it has become apparent that this is not true. A whole generation of professionals (and parents) heard about this, and sometimes you will still find someone who was taught this. Give your friend a copy of this book or another recent one on autism!

CHAPTER 2

What Causes Autism?

The earliest descriptions and discussions of autism, not surprisingly, focused on today what we might call ''classical'' autism—that is, autism as strictly defined and much less on what we would now think of as autism spectrum disorders or the broader autism phenotype, as it is sometimes called. As we mentioned in the first chapter, Kanner's first paper on autism was very influential in several different ways although some of his first observations have been modified over time. His description of autism was unusually clear about what he saw as the central features present in autism (problems in social interaction and unusual responses to the environment). He also was clear in suggesting that autism was congenital; that is, children were born with it although we now know that sometimes children seem to develop autism in the first years of life. Kanner speculated that autism was not associated with intellectual disability (mental retardation) because children did well on some parts of intelligence quotient (IQ) tests. Some aspects of his report misled people, for example, into thinking parents of children with autism might somehow cause the disorder. The early (and mistaken) notion that autism was more common in families where parents were more successful indirectly contributed to a very unfortunate development in the 1950s: blaming the parent (usually the mother) for the child's troubles. Bruno Bettleheim of the University of Chicago advocated removing children from the home in an attempt to address what he saw as the fundamental problem. The idea that parents somehow caused autism damaged a generation of parents who felt responsible for their child's difficulties. However, beginning in the 1960s, and particularly in the 1970s, research began to show that autism was a brain-based disorder.

As children with autism were followed over time, it was clear that many of them—perhaps 20% or so—would develop seizures. Other children exhibited unusual features on neurological examination such as persistent ''primitive'' **reflexes** (which are present at birth but typically disappear in children after a few months). Some studies reported that children with autism were more likely to

have had complications either before or during birth. Still other studies reported associations of autism with a number of medical conditions that were known to affect brain development. Most importantly, it became clear that autism has a strong **genetic** aspect. Although we still don't know the absolute cause of autism, the best evidence suggests that autism is a brain-based disorder with a very strong genetic component. Although the exact genetic cause (or causes) is not known, we are much closer to finding genes than we were even a few years ago.

Genetic Causes of Autism

In the 1970s, an article written by some prominent **geneticists** suggested that there was no genetic contribution in autism. However, autism was relatively rare and the data was very limited. Shortly thereafter, a very important paper by Susan Folstein and Michael Rutter (1977) appeared, which suggested that rates of autism in identical (or monozygotic) twins were much higher than rates in same-sex fraternal (or dizygotic) twins.[1] Identical twins have identical genes, while fraternal twins share only some genes. The implication of this finding was that there was potentially a very strong genetic contribution in autism. A number of studies have now shown that this is the case (see Rutter, 2005 for a detailed discussion). Several different additional findings have emerged.

As scientists began to look into the issue of the genetics of autism, it became apparent that rates of autism were increased in the brothers and sisters of children with autism. Rates reported vary between 1 in 10 and 1 in 50. This does not seem like a very high rate *unless* one realizes that the rate of classical or strictly defined autism in the general population is between 1 in 800 and 1,000 or so and that, although autism is by no means common in siblings of autistic children the rate is clearly increased— relative to the general population.

GENETICS OF AUTISM

Strong role for genetic factors suggested by:

- High rates of concordance in identical twins (if one twin has it, the other one is very likely to have it).
- Increased risk for autism in siblings (2–10%) (this is significantly greater than the population rate).

[1]Monozygotic or identical twins are always same sex of course but genetic studies of fraternal twins with autism have generally focused on same-sex twin pairs because of the gender difference in autism, or, put another way, these studies have typically not used boy-girl fraternal twin pairs because of this sex difference.

What genes are involved?

- It appears that multiple genes contribute to autism.
- Attempts are under way to identify these genes.

What happens once genes are identified?

- It will be possible to develop animal models.
- We will better understand how the genes work in the brain.
- There may be implications for diagnosis and screening.

Other work also began to look at associated problems in siblings and suggested that even when siblings did not have autism, they did seem to have an increase in other problems, including language and learning difficulties. It still is not exactly clear what is inherited in autism. It is possible that what is inherited is a more general predisposition to difficulties rather than to autism as such. Recent work on family members also suggests that there may be higher rates of mood and anxiety problems in family members as well as, perhaps, more social difficulties.

Although research has increasingly highlighted the importance of genetic factors in autism, final answers are not yet in. The genetics of autism is not straightforward or very simple, and it appears that multiple genes are probably involved; estimates of the number of genes range from 4 to 20 or even more. To make life more complicated, it may also be the case that not all forms of autism have the same genetic basis but might come about in other ways; for example, there might be a specific problem at the moment of conception when some genetic material might be lost or a genetic change (**mutation**) might occur. It might be that other things are involved, for example early birth difficulties might interact with a genetic predisposition to cause autism. Major efforts are now under way to identify potential genes in autism. Genes that may be involved (known as ''candidate genes'') are presently being investigated. It seems likely that some genetic cause (or causes) of autism will be identified over the next few years.

