The pervasive developmental disorders (PDDs) are a group of conditions that share some general clinical features but likely reflect diverse etiologies. The conditions have their onset in infancy or early childhood and are associated with characteristic patterns of delay and deviance in the development of basic social, communicative, and cognitive skills. Of the various conditions sometimes included within the overarching PDD class, infantile autism, or as it more recently has been termed, “autistic disorder” (American Psychiatric Association, 1994) has been the most intensively studied and will be discussed in greatest detail in this chapter. The convergence of diagnostic criteria in the tenth edition of the International Classification of Disorders (ICD-10; World Health Organization, 1992) and the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV; American Psychiatric Association, 1994) have allowed for international diagnostic consensus in autism and other conditions within the PDD class. In general, the PDDs are associated with some degree of mental retardation, although the pattern of developmental and behavioral features differs from that seen in children with primary mental retardation. The term “pervasive developmental disorder” emphasizes the pervasiveness of difficulties across various domains of development as well as the important developmental aspects of these conditions.

DIAGNOSTIC CONCEPTS

Historical Background

Over the past century, a major point of debate has been the relationship of the severe psychiatric disturbances of childhood to adult psychoses. The concept of childhood “psychosis” became more or less synonymous with the term “childhood schizophrenia”; an assumption based on the severity of the conditions (Volkmar & Cohen, 1988). However, various lines of evidence (e.g., Kolvin, 1971; Rutter, 1972; Volkmar, Cohen, Hoshino, Rende, & Paul, 1988) now suggest that autism differs from schizophrenia in a number of ways, and the term “psychosis” has seemed less appropriate, in general, to apply to children, particularly to younger and lower-functioning children. Other diagnostic concepts have proven more enduring, and still others (e.g., multisystem developmental disorder) await verification. The “nonautistic PDDs,” such as childhood disintegrative disorder (CDD), Rett’s syndrome, and PDD-NOS, have
been less commonly studied than autism. These disorders have been included in both DSM-IV and ICD-10. In many instances, modifications in the original description of the concept have been made based on research.

**Autism**

Of all the diagnostic concepts proposed, Leo Kanner's (1943) description of early infantile autism has proven to be remarkably enduring. He described children with a congenital inability to relate to other people (autism), which was in striking contrast to their relatedness to the inanimate environment. These children also exhibited a number of unusual developmental and behavioral features (e.g., insistence on sameness and resistance to change, stereotyped mannerisms, and when it developed at all, unusual language characterized by echolalia, pronoun reversal, and extreme literalness).

Although Kanner's phenomenological description of the condition has been enduring, certain aspects of his original report were mistaken; for example, he suggested that autism was not associated with mental retardation, that autistic children were more likely to come from more educated families, and that the condition was not associated with other "organic" conditions. It is clear that most autistic children are also mentally retarded; the condition can be observed in association with a host of medical conditions, for example, tuberous sclerosis, congenital rubella, and fragile-X syndrome (Dykens & Volkmar, 1997), although medical conditions are most likely to be associated with more severe impairment (Rutter, Bailey, Bolton, & LeCouteur, 1994). In some cases, children with autism develop seizure disorders, with peak onset in early childhood and adolescence (Volkmar & Nelson, 1990). Other neurological abnormalities are also frequently observed and consistent with an as yet unspecified underlying "organic" etiology (Minshew, Sweeney, & Bailey, 1997).

Kanner's initial observation of deviance in parent–child interaction was taken by some to suggest a role of parental psychopathology in syndrome pathogenesis. It now appears that deviant patterns of parent–child interaction stem primarily from the disturbance in the child and that parental psychopathology is no more frequent in parents of autistic children than those of children with other developmental disorders (DeMyer, Hingtgen, & Jackson, 1981).

**Nonautistic PDDs**

**Childhood Disintegrative Disorder**

Theodor Heller, a Viennese educator, described a condition in which young children who had previously developed normally exhibited marked developmental and behavioral deterioration with only minimal subsequent recovery. In the years subsequent to Heller's description, perhaps 100 cases of the condition have appeared in the world literature (Volkmar, Klin, Marans, & Cohen, 1997). Generally, early development is entirely normal, and the child progresses to the point of using language prior to the onset of a profound developmental regression; once established, the condition behaviorally resembles autism, although the prognosis may be somewhat worse (Volkmar & Cohen, 1989). In some instances, the condition has been reported in association with a specific disease process, for example, a progressive neurological condition. However, it is clear that such a medical condition is observed only in a minority of cases. The validity of CDD apart from autism was supported through comparison of onset date during DSM-IV field trials for autistic disorder (Volkmar & Rutter, 1995). Patients with CDD are more likely to be mute, with profound mental retardation, and have a worse prognosis.

