



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,

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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 *resistance 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

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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*,

¹Note 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.

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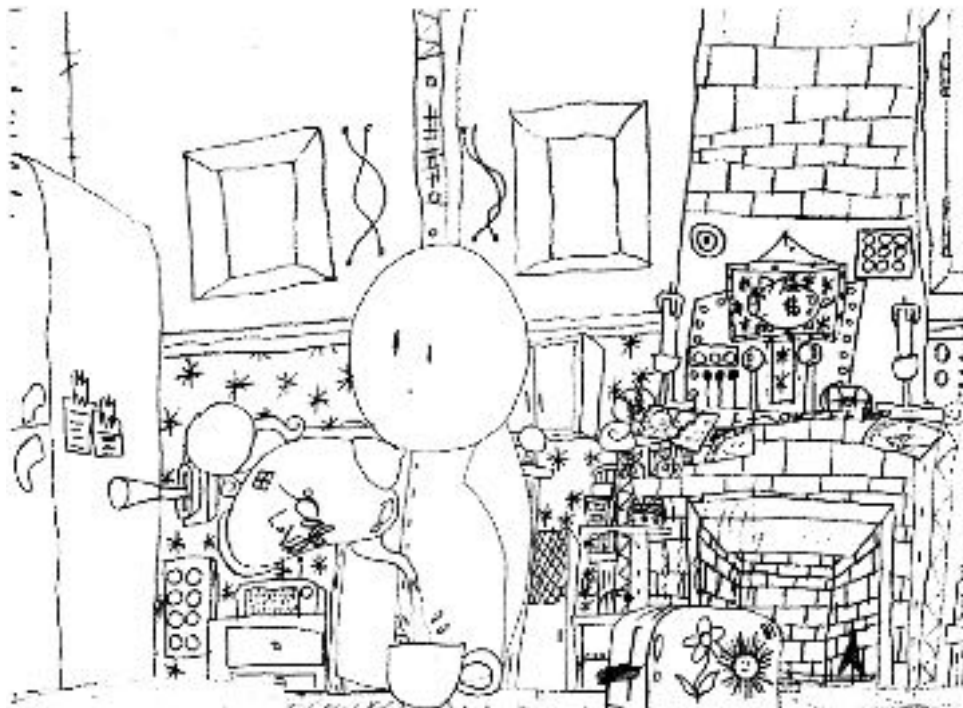


FIGURE 1.1 “Bim gets breakfast in the love kitchen.” Drawing by a child with autism. Bim is the child’s made-up cartoon character. We thank the child and his parents for permission to reproduce. Reprinted, with permission, from F. Volkmar and D. Pauls, Autism. *The Lancet*, 2362, 1134, 2003. Reprinted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

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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 educated 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

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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).

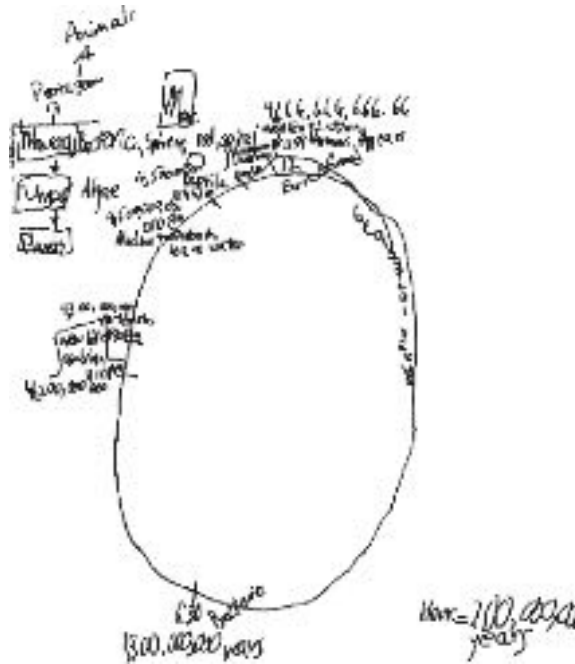


FIGURE 1.2 Autobiographical statement and drawing of area of special interest, 10-year-old boy with Asperger's disorder.

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

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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

Auditory perception
Rote verbal capacities
Verbal memory skills
Verbal output

Weaknesses

Tactile perception
Motor coordination
Visual-spatial skills
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.

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- 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,

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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).

Sex Differences

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

Feature	Autistic Disorder	Asperger's Disorder	Rett's Disorder	Childhood Disintegrative Disorder	Pervasive Developmental Disorder-NOS
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.

Rates of Other Disorders

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

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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.

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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!