Seizure Disorders and Electroencephalographic Abnormalities

One of the important things that helped doctors realize parents weren't to blame for autism was an increasing awareness of the higher-than-expected risk autistic children had for developing seizures. Seizure disorders (also referred to as **epilepsy** or **convulsions**) are a group of conditions that result from abnormal electrical activity in the brain. The symptoms of seizure disorders are quite varied. They can range from brief episodes where the child seems to ''tune out'' to

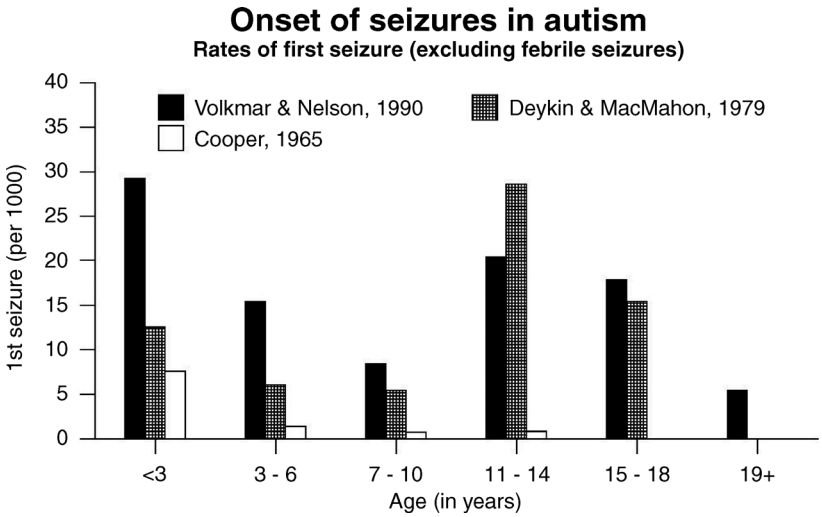

FIGURE 2.1 **Rates of first seizure in samples of individuals with autism**

(Volkmar & Nelson, 1990; Deykin, & MacMahon, 1979) and a normative British sample (Cooper, 1965).

much more obvious convulsions where the child falls to the ground, loses consciousness, and has alternating periods of muscle contraction and relaxation. There are many different kinds of epilepsy (see Chapter 12).

One of the ways doctors look for seizure activity is through the **electroencephalogram** (or **EEG**), which measures electrical activity in the brain. Both early and more recent studies suggest that as many of 50% of individuals with autism have abnormalities in their EEGs; findings on the EEG are diverse and not specific to autism, but the higher rates of abnormality are, of course, suggestive of some basic problem with the way the brain is "wired." In the "normal" population of children, rates of first seizure are highest around the time of birth and then greatly decrease over time. Figure 2.1 presents information from two studies of children with autism or autism and pervasive developmental disorder not otherwise specified (PDD-NOS), as well as data from a large normative sample of British children. The rates for developing seizures are higher in children with classical autism.

Other Neurological Problems

A number of other neurological problems are observed in autism. Again, these are of many different types; not every child has every problem, and some children will have none. Some children with autism have delays in the development of hand dominance (preference for right or left hand) later than typically developing

children. They can also have general decreases in muscle tone in the body and be somewhat ''floppy'' as babies (technically called **hypotonia**). Sometimes individuals with autism have unusual reflexes; often, these are reflexes that are usually seen only in very young babies but can persist into adulthood in individuals with autism. For example, if the doctor brings a reflex hammer toward the baby's mouth, she may start to suck as if anticipating the bottle or breast; this *visual rooting reflex* is sometimes seen even in adults with autism, whereas in most people it disappears very early in childhood. Other problems may be seen in the way that individuals with autism walk or with their posture.

Neuroanatomy and Brain Imaging Studies

Various methods can be used to study the brain, ranging from actual studies of brain tissues obtained at the time of death (postmortem studies) to studies of the living and active brain through **functional magnetic resonance imaging (fMRI)**. A number of findings deserve mention. Both autopsy and brain imaging studies have suggested that at least some individuals with autism have increased brain size and that this develops in the first year or so of life. Several studies have suggested the possibility that there are some alterations in brain structure, particularly in those parts of the brain that process more emotional or social information (the limbic system) and possibly in the cerebellum. The **cerebellum** is the part of the brain that, among other things, helps coordinate and control movement. One investigator has, in particular, noted specific changes in the cerebellum in individuals with autism; unfortunately, other investigators have generally not been able to find this.

AREAS OF POSSIBLE BRAIN INVOLVEMENT IN AUTISM

Areas of Possible Difficulty	Functions
Prefrontal cerebral cortex	Social thinking
Hypothalamus	Attachment behaviors
Amygdala	Social orientation, emotional learning
Fusiform gyrus	Face recognition
Middle temporal gyrus	Recognition of facial expression
Pulvinar	Emotional relevance

In the last few years, several interesting findings have emerged from studies of functional neuroimaging in autism. A paper from our (Yale) group documented that children with autism and Asperger's syndrome seem to process the information in faces differently in the brain; basically, they use the object processing areas, whereas most of us use a very specialized face processing center in the brain. This

may be one of the reasons that faces don't seem to have the same "specialness" for people with autism that they do for typically developing people. It might also account for an interesting finding in autism—people with autism do just as well identifying faces upside down as opposed to right side up. After about 6 months of age, typically developing babies (and people in general—try this with your driver's license the next time you go through the airport screener!) have real trouble identifying upside-down faces (known as the *facial inversion effect*).