**Asperger's Syndrome**

In 1944, Hans Asperger, an Austrian pediatrician with interest in special education, described four children who had difficulty integrating into groups. Despite preserved intellectual skills, the children showed marked paucity of nonverbal communication involving both gestures and affective tone of voice, poor empathy and a tendency to intellectualize emotions, an inclination to engage long-winded, one-sided, and sometimes pedantic speech; rather formalistic, all-absorbing interests involving unusual topics which dominated their conversation; and motoric clumsiness (Klin, 1997). Unlike Kanner's patients, these children were not as withdrawn or as aloof; they also developed, sometimes precociously, highly grammatical speech, and their difficulties were not diagnosed in the first years of life. Discarding the possibility of a psychogenic origin, Asperger highlighted the familial nature of the condition and even hypothesized that the personality traits were primarily male transmitted. Asperg-
er’s work, originally published in German, became widely known to the English-speaking world only in 1981, when Lorna Wing published a series of cases showing similar symptoms. Since then, several studies have attempted to validate Asperger syndrome (AS) as distinct from autism without mental retardation; analysis of neuropsychological characteristics is consistent with a nonverbal learning disability profile rather than high-functioning autism (Volkmar et al., 1996; Klin, Volkmar, Sparrow, Cicchetti, & Rourke, 1995). Additionally, AS individuals are likely to show interest but little facility in social relationships.

Rett’s Syndrome

Andreas Rett (1966) described the syndrome now commonly referred to as Rett’s syndrome. Although exhibiting some “autistic-like” features, particularly during the preschool years, this syndrome appears to differ from autism in several ways: It is reported only in females, the “autistic-like” phase is relatively brief, it is associated with characteristic motor behaviors (stereotyped hand “washing” or “wringing” movements), abnormalities in gait or trunk movement (e.g., apraxia or gait scoliosis), and breath-holding spells. Early growth and development are normal but followed shortly by developmental regression, relative failure of head growth, loss of acquired speech, and loss of purposeful hand movements. Loss of interest in people and decreased interpersonal contact occurs but eye contact is maintained. This deterioration occurs usually within 1 year (Van Acker, 1997). Eventual mental retardation is even more severe than in autism.

PDD-NOS

The term “PDD-NOS” is used in DSM-IV (American Psychiatric Association, 1994) to describe children with some, but not all, features of autism. These children exhibit patterns of unusual sensitivities and difficulties in social interaction, but not sufficient for a diagnosis of autism. The term “PDD-NOS” is problematic in several respects. The definition is essentially a negative one. The lack of an explicit definition means that it is used rather inconsistently (Towbin, 1997). Research on the condition has been uncommon. More commonly, PDD-NOS has been used for children with better cognitive and communicative skills, and the most common reasons for referral in such cases include concerns of parents about the child’s emotional and social development rather than, as in autism, the failure to develop language.

CLINICAL DESCRIPTION OF AUTISM

Onset and Characteristics of Early Development

Kanner (1943) originally suggested that autism was present from birth. Subsequent research has suggested that the condition is usually apparent within the first year of life but sometimes appears to have its onset within the second or third year of life (Short & Schopler, 1988; Volkmar, Stier, & Cohen, 1985). Age and type of onset have some value in the differential diagnosis of autism, although various extraneous factors may act to delay case detection (e.g., as parental sophistication or denial and level of associated mental retardation in the child).

Most studies of early development in autism rely on parental retrospection or, less frequently, contemporaneous videotapes or movies of the child (Stone, 1997). Although parents often have concerns from the first months of life, they may seek guidance from health professionals only when the child is 16–24 months and still not speaking. The parents may report concern that the child might be deaf, although they paradoxically often note that the child is exquisitely sensitive to certain sounds in the inanimate environment (e.g., the noise of the vacuum cleaner).

