Table 20-1 Pervasive Developmental Disorder: Characteristics

FEATURE	AUTISM	ASPERGER	RETT	CDD	PDD-NOS
Epidemiology	10 cases per 10,000; 5:1 male-to-female ratio	2.5 per 10,000; 5:1 male-to-female ratio	0.44 to 2.1 per 10,000; diagnosed only in girls; associated with *MECP2* gene mutation	0.11 per 10,000	2 to 16 per 10,000
Onset	Before age 3 yr	Usually first recognized in preschool age; Motor delays seen	Before age 5 yr; Head circumference growth dramatically decelerates and child begins losing skills.	Child appears normal until age 2 yr, then dramatic autistic-like picture	Variable; Child does not meet full criteria for other PDD
Language/ communication impairment	Marked	Good verbal ability but poor communication	Very marked	Marked (normal previously)	Variable
Motor issues	Poor; May be preserved previously	Fine and gross motor difficulties	Significant loss of motor abilities, hand-washing stereotypes; A characteristic hand-wringing movement exists.	Often preserved but child may lose some skills	Variable
Restricted interests	Marked, mannerisms, trouble with change, occasionally savant ability	Highly circumscribed interests that may interfere with functioning	Significant psychomotor retardation	Marked, as in autism	Variable; mannerisms may be less prominent, but child is often troubled by change.

CDD, Childhood disintegrative disorder; *PDD-NOS,* pervasive developmental disorder-not otherwise specified.

OCD), pronoun reversal, nonsense rhyming, and other abnormalities. Intense absorbing interests, ritualistic behavior, and compulsive routines are characteristic, and their disruption often invokes tantrum or rage reactions. Head banging, teeth grinding, rocking, diminished responsiveness to pain and external stimuli, and self-mutilation may be noted.

Although the etiology of autistic disorder is unknown, there is an increased risk of autistic disorder in siblings compared to the general population. Twin studies have revealed high levels of concordance for monozygotic twins. Family studies reveal prevalence rates of between 2% and 10% in siblings, and, when absent, there may be increased risk for language, learning, and social development problems.

It is proposed that the brain connectivity is adversely affected. Abnormalities in the limbic system, temporal, and frontal lobes have been suggested. Some postmortem studies reveal abnormalities in the brain microarchitecture, size, and neuronal packing. Functional magnetic resonance imaging (MRI) studies show hypoactivity of the fusiform gyrus of the amygdala, a location involved in face processing tasks and facial expression recognition involved in social and affective judgments.

The American Academy of Pediatrics recommends screening for autism at 18 and 24 months of age. Comprehensive testing should be done if there is an affected sibling or parental, other caregiver, or pediatric concern.

There are no definitive laboratory studies for autistic disorder, but they can help rule out other diagnoses. A hearing test (may account for the language deficits), chromosomal testing (to identify fragile X syndrome, tubular sclerosis, and genetic polymorphisms), congenital viral infections, and metabolic disorders (phenylketonuria) should be performed. Electroencephalography abnormalities may be seen in 20% to 25% of children with autism but are not diagnostic. Psychological tests in children with autism often show strengths in nonverbal tasks (e.g., puzzles) and marked deficiency in verbal cognitive abilities. IQ is usually low, though savant skills and hyperlexia (a precocious interest in letters and numbers) are sometimes observed. Speech pathology consultation can be helpful in evaluating the communication difficulties.

Common comorbidities are mental retardation (in up to 80%), seizure disorder (in 25%), anxiety disorders, OCD, and attention-deficit/hyperactivity disorder. Seizures often start around the onset of puberty. Higher IQ and better language skills are related to improved prognosis. Good communication by the age of 6 years and average nonverbal cognitive skills predict the likelihood of living independently or in a less structured group living situation.

The earliest studies of autism suggested a relatively poor prognosis, with only a small number of individuals (1% to 2%) being able to function independently as adults. Recent research reveals major gains, but not a cure, with early diagnosis and treatment.

SCHIZOPHRENIA

Schizophrenia generally presents in adolescence or early adulthood. The same diagnostic criteria are applied as in adults but must be interpreted in terms of the developmental stage of the child (Table 20-3).

Childhood-onset schizophrenia is a rare disorder (<1 in 10,000 children) and usually indicates a more severe form of schizophrenia. The frequency increases between 13 and 18 years of age. Boys tend to be affected about twice as often as girls, regardless of ethnic or other cultural factors. The etiology of schizophrenia is unknown. Numerous studies have shown genetic predisposition and linkages for the disorder. In addition, family studies consistently show a higher risk in monozygotic twins compared with dizygotic twins and siblings. First-degree relatives of patients with schizophrenia have a 10-fold higher risk.

The symptoms of schizophrenia typically fall into four broad categories:

- **Positive symptoms** include hallucinations and delusions. Hallucinations are auditory or visual misperceptions that occur without external stimuli. Delusions are fixed false