Another, possibly related finding is that higher functioning individuals with autism—and, for that matter, many babies with autism—tend to look at mouths rather than eyes and the upper parts of the face when watching very intense social interactions. Our group originally demonstrated this by having very able people with autism and typically developing viewers watch clips from the movie classic *Who's Afraid of Virginia Woolf* and discovered great differences in how the very cognitively able viewers with autism watched the movie. In contrast to typically developing viewers, who spend more time looking at the eyes and top half of the face, the viewers with autism tended to focus on mouths and objects (the latter usually more or less totally extraneous to the plot of the movie). This is demonstrated visually in Figures 2.2 and 2.3. Figure 2.2 shows the differences in visual scanning of a viewer with autism (bottom line), who focuses on mouths (and

FIGURE 2.2 **Visual focus of an autistic man and a normal comparison subject showing a film clip of a conversation.**

Typically developing person (top line) goes back and forth between the eyes in viewing a social scene; a high-functioning person with autism goes back and forth between the mouths of the speakers.

Reprinted, with permission, from Klin, A., Jones, W., Schultz, R., Volkmar, F., & Cohen, D. (2002). Defining and quantifying the social phenotype in autism. *American Journal of Psychiatry, 159,* 895–908.

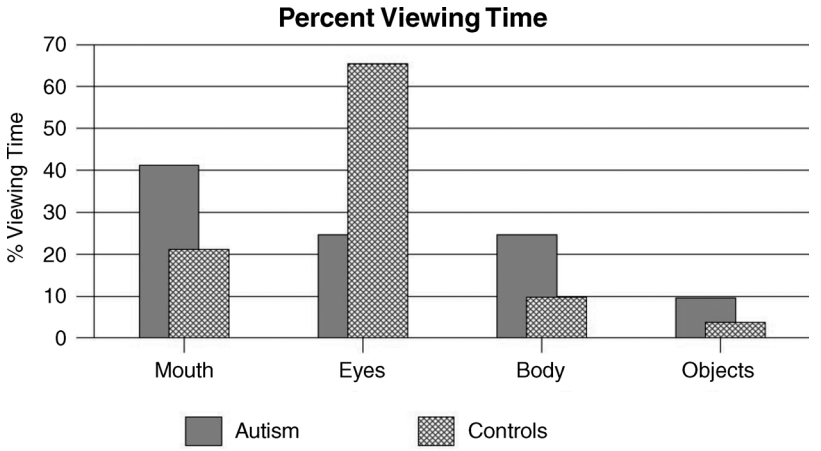

FIGURE 2.3 **Percent viewing time spent focused on mouth, eye, object, and body regions in viewers with autism and typically developing persons.**

All differences are significant.

Data adapted, with permission, from A. Klin, W. Jones, R. Schultz, F. Volkmar, & D. Cohen. (2002). Visual fixation patterns during viewing naturalistic social situations as predictors of social competence in individuals with autism. *Archives of General Psychiatry, 39*(9), 809–816.

then only on the person speaking), versus the typical viewer, who focuses on the eyes. Figure 2.3 shows the data for groups of cases—there is little overlap of the groups in terms of where they watch. Given that most (maybe about 90%) of the important social information is conveyed in the top half of the face, it is probably not such a surprise that viewers with autism are missing most of the relevant action. These differences in visual tracking seem to develop very early (see Chapter 7) and may be a reflection of different brain mechanisms used to process social information. This is a very active area of research right now.

Neurochemistry

Nerve cells use different kinds of chemicals to communicate with each other. A number of these systems have been studied in autism, and there is some suggestion of alterations in these systems. Most of the work has centered on the chemical **serotonin** (also sometimes referred to as 5-HT or 5-hydroxytryptamine). A number of studies have shown that levels of serotonin in the blood are often increased in individuals with autism. Unfortunately, the relationship between blood levels and brain levels of this chemical are not always clear. Other studies have focused on the chemical **dopamine**, which is involved in parts of the brain

that control movement and is part of a broader system that relates to levels of alertness and what is technically called *arousal*. Many of the drugs used to treat symptoms of autism affect these chemicals (see Chapter 15).

NEUROCHEMISTRY OF AUTISM

Neural transmitter	Function, relevance to autism
Serotonin	Regulates sleep, mood, body temperature High levels in blood of many individuals with autism Affected by some medications
Dopamine	Control of motor functions One class of medicines used in autism (neuroleptics) block dopamine function
Norepinephrine	Involved in states of arousal, stress response, memory, and anxiety; affected by some medications

Risks During Pregnancy and Childbirth

Could autism be caused by problems during pregnancy, labor, and delivery? A number of studies have looked at this question. Generally, they have employed some rating scale that looks at the degree of risk during the pregnancy and/or during labor and delivery. Early studies seemed to show that there was an increased risk based on the use of these rating scales. Factors that seemed to be associated with increased risk for autism included older age in the mother, prematurity, and some other problems during labor and delivery. Several studies have also suggested that doctors or nurses may be more likely to notice something wrong with the newborn, even if it is very minor. This suggests an important point and a major problem in understanding whether problems during pregnancy or labor and delivery might cause autism; that is, it would be reasonable to assume that if there were something wrong with the child from the moment of conception that was picked up at birth, we might be seeing problems at birth that may result from some vulnerability in the child. Thus, it would be just as reasonable to assume that problems in the child cause difficulties in the pregnancy. The growing body of work on genetic factors in autism, which is discussed shortly, would be most consistent with this idea.