The child may not respond differentially to parents but may be particularly attached to a highly unusual object. The young autistic child may also have interest in nonfunctional aspects of objects (e.g., their smell or taste), and normal use of materials for play is typically absent. Younger autistic children often exhibit unusual stereotyped behaviors or motor mannerisms, such as hand flapping or toe walking, and seem to prefer such activities to those involving social interaction, although such behaviors become more prominent as the child becomes older. Bizarre affective responses may be observed; for example, the child may become highly agitated if the same route or routine is not precisely followed.

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Autism and PDDs

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differentiated social responsiveness may be observed in young autistic children (Sigman & Ungerer, 1984), the usual robust patterns of attachment do not develop, and autistic children may not respond differentially to their parents until the elementary school years. Deficits in social interaction remain a source of marked disability even for the highest intellectually functioning autistic adults (Volkmar, Klin, & Cohen, 1997) and may include difficulty dealing with social rules, social interchange, and the pragmatics of social communication.

Social Development

Autism was initially described as a disturbance of affective contact (Kanner, 1943), and social deviance is the major defining feature of the condition. Most recently, research into social development in autism has supported the notion that social criteria tend to be the most potent predictors of diagnosis (Volkmar, Carter, Grossman, & Klin, 1997; Lord, 1995). Social development in autistic children does occur but is qualitatively different from that of typical children, and syndromic behavior is expressed differently at each developmental level. Relative to nonautistic developmentally delayed children, autistic children tend to be significantly more delayed in the domain of social behavior and social skills (Klin, Volkmar, & Sparrow, 1992).

For normally developing infants, social stimuli are particularly interesting; the predisposition to form social relationships appears to be an important foundation for the development of other skills. In contrast, for autistic infants and young children, the human face holds little interest, and lack of eye contact, lack of preference for speech sounds, poor attachments, and a general lack of social interest are typical (Volkmar, Carter, et al., 1997; Klin, 1992).

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Communicative Development

Qualitative differences in communication are present in all children with autism. One of the first notable differences in development is often a delayed onset of intentional communication (both nonverbal and verbal) (Schuler, Prizant, & Wetherby, 1997). Children with autism may be less motivated to communicate at an early age and typically exhibit delays in the acquisition of conventional nonverbal gestures, speech, and language (Wetherby & Prutting, 1984). As infants, children with autism have been found to use a lower frequency of pointing and showing gestures, head shakes, and head nods and may use idiosyncratic behaviors to have their needs met (Schuler et al., 1997). Approximately half of children with autism never gain useful communicative speech, and those who do frequently use immediate and delayed echolalia (e.g., repetition of words and phrases) for communicative purposes (Prizant & Rydell, 1984). Qualitative differences in language acquisition may also include delays in syntactic development, abnormal use of words and phrases, pronoun reversal, and the use of unusual intonation patterns (Lord & Paul, 1997).

Although linguistic form and speech development are often affected, it is the social use of language (i.e., pragmatics) that differentiates children with autism from those with a more circumscribed language disorder. Autistic children who develop intentional communication may communicate less for social purposes than do typically developing children; they may have difficulty monitoring the appropriateness of their discourse and may communicate at a reduced rate (Lord & Paul, 1997). Wetherby
(1986) found that while typically developing children engage in reciprocal interactions for a variety of purposes (e.g., requesting, social interaction, and commenting), children with autism may communicate primarily for instrumental purposes (e.g., requesting and/or protesting activities). In particular, joint attention skills or the ability to establish and maintain a common focus of attention on an entity or event are often restricted in young children with autism (Mundy, Sigman, & Kasari, 1990; Wetherby & Prutting, 1984). Likewise, older children with autism have been found to exhibit difficulty with maintaining conversations and inferring the implicit intentions of their communicative partner (Lord & Paul, 1997). For example, children with high-functioning autism or AS may use unusual discourse (e.g., pedantic speech) and may have difficulty providing relevant and coherent information (Volden & Lord, 1991).

Cognitive Development

Approximately 75–80% are mentally retarded, with about 30% falling within the mild to moderate range and about 45% falling within the severe to profoundly mentally retarded range. Mental retardation is not simply a consequence of negativism while taking intelligence tests, lack of motivation, or oversensitivity to being tested. The typical profile on psychological testing is one marked by significant deficits in abstract reasoning, verbal concept formation, and integration skills and on tasks requiring a degree of social understanding. In contrast, relative strengths are usually observed in the areas of rote learning and memory skills and visual–spatial problem solving, particularly if the task can be completed “piecemeal,” that is, without having to infer the context or “Gestalt” of the task.