At the same time, it is reasonably clear that horrendous difficulties during labor and delivery, particularly when associated with severe fetal distress, won't *help* any child and have the potential to further cause trouble for a child who was going to have autism.

Environmental Causes of Autism

Interest in the possibility that environmental factors contribute to autism stems from several sources, including reports of ''cluster'' cases, an assumption that the rate of autism has risen over time, and associations with potential environmental toxins like mercury (or thimerosal in vaccines). It is indeed clear that even in identical twins, while the rate of concordance for autism is high, it is not 100%.

However, as we discussed in Chapter 1, it is not so clear that the rate of autism is increasing. It is reasonably clear that we are doing a better job of finding cases, that there is much more awareness of autism, and that we've changed the ways we diagnose autism and have expanded the number of children diagnosed. As we discuss in Chapter 10, the vast majority of serious scientific studies have not supported the idea that vaccines (or thimerosal) cause autism. Furthermore, some of the evidence proposed for environmental factors is based on case reports, which are often difficult to interpret. For example, there was an early impression that autism was associated with congenital rubella infection, but, over time, these usually very delayed children looked less and less autistic. At present, there is not particularly strong evidence for specific environmental etiologies, although clearly more work is needed in this area. Some good summaries of work in this area are included in the reading list at the end of this chapter.

Medical Conditions and Autism

It took many years before people considered the possibility that autism was associated with some medical conditions, such as seizures. As autism came to be better recognized, it became associated with a number of other medical conditions, and there was interest in the possibility that some of these conditions *caused* autism. However, much of this work was based on **case reports**, wherein a doctor sends a letter or a short paper to a professional journal, which reports that autism is associated with_____syndrome (you can fill in the blank with essentially any known medical condition). Case reports have some value but also have many limitations since there is a bias for only positive reports to be published (similar to the situation with the regular media!). The issue is not whether you *ever* see autism and condition X, but whether in larger groups of individuals the frequency of condition X is significantly greater in autism than you would expect, given how common condition X is in the general population.

Another problem relates to the diagnosis of autism in the first place. Some researchers take a very broad view of autism; others, a more narrow one. If a broad view is taken, estimates of the rate of autism will naturally tend to be higher and there will be the impression that autism is more likely associated with other medical conditions. In other words, if a broader definition of autism

is used, there will be more diagnoses of autism among people with severe and profound intellectual disability, in part because levels of repetitive movements and unusual behaviors are higher in this group of cases. Also in this group of cases, with lower IQs, about half the time there is an associated medical condition, and thus a condition that might contribute to the person's handicap is much more likely to be found, regardless of whether they have autism! Two rather different views emerge from the published research on medical conditions associated with autism. If a very broad view of autism is taken, perhaps one-third of cases of autism might be related to some condition; however, if a narrow view is taken, probably only 10% of cases are.

Various conditions have been identified as possibly being associated with autism, including **phenylketonuria**, **congenital rubella**, **tuberous sclerosis**, and **fragile X syndrome**. However, careful research has led us to rethink how strong these relationships are, and at present the strongest associations are with fragile X and tuberous sclerosis.

Fragile X Syndrome

Fragile X syndrome is a common syndrome associated with intellectual disability and, sometimes, with autism. It is probably second only to Down syndrome as the most identified genetic source of mental retardation. Fragile X syndrome particularly affects boys and has sometimes been associated with autism. All of us have 23 pairs of **chromosomes**. Boys have an X chromosome (from their mothers) and a Y chromosome (from their fathers). It is called fragile X syndrome because the X chromosome was noted to sometimes break or be ''fragile'' when examined. Because boys have only one X chromosome, they are more likely to have the disorder (i.e., there is no extra X to make up the difference). In girls, who have two X chromosomes, the disorder may be expressed in a somewhat milder form. Fragile X is one of the more common causes of intellectual disability/mental retardation, perhaps affecting 1 in 800 to 1,000 children.

Associated problems in fragile X syndrome include mild intellectual disability/mental retardation (although sometimes IQ is in the normal range). In addition, language problems, attentional difficulties, and symptoms suggestive of autism (problems with eye contact and self-stimulatory behaviors) may be observed. Boys with the disorder may have some unusual body features, such as large ears and genitals; the face may be long and narrow, and the palate (the roof of the mouth) may be unusually high and arched. Motor and learning problems are relatively common. Low muscle tone as well as dental and eye problems may be observed. Sometimes individuals with this condition have seizures.

FRAGILE X SYNDROME

- Symptoms: social anxiety, "autistic-like" symptoms, and sometimes autism.
- About 1–2% of individuals with autism have fragile X.
- A simple blood test can be used to determine whether fragile X is present.
- Syndrome can be seen in boys and girls.
- The genetic basis of this condition has been well described.