Developmental and psychological testing of infants and young autistic children can be challenging but often reveals difficulties with tasks that require more verbal language (either receptive or expressive), symbolic thinking, or social interaction (e.g., tasks that involved imitation). Nonverbal problem-solving abilities, such as matching shapes and solving simple inset puzzles, is closer to age-expected levels (Klin & Shepard, 1994). Deficits in sensorimotor skills, as opposed to more symbolic or verbal skills, are more variably noted (Curcio, 1978; Morgan, Curter, Coplin, & Rodrique, 1989).

Based on the unusual neuropsychological profile in individuals with autism and two decades of cognitive experiments, several influential cognitive theories of the social dysfunction in autism have been proposed. One hypothesis posits that there is a lack of a central drive for coherence, with the consequent focus on dissociated fragments rather than integrated “wholes,” leading to a fragmented and overly concrete experience of the world. Another hypothesis posits that the commonly found difficulties in abstracting rules, inhibiting irrelevant responses, shifting attention, and profiting from feedback, as well as maintaining “on line” different pieces of information while a decision is made—the so-called executive functions—underlie the social, communicative, and behavioral aspects of autism. As executive functions are thought to be mediated by frontal areas, this hypothesis highlights the similarities between autism and conditions resulting from frontal lobe lesions.

Yet another hypothesis, so far the most influential, posits that autism is caused by the child’s inability to attribute mental states such as beliefs and intentions to others. Devoid of a “theory of mind,” individuals with autism are thought to be unable to infer their thoughts and motivations, thus failing to predict their behavior and adjust accordingly, resulting in lack of reciprocity in communication and social contact (Baron-Cohen, 1989). This line of research is of considerable interest in that it more parsimoniously accounts for observed deficits in social interaction and play in autistic children. However, the theory is limited in several important respects. The theory is highly cognitive, and the social deficits in autism are viewed as secondary to an essentially cognitive deficit. Because theory-of-mind capacities are not apparently exhibited much before 1 year of age, at the earliest, the theory does not account for the very early onset of the condition. Comparison of preverbal social behavior in autistic and nonautistic children showed that autistic children lacked early emerging social behaviors that would predate the emergence of the cognitive capacity to appreciate other’s minds (Klin et al., 1992). Moreover, experimental work using the theory has tended to focus on verbal subjects, and it is quite unclear how or whether the theory has applicability to lower-functioning subjects (Klin & Volkmar, 1993). At least some work has suggested that apparent theory-of-mind problems are more a function of verbal
ability and developmental level than of diagnostic category (Prior, Dahlstrom, & Squires, 1990; Tager-Flusberg & Sullivan, 1994).

Neurobiological Studies

Considerable evidence suggests the operation of some as yet unspecified neurobiological factor in pathogenesis. For example, autistic children are more likely to exhibit physical anomalies, persistent primitive reflexes, various neurological “soft” signs, and abnormalities on electroencephalogram computed tomography, or magnetic resonance imaging scans (Minshew et al., 1997). As many as 25% of autistic individuals develop seizure disorders (Volkmar & Nelson, 1990). There is some suggestion of reduced obstetrical and neonatal optimality, although this may reflect problems in the fetus rather than the pregnancy per se (Tsai, 1987).

There is now evidence suggesting the operation of genetic mechanisms in at least some cases (International Molecular Genetic Study of Autism Consortium, 1998; Rutter et al., 1994). Recent research suggests that siblings of autistic children are at significantly greater risk for also exhibiting autism and other developmental difficulties, and that monozygotic twins are more likely than fraternal twins to be concordant for the disorder (Folstein & Rutter, 1977; Rutter, Bailey, Simonoff, & Pickles, 1997). In addition, a recent study of families with more than one child on the autism/PDD spectrum showed preliminary evidence of chromosomal abnormalities (International Molecular Genetic Study of Autism Consortium, 1998).