Early reports suggested a very strong association between autism and fragile X syndrome, with claims that as many as 60% of individuals with autism had fragile X. There was much optimism that a genetic cause of autism had been found. However, subsequent (and better) studies have suggested that the association between fragile X syndrome and autism is not nearly as strong as it first appeared. In the early studies there was not careful attention to appropriate controls or to diagnosis of autism. Recent research indicates that between 1% and 2% of individuals with autism have fragile X; this rate is not much different from what would be expected from any sample of children with mild intellectual disability—that about 1% of individuals with fragile X have autism. Thus, although it remains important to consider testing for fragile X, the rate of the condition in autism is relatively small and accounts for only a very small subgroup of children with autism.

At the present time, the main impact of diagnosing fragile X relates to the implications for genetic counseling of parents and sisters of affected individuals; that is, the treatment of the child with autism and fragile X is no different than that for the autistic child without fragile X. However, for parents who know that they have a risk for subsequent children with fragile X, prenatal testing and termination of pregnancy (if that is an option for the parents) is available. In the past, the diagnosis of fragile X syndrome was made on the basis of an actual examination of the child's chromosomes, obtained through a blood sample and grown in a laboratory. This time-consuming and costly procedure has now been replaced in many centers by a more direct DNA test for the fragile X abnormality. The genetic cause of fragile X has now been identified.

Tuberous Sclerosis

Tuberous sclerosis affects about 1 in 10,000 people and, although rare, has been noted to be significantly associated with autism. It is seen equally as frequently in boys and in girls. Symptoms include the growth of unusual tissue or benign tumors in the skin, eye, brain, and other organs. Over half of infants with the disorder will have white patches on their skin at birth. The tubers may be seen in

the brain and can be detected by **computed tomography (CT)** or **magnetic resonance imaging (MRI)** scan. The tumors associated with this disorder are often seen in the preschool years and may increase in frequency during **puberty**. These growths are ''benign'' in the sense that, unlike cancer, they do not spread, but their effect on growth and development can be very serious. Individuals with this condition often have **developmental delay** and intellectual disability and seizures.

The disorder is inherited as an *autosomal dominant* trait, meaning that it is on the autosomes (i.e., not on the sex chromosomes) and that if you get one copy of this gene from either parent, you are likely to have the disorder. A gene for the disorder has been located on chromosome 9.

Although the effects of the disorder can be severe, the degree of severity in tuberous sclerosis is variable. Children with tuberous sclerosis may have speech delays and learning problems. They often have motor problems as well. Sometimes the first symptoms are seen in infancy or early childhood, often with the onset of seizures. Between 50% and 60% of affected individuals show intellectual disability, and about 80% have seizures. Sometimes the findings include a specific abnormality in brain wave (EEG) testing. The seizures may include a specific kind of muscle spasm (myoclonic jerks). Sometimes individuals seem to be much less severely affected; it seems likely that this happens in some special circumstances, for example, as a result of spontaneous genetic change not inherited from the parents.

Interestingly, early reports on tuberous sclerosis appeared in the 1930s and described some problems suggestive of autism (which was not described until the 1940s!). These problems included stereotyped movement, abnormal speech, and social problems. Hyperactivity, aggression, and other behavior difficulties have also been reported.

In studies of individuals with autism, about 1–2% also have tuberous sclerosis; this figure is higher only if individuals with autism *and* seizures are included (about 8–12% of such cases may have tuberous sclerosis). But not every child with tuberous sclerosis has autism. The ratio of boys to girls in autism associated with tuberous sclerosis is about the same; this is in contrast to autism in general, where the rate of autism is clearly several times higher in boys than in girls. Sometimes tuberous sclerosis is not associated with intellectual disability; studies of this small subgroup do not seem to suggest high rates of autism or similar problems, but the final answers are not yet in. Promising work on the genetic causes of tuberous sclerosis is now under way.

Disorders of Metabolism: Phenylketonuria (PKU)

PKU is caused by a problem in the body's use of the amino acid phenylalanine. As a result, levels of this amino acid build up in the body and eventually are

excreted in the urine. This disorder is rather rare, affecting about 1 in 10,000 babies. If it is not treated, PKU can cause severe intellectual disability, growth problems, and seizures. Although the baby may otherwise appear normal at birth, symptoms gradually develop, including problems in feeding and development. Fortunately, with the recognition of the cause of the disorder, doctors realized that the condition could be treated with a special diet that eliminates phenylalanine. This is one of a handful of dietary treatments medically proven to have a major role in preventing/treating developmental problems. PKU is now screened for at birth in this country, since prompt treatment allows children with PKU to have normal and productive lives.

Early papers suggested that PKU was a risk factor for autism. However, more recent research has questioned this view. Well-controlled studies do not seem to suggest that there are higher-than-expected rates of PKU or other disorders of metabolism in autism. It seems likely that early reports of such associations probably equated ''autistic features'' (usually meaning stereotyped, self-stimulatory behaviors) with autism. It is still appropriate to consider screening children with severe developmental difficulties for inborn errors of metabolism, but there is not a clear relationship of these disorders to autism.

Congenital Infections

There have been some reports associating autism with infections either before or at the time of birth or shortly thereafter. The kinds of infections for which this have been claimed are quite varied and include congenital rubella, cytomegalovirus, herpes simplex, and human immunodeficiency virus (HIV—the AIDS virus). A few papers have also reported that there might be some association of autism with the time of year when children are born that might suggest some fluctuation in association with the prevalence of other infections. However, other studies have not seen such associations. Probably the most interest in terms of infection has centered around congenital rubella.