Unfortunately, neurobiological findings vary considerably and findings are often subtle. Neuroanatomical models of the disorder have placed the “site” of the lesion at various points on the neuraxis from brain stem to cerebellum to cortex. Autistic children exhibit elevated peripheral levels of serotonin, a central nervous system neurotransmitter, yet the significance of this observation is unclear (Anderson & Hoshino, 1997). Also, elevated levels of plasma betaendorphin, and ACTH found in autistic children suggest abnormal functioning within the hypothalamic-pituitary-adrenal axis (Tordjman et al., 1997). This suggests a biochemical abnormality in association with autistic individuals’ heightened response to stress, and stands in contrast to prior studies of cortisol or ACTH secretion, which have not shown significant and robust differences when compared with nonautistics.

Epidemiology

Various problems complicate epidemiological studies, including the relative infrequency of the conditions, difficulties in case identification, sampling techniques, changes in diagnostic criteria, and scope of syndrome definition. Over 20 epidemiological studies worldwide have been conducted (Forbonne, 1998). Prevalence rates ranged from 0.7 per 10,000 to 21.1 per 10,000. The median value of prevalence estimates is 4.5 per 10,000. Since the mid-1980s, a series of studies have reported higher prevalence rates, although it is still unclear whether these reports provide evidence for either a more accurate (and higher) prevalence rate of autism or for an increase in prevalence of autism in the past decade. Possible reasons for the increased rates are (1) the adoption of broader definitions of autism; (2) the inclusion of smaller size target populations, given that in general, smaller studies have yielded the higher rates; and (3) better detection of cases in the extreme ranges, that is, the severely mentally retarded and the nonretarded individuals with autism. Most studies suggest that autism is more frequent in males, usually four or five times as common as in females. When girls are affected, however, they are more severely affected, particularly in terms of lower IQ. The significance of the observed sex difference is unclear but may reflect the operation of underlying genetic mechanisms (Bryson, 1997).

Epidemiological information on other “nonautistic” PDDs is more limited. It does, however, appear that PDD-NOS is much more common than more strictly defined autism. The other PDDs are apparently less common than autism. For example, CDD is perhaps 10 times less common than more strictly defined autism. Although the reliability for diagnostic ascertainment of these autistic-like conditions is still questionable, there is compelling evidence to suggest that perhaps one in every 1,000 children exhibits social disabilities consistent with the autistic spectrum of disorders.

Course and Prognosis

Younger children more typically display the “pervasive” unrelatedness alluded to in DSM-IV (American Psychiatric Association, 1994).
criteria for the condition. Although some evidence of differentiated responsiveness to parents may be observed as the child reaches the elementary school years, patterns of social interaction remain quite deviant, and the child’s behavior can be quite problematic. Often, some gains in communicative and social skills are observed during the elementary school years. During adolescence, some autistic children exhibit behavioral deterioration, and a smaller number improve (Rutter, 1970). As adults, even the highest-functioning individuals exhibit marked difficulties in social interaction (Volkmar & Cohen, 1988). Various interactional styles can be observed in the autistic child, ranging from aloof to passive and to eccentric; these styles appear to be closely related to developmental level (Wing, 1997).

Available data suggest that the outcome for autistic children is quite poor, with perhaps only one-third able to achieve some degree of personal independence and self-sufficiency as adults (DeMyer et al., 1981). In general, two major factors appear predictive of ultimate outcome: the acquisition of truly communicative speech by age 5 and IQ. However, much of the available outcome information is based on samples collected during the 1960s and 1970s. During this period, fewer services were available, and services provided were often not provided until the school years. There is some reason to hope that over the past decade, the mandates for earlier intervention, earlier recognition of the disorder, and more intensive behavioral and social–communicative interventions (Lovaas, 1987; Wetherby, Schuler, & Prizant, 1997) have improved the long-term outcome for the disorder. Research in this area is critically needed.

**DIAGNOSIS OF AUTISM**

**Categorical Definitions**

Categorical definitions of autism typically have emphasized four features essential for diagnosis: (1) early onset, (2) social dysfunction, (3) communicative dysfunction, and (4) various unusual behaviors, such as stereotypies and resistance to change, which are typically subsumed under the term “insistence on sameness.” Most categorical definitions emphasize that deviance in social and communicative development is not just a function of developmental level. During field trials for the DSM-IV, efforts were made to make diagnostic criteria convergent with ICD-10 definitions (Volkmar et al., 1997).