Congenital rubella occurs when a baby still in its mother's womb is infected with the rubella (German measles) virus. Women who have not had rubella or who have not had the immunization for it are at very high risk for having a baby with congenital rubella if they develop rubella while they are pregnant. The risk is greatest during the first 8 weeks or so of the pregnancy (a time when sometimes women may not realize they are pregnant).

The virus often does severe damage to the developing baby. The baby may be born with problems in the heart, eyes, and ears. The head may be small, and there may be problems with other parts of the body. Hearing loss may develop; deafness and blindness are relatively common. Intellectual disability and various behavior problems may be observed. While a few children do not have symptoms, most

do. Fortunately, greater awareness of the seriousness of this condition and the development of a vaccine have reduced the frequency of this condition. The vaccination of young children has been very helpful in this effort.

Early reports on congenital rubella suggested that these children often seemed to have autism. As we mentioned earlier in the chapter, there were a number of issues with this conclusion since these children had multiple problems—they were often deaf or had impaired vision and severe learning difficulties so being sure of the diagnosis was complicated. As with other conditions, the presence of **autistic-like** features was taken as suggestive of autism; however, follow-up studies have shown that over time the social and other problems of these children seem to improve in ways that would not be typical of autism.

Psychological Models of Autism

Following the error of the early ''blame the parent'' notion, speculation about how autism might be understood through psychology was held back in some important ways. Over the last two decades, new theoretical models have been proposed that try to understand the developmental and behavioral aspects of autism from the point of view of psychological development; it must be emphasized that this is an attempt to understand brain-based difficulties and *not* to blame the parents. These attempts are of some interest in terms of research and may, perhaps, lead to some treatment advances. It is important to realize that several rather different approaches have been used. One attempts to view the social problems in autism as one of many different difficulties caused by the same factor (or factors). The other view emphasizes the social difficulties as primary in some basic way, that is, as leading to other problems. These all have their pros and cons and none has, at least as yet, emerged as the ''winner.'' At present, they all have something to offer in terms of alternative models of how we might understand autism.

The **Theory of Mind** approach has emphasized the idea that there is a basic problem for children with autism in empathizing with others, that is, having a ''theory of mind,'' or theory of what motivations, intention, and so on, impact on the behavior of others. This approach, first proposed by Simon Baron-Cohen (see reading list) has been remarkably productive in terms of research. The simplicity and elegance of this theory have added to its attractiveness. There are, however, two problems with this model. One is that the severe difficulties in social interaction impact behaviors seen in very, very young children—children of a few weeks of age. This is a time well before the ability to ''put yourself into the other's place'' has really developed. Another problem is that many higher functioning individuals on the autism spectrum can do ''theory of mind'' tasks just fine, and yet these individuals are still very socially disabled.

Another approach, termed the **executive dysfunction hypothesis**, emphasizes deficits in ''executive functions'' (a topic we discuss in greater detail in Chapter 6). The notion of executive functions refers, basically, to the whole range of abilities involved in planning and organization. For example, seeing the multiple steps involved in a complicated task, plotting a solution in terms of getting to the desired result, keeping the desired result in mind, and being able to work out alternatives when this is needed (Pennington & Ozonoff, 1996). Within this view, autism is related to difficulties in dealing with change and a tendency to engage in repetitive behavior and **perseveration** as well as to problems in developing planning and problem-solving abilities due to a lack of coordinated reasoning and ongoing adjustment to feedback (Ozonoff, 1997). As we discuss later in this book, there is no question that children with autism spectrum disorders often have severe problems in this area. From the point of view of a more general theory, however, there are some difficulties. Probably most importantly, difficulties in this area are not unique and specific to autism; that is, children with attention deficit hyperactivity disorder also have problems with organization (but don't have social troubles of the same type seen in autism).

A somewhat different theory proposes that the difficulties in autism relate to ''**weak central coherence**.'' The idea here is that people with autism have trouble getting the ''big picture'' (Happé, Briskman, & Frith, 2001); they don't see the interconnections of things—a ''not seeing the forest for the trees'' problem. This theory would account for some of the people with autism who are gifted in one area but very deficient in another area. Although very attractive in many ways, the experimental evidence has been somewhat weak and contradictory. Other approaches, for example, Klin and colleagues (2003) focus more on the social difficulties being a primary cause of autism, with many of the symptoms arising from the limited interest in people and the negative consequences of brain and psychological development.

Understanding the Causes of PDDs Other than Autism

Our understanding of the causes of PDDs other than autism is not generally as far advanced as in autism, with the major exception of Rett's disorder. Again, we understand that all these conditions have a basis in problems in the brain. This is suggested by such things as rates of seizure disorder and, occasionally, other abnormalities as well. The role of genetic factors in Rett's syndrome is now clearly established, as a gene has been found to be involved in most cases (see Chapter 13). Compared to Rett's syndrome, childhood disintegrative disorder is apparently less common and has been even less frequently studied. For many years doctors presumed that there was some specific medical process that could always

be identified to explain why children developed normally for several years and then had a major deterioration. It is clear that this is the exception rather than the rule. Occasionally, such a process is identified that is similar, in some ways, to the dementias of adults (such as Alzheimer's disease, where there is progressive loss of functioning). Interestingly, however, in CDD behavior and developmental skills usually deteriorate and then stay at the same, relatively low, level. This kind of plateau is not usually observed when a progressive medical condition is present. As in autism, the involvement of the brain is suggested by the high rates of EEG abnormality and seizure disorder. Information on brain structure and functioning is very limited, although research on this aspect of CDD is now under way. It is possible that the condition might develop in several different ways.