**Dimensional Definitions**

Dimensional approaches also have been used in the diagnosis of autism. These methods attempt to assess dimensions of function/dysfunction that are relevant to the diagnosis. Most dimensional assessment instruments are designed for school-age children. These instruments rely either on parental or teacher report or on direct observation in structured settings; in most instances highly deviant behaviors are rated or sampled. Reliance on parental retrospective brings attendant issues of reliability; direct observational procedures may prove less useful for sampling low-frequency behaviors. In typically developing infants and young children, the frequency of apparently “autistic-like” behaviors raises particular problems for most “deviance model” assessment instruments.

Another approach relies on dimensional assessment instruments that are more truly developmental in nature. The utility of normative assessments of cognitive or communicative ability is well established (Lord, 1997; Lord & Paul, 1997; Sparrow, 1997). The availability of an instrument that normatively assesses social skills, the Revised Vineland Adaptive Behavior Scales (Sparrow, Balla, & Cicchetti, 1984), offers considerable potential in this regard.

**CLINICAL ASSESSMENT**

The clinical assessment of a child with autism or another PDD is most effective when carried out by an experienced interdisciplinary team. Two fundamental considerations should guide the assessment process: (1) an awareness of the challenges that autistic children pose for usual assessment methods and (2) an awareness that some modifications in more usual assessment procedures may be helpful to parents (Klin & Shepard, 1994). For example, to the extent possible, parents should be encouraged to observe the evaluation of the child. This procedure helps to demystify assessment procedures, provides a common set of observations for subsequent discussion, and helps to establish a long-term collaborative relationship.

A careful history should be obtained, including information related to the pregnancy and
neonatal period, early development and characteristics of development, and medical and family history. Information on the nature and age at apparent onset of the condition can provide important information relevant to differential diagnosis. Questions about development can sometimes be helpful if framed for parents around a specific time or event (e.g., the first birthday).

Assessment of the child should include both psychological and communication assessments which aim to establish levels of functioning. Instruments should be selected with consideration of the child's apparent developmental levels. For cognitive assessment, tests that are not highly dependent on verbal abilities should be used, for example, the Bayley Scales (Bayley, 1969), the Uzgiris–Hunt Scales (Uzgiris & Hunt, 1975; Dunst, 1980), the Leiter International Performance Scale—Revised (Roid & Miller, 1997), and the Kaufman Assessment Battery for Children (Kaufman & Kaufman, 1983). For children with nonverbal mental ages over 2 years, several nonverbal tests are available (see Sparrow, 1997, for a discussion of assessment procedures and instruments). When direct assessment of the child is difficult, results of the Revised Vineland (Sparrow et al., 1984) may be helpful. Relative to developmentally disordered, non-PDD comparison groups, the social development of autistic children, as assessed by the Vineland, is lower than expected given the child's overall developmental level (Volkmar, Carter, et al., 1997).

Several instruments are available for assessing communicative skills in young children with autism. Tools that appear to be the most successful involve the use of objects rather than picture stimuli. The Reynell Developmental Language Scales—U.S. edition (Reynell & Gruber, 1990), for example, provides a measure of formal aspects of language comprehension and expression (e.g., vocabulary and syntax) using a variety of miniature objects and figurines. The Communication and Symbolic Behavior Scales—Normed edition (Wetherby & Prizant, 1993) incorporates motivating toys and "communicative temptations," which are useful for providing a profile of a child's communicative skills (e.g., nonverbal and verbal communicative means, communicative functions, and reciprocity), as well as a child's social—affective and symbolic behavior.

Psychiatric examination of the child should include observation during more and less structured periods. Areas for assessment, observation, and inquiry include social development (interest in social interaction, patterns of gaze and eye contact, differential attachments, style of social interaction), communication (receptive and expressive language, nonverbal and pragmatic communication, communicative intents, echolalia), responses to the environment (motor stereotypies, idiiosyncratic responses, resistance to change), and play skills (nonfunctional or idiiosyncratic uses of play materials, developmental level of play). The child's capacities for self-awareness (e.g., interest in mirror image and awareness of his or her own body) and motor skills should be observed. Problem behaviors that are likely to interfere with remedial programming should also be noted (e.g., marked aggression or problems in attention).

Given the difficulties in assessing infants and younger children, several assessment sessions may be required. If a multidisciplinary treatment team is providing the evaluation, it is important that team members maintain close communication with each other to avoid fragmentation and duplication of effort. When possible, the evaluation should be sufficiently integrated so that parents receive a single coherent picture of the child and his or her difficulties; such a report also has the practical advantage of facilitating discussion between team members who must be able to reconcile, or understand, apparent discrepancies in the results.

For younger children, consultations with other medical professionals, such as pediatric neurologists or geneticists, may be indicated. History or examination may suggest the need for specific laboratory studies or medical procedures. For example, a family history of mental retardation, severe mental retardation, or dysmorphic features in the child suggest the need for genetic screening and chromosome analysis (including screening for fragile X); symptoms suggestive of seizures (apparent periodic unresponsiveness) suggest the need for an electroencephalogram and possible neurological consultation. Computed tomography or magnetic resonance imaging scans sometimes reveal disorders such as tuberculous sclerosis or degenerative central nervous system disease. A careful history of the pregnancy and neonatal period should be obtained to ascertain possible pre- or postnatal infections such as congenital rubella.

Usually the child's hearing has been tested prior to comprehensive evaluation. If this has
not been done, or if it was not possible to elicit the child’s cooperation, brainstem auditory-evoked response procedures should be used. In most instances, extensive medical evaluations fail to reveal an associated medical condition; this suggests reasonable care in obtaining additional assessments. On the other hand, certain features may suggest the importance of extensive medical investigations, for example, the abrupt behavioral and developmental deterioration of a child who was previously developing normally.

The differential diagnosis of autism and other PDDs includes language and other specific developmental disorders, mental retardation, sensory impairments (particularly deafness), and reactive attachment disorders. Usually children with language disorders do not exhibit the pattern of serious social deviance and deficit exhibited in autism; nonverbal communicative abilities are an area of evident strength. In mental retardation, social and communicative skills are usually on a par with overall cognitive skills. Deaf children may exhibit some difficulties in social interaction and repetitive activities; however, they are usually interested in social interaction and may make use of gesture for communicative purposes. Children with reactive attachment disorders have, by definition, experienced marked psychosocial deprivation, which results in deficits in social interaction (most notably in attachment). However, the quality of social deficit is different than in autism; the child may be withdrawn or indiscriminantly attached to others. The disturbance tends to improve relatively quickly after an appropriately responsive psychosocial environment is provided.

In young children, the task of differential diagnosis is complicated by the inherent difficulties in child assessment, the frequency of autistic-like behaviors in other conditions, and the fact that autism can be associated with deafness and with mental retardation as well as with other medical conditions. Differential diagnosis is often most complicated in young children without expressive language, odd social behavior, and some apparent degree of cognitive delay. Consideration of the pattern of developmental deviance is often helpful in such instances. The assessment of relative levels of sensorimotor and cognitive skills in relation to communication and social ones is important. In general, the presence of both communication for more social purposes and of some evidence of differential social responsiveness argues against the diagnosis of autism. Often the issue of diagnosis in such cases is clarified with certainty only over time. It is appropriate to share with parents a sense of the clinician’s degree of confidence in the diagnosis. It is also important to realize that the diagnosis may have important, if not necessarily intended, implications for other purposes, such as educational programming, special services in the community, and so forth. It is critical that the importance of educational and other interventions be emphasized regardless of how “classically” autistic the child appears to be.

INTERVENTIONS

In the absence of a definitive cure there are a thousand treatments. Essentially, every conceivable treatment has been used for autism. With the exception of a few areas (notably behavior modification and pharmacological intervention and, to a lesser extent, educational interventions), most proposed interventions have not been rigorously studied, and it has been difficult to assess treatment effects systematically. These poses difficulties for professionals who are asked by parents to recommend or evaluate a therapy. Unfortunately, short-term changes readily occur when treaters and/or evaluators are not blind to the hypothesis under study; short-term changes may be neither sustained nor clinically meaningful.

In some instances, particularly with single case reports demonstrating improvement, it is unclear whether the individual was autistic and which specific factors are responsible for improvement. The observation that a few autistic individuals achieve relatively good outcomes is gratifying but also complicates the interpretation of single case studies. To further complicate the problem, there is no “untreated” autistic child; that is, even by the time the diagnosis is definitively made, parents often have tried multiple interventions.