CHILDHOOD DISINTEGRATIVE DISORDER

Causes of Childhood Disintegrative Disorder

- Early impression of possible psychological causes seems wrong.
- The distinctive and unusual pattern of onset suggests some specific disease process.
- Most of the time, despite intensive searching, no specific medical cause is found.
- Occasionally the condition is associated with some neurological disorder similar in some ways to adult dementia, but this is not usually the case.

In Asperger's syndrome (AS) there have been several reports of associated abnormalities, but these are mostly based on reports of single cases rather than group studies. One interesting finding has been that Asperger's is frequently associated with **nonverbal learning disability** (NLD) and the difficulties in AS have been taken by some to suggest difficulties in the right part of the brain (in contrast to autism, where the presence of **language** problems has often been taken to suggest problems in the left part of the brain). Although research on the issue of genetic contributions in Asperger's is not as well advanced as that in autism there is already some evidence for a strong genetic component with high rates of social difficulty in members of the immediate family.

ASPERGER'S DISORDER

Causes of Asperger's Disorder

- Genetics:
  - Asperger commented on high rate of similar conditions in fathers.

  - Recent research does suggest higher rates of social problems in male relatives.
  - Female relatives tend to have higher rates of anxiety and depression.
  - It is possible that this is even more strongly genetic than autism.
- Brain functioning:
  - Association with Nonverbal Learning Disability Profile.
  - Suggestion that problems are more likely in the right cerebral cortex.

Research on the causes of PDD-NOS is the least advanced of all the PDD conditions. There is a strong suggestion of a possible genetic component, since many individuals with autism have relatives with language, learning, or social difficulties. It is possible that what we now see as PDD-NOS may, some day, be identified as a variant of autism—for example, one which comes about when some, but not all, the **genes** that cause autism are present. It is also likely that there really may be important distinctions within the broad group of PDD-NOS cases. For example, some cases may have a more genetic basis and may be more closely related to autism; others may have a different basis and might be close to other conditions (e.g., language or attentional problems). It is also possible that some combination of factors might cause PDD-NOS.

Summary

We have now come to appreciate that genetic factors are very much involved in autism. In some ways this has been a surprise, since early work did not seem to suggest a strong genetic basis. This early work was very limited, and only when the first studies of twins were done was the possible genetic basis of autism recognized. Studies of twins showed that if the twins were identical (with exactly the same genetic makeup) and if one had autism, there was a very high chance the other twin would as well; if the twins were fraternal (not exactly the same genetic makeup but sharing as many genes as any siblings would), the rate was much lower. As time went on, it also became clear that a range of other problems—in language and learning and social interaction—might be inherited. Active research around the world is being conducted to look for the genes that cause autism.

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QUESTIONS AND ANSWERS

1. **I have one child with autism and am thinking about having a second. What are the chances my second child could have autism?**

   In general, having had one child with autism increases your risk of having another by probably between 2% and 10%. This does not sound like much of a risk until you think that roughly 1 child in a 800 to 1,000 in the general population has autism, which means your chances are substantially increased. We have seen families with three and four children with autism. Keep in mind that this is a question we can answer in general terms—for a specific answer relevant to you, speak with a genetic counselor, who can take into account all the special factors in your situation, such as family history.

2. **Are a brain scan and an EEG always necessary in evaluating a child with autism?**

   In general without a specific clinical reason to do it, the likelihood of finding something is small. If there are specific clinical reasons to do these tests, for example, if you suspect seizures or if the child's history and behavior are highly unusual, then they should be done.

3. **Are there any lab tests that diagnose autism?**

   At present, the answer is no. When genes for autism are found, there may be some such tests in the future. At the moment, the only additional lab test that makes sense is the test for fragile X (a blood test). Special genetic tests are now able to look for missing genes. Other tests may be needed, given the child's history and examination.

4. **Can autism be diagnosed from an EEG?**

   No. Autism is diagnosed based on history and clinical examination. The EEG is useful in the diagnosis of seizure disorders, which are sometimes associated with autism.

5. **What will it mean if genes are found for autism?**

   A number of things will have to happen before the findings can translate into new treatments. These include discovering how the gene works and how it operates in development and in the brain, and development of an animal model. Developing an animal model is important, since this would help with understanding what is happening in the brain and give us more potential for testing possible treatments. There *may* be some important implications quickly for screening. There also *may* be some possibility of understanding the broader spectrum of autism and related conditions. Keep in mind that this is a very active area of research and that the answer to this question may change dramatically in the next several years.