The available evidence suggests the importance of appropriate, intensive educational interventions to foster the acquisition of basic social, cognitive, and communicative skills (Schuler, Prizant, & Wetherby, 1997; Olley & Stevenson, 1989), which are, in turn, related to outcome. Behavior modification techniques can be quite helpful. Early and continuous intervention is highly desirable (Rogers, 1996).
Some reports have suggested marked improvement following early, intensive intervention, such as Applied Behavioral Analysis (Lovas, 1987). However, questions have been raised about the methodological weaknesses of the research supporting this approach (Gresham & MacMillan, 1998), casting doubt on the high rate of “recovery” of lower-functioning autistic children. Although behavioral approaches have the advantage of addressing a range of behaviors in autism, they should not be considered a comprehensive program to the exclusion of other developmentally based communicative strategies.

Dawson and Osterling (1996) identified several common elements of appropriate early intervention programs designed for young children with autism. The first of these elements, curriculum, included the facilitation of basic skills such as attending to social cues within the environment, imitation, spontaneous communicative intent, symbolic play, and social interaction with both adults and peers. The second element emphasized the need for balancing highly structured teaching environments with conscious efforts to support generalization of those skills to more natural environments. In addition to teacher-directed therapy sessions, a child’s motivation and/or ability to initiate communication spontaneously throughout his or her daily schedule should be targeted in order to foster communicative skills within naturally occurring contexts (Prizant & Wetherby, 1993; Schreibman & Pierce, 1993). Dawson and Osterling (1996) also emphasized the need for predictability and routine, family involvement, and intensive service delivery (especially in the early years).

Educational programs should be highly structured and oriented around the individual needs of the child. Intervention programs should be comprehensive and include the services of various professionals including special educators, speech pathologists, occupational therapists, and so forth. Parental involvement should be encouraged to enhance consistency in approaches at home and in school and to facilitate generalization of skills across settings. Professionals should work with parents to obtain appropriate educational placement and help parents become aware of other community resources, such as respite care.

A marked shift in social policy has resulted in most state agencies attempting to maintain children in their families and communities. Unfortunately, many necessary services may not be provided as a result of this trend. A similar issue has arisen with regard to the integration of autistic children into regular classroom settings. Given the nature of social deficits in autism, there is considerable reason to worry that autistic children may not be as able as mentally retarded, nonautistic, children to profit from such an approach. In considering various alternative educational placements, the individual needs of the child should be paramount. Competency in social skills is as important as level of cognitive functioning when considering the appropriate setting.

In general, pharmacological interventions with infants and young autistic children are best avoided. The best studied agents (i.e., the major tranquilizers) have some limited utility in selected cases, but their many side effects (particularly sedation) may prove problematic (Campbell, Anderson, Green, & Deutsch, 1987). The newer “atypical” neuroleptics may hold some promise for treating specific symptom clusters (Mcdougle et al., 1997), but efficacy has not been established through controlled trials as of yet. These agents may be indicated in some situations but are typically used in older children and, even then, at the lowest effective dose for the shortest period. Although stimulants have been used to treat inattention and hyperactivity in children with PDDs, they have no demonstrable effect on the “core” symptoms of autism (Mcdougle, 1997). Their use is entirely empirical, and often they cause serious adverse effects in autistic children. They should not be a first-line pharmacological intervention. The efficacy of other pharmacological agents has not been clearly established.

Many nontraditional treatments are presently available. In discussing such treatments with parents, it is helpful to explore the rationale for the proposed treatment, the evidence (if any) of efficacy, and its potential costs (in both financial and human terms) to the child and family. Treatments that are minimally disruptive of the child’s educational program and that represent little apparent risk to the child are of less concern than those that entail considerable disruption of the child’s educational program or the family’s life.

In the clinical management of the autistic child, it is important not to lose sight of the needs of the family. Mothers of autistic children show high levels of stress and reactive depression, and the marriage may suffer as well.
IMPLICATIONS

Considerable progress in understanding the nature of autism and related disorders has been made over the past 50 years. Given the early onset of the condition, it is somewhat paradoxical that our knowledge of autism in infants and very young children remains limited in important respects. Knowledge of the other PDDs in infancy and early childhood is even more limited. Medical health professionals have important roles to play in evaluation and provision of remedial programming. Although it now appears that these conditions arise as the result of some insult to the developing central nervous system, precise and testable pathophysiological mechanisms remain to be identified. The study of infants and young children with autism may have important implications for both clinical service and our understanding of the course of early child development.

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