CHAPTER 3

Getting a Diagnosis

A diagnosis is a label that serves as a shorthand way for professionals and others to communicate with each other very quickly. You might think getting a diagnosis would be a simple business, but, unfortunately, for autism there is not (at least at the time this chapter is written and probably not for some time) a simple blood or laboratory test to determine who is autistic. There now are blood tests for a few conditions that are associated with autism, such as fragile X syndrome and Rett's disorder. In autism, we have to rely on the judgment of (hopefully) experienced clinicians, who may do any of several things to help them arrive at a diagnosis. Usually, getting a diagnosis is one part of getting an **assessment**. The goals of the assessment often include things in addition to the diagnosis, for example, clarifying the child's strengths and weaknesses, challenges the child has in learning, and so forth. In this chapter, we review some important aspects of getting a diagnosis and doing an assessment. In subsequent chapters, we'll talk in more detail about some of the assessment **instruments**, for example, as they are relevant to younger children (Chapter 7), school-age children (Chapter 8), or adolescents and adults (Chapter 9). We also devote some discussion to testing and assessment issues in discussing building skills (Chapter 6).

You may well ask, ''Does a diagnosis make a difference?'' For some purposes (e.g., for getting early intervention services), providers may not be so concerned about a precise diagnosis but rather about the child's need for services. In part, this reflects an awareness, particularly in very young children, that diagnoses can sometimes be hard to make. However, a diagnosis can be helpful. A diagnosis may help to ''frame'' the child's needs, for example, for educational and speech and language services. It is important to realize that diagnosis helps us to know only the general kinds of problems or issues presented; it does not tell us a lot about the specifics of an individual child because there is such a range of abilities and needs among children with autism spectrum disorders. Sometimes parents want the label (for educational purposes) that may get their child the most services.

As with many other things, life is lived in the details, and it is the specifics about autism that are very important. For example, is the child verbal or nonverbal? Does he have any motor difficulties? How socially related is the child? Does he have behavior problems that interfere with programming for him? These issues are particularly important for autism and related disorders, given the wide range of disabilities we see in children with these conditions. Since we don't yet know the exact cause of autism, we presently rely on observation and history to make the diagnosis. Various guidelines, rating scales, and checklists have been developed, and they may help in making a diagnosis, but they never replace the importance of a skilled clinician's observations and assessment.

This chapter covers aspects related to getting a diagnosis of autism and autism spectrum disorders (ASDs). We discuss some of the more common ways that parents and professionals become concerned about the child and the kinds of behaviors that very young children with autism exhibit. We discuss the uses and limitations of diagnosis, as well as what a good diagnostic **evaluation** consists of. We also address some of the more frequent sources of disagreement on diagnosis and, finally, what should happen after a diagnosis is made. As with other chapters in this book, it is important to realize that when we give examples, not every child will behave or develop in the same way and that the examples are just that. This chapter will be of greatest interest to parents of younger children, as well as to parents who are trying to understand what goes into an assessment of their child. In the next chapters, we talk about the critical issue of translating the diagnostic assessment into services, and the chapters following that discuss age-related aspects of ASDs.

First Concerns

There are many ways that parents become aware that something is wrong with how their child is developing:

- Sometimes parents gradually become aware that there is something about their child that is different—maybe she seems less interested in her parents than they expect or has some unusual reaction to sounds or noise.
- Other parents trace their concerns to a very specific event, such as seeing their child with other children of about the same age.
- Occasionally, a grandparent or friend, or sometimes a day care provider or the child's doctor, may mention that they are worried about how the child is doing.
- Sometimes parents will say that, as they think about it, maybe there were some signs of trouble even earlier than they first thought.

- Sometimes parents will say that their child was, as an infant, ''too good,'' making few demands on them.
- At other times parents will tell us that the child had difficulties from shortly after birth, for example, being difficult to console or being very demanding.
- Somewhat less commonly, parents will feel as if their child was doing reasonably well until, say, 18 months of age, when either he lost ground or seemed to stop moving forward developmentally.
- Parents who have already had one child may make comparisons and realize that their new child is developing in a very different way.

Probably the most common cause of concern is speech delay. Concerns that the child may be deaf are also very frequent, although usually, unlike deaf children, the child with autism seems to respond to some sounds. The child with autism may use pointing to get things or may pull a parent by the hand (often with no or limited eye contact) to get something, but does not seem interested in sharing attention. For example, he rarely points to show things to parents. Some parents, especially, first-time parents, may not have realized that there was anything unusual about this behavior and do not ask their doctor about it until 18 or 20 months, when their child is still not speaking.

In other cases, parents may be worried about their child's development even earlier. When this happens, it is often the child's lack of social relatedness, that is, his lack of interest in parents and other people, that causes concern. In our experience, parents are more likely to be concerned about this when they have had experience with children. Occasionally, parents will be worried because their child does not seem to enjoy contact with them, but she is interested in odd or unusual aspects of the environment, such as rocking by herself in a corner. Or parents may be concerned that their child has chosen an unusual transitional object to comfort herself. Rather than choosing something soft (and typical) such as a blanket or toy, she may choose something hard (and unusual). The child may also be less interested in the actual object than in the ''kind'' of object (e.g., carrying a specific magazine around and taking it to bed with her but not caring which issue of the magazine it is). Sometimes the child will have dreadful and almost ''catastrophic'' responses to certain events in the environment. For example, when the vacuum cleaner is used, he runs upstairs crying and cannot be consoled for hours. Other children may have unusual aversions to food or certain smells. Some of the early warning signs for autism are listed in Table 3.1.

When parents of children with autism are asked when they were first worried about their child, it is clear that many are concerned by a year and most by 16 to