PERVASIVE DEVELOPMENTAL DISORDERS

The broad diagnostic category of pervasive developmental disorders (PDDs), now often called autistic spectrum disorders (ASDs), encompasses several related yet distinct disorders, including autistic disorder, childhood disintegrative disorder, Rett's disorder, Asperger's disorder, and pervasive developmental disorder, not otherwise specified. The ASDs are characterized by significant and pervasive impairments in several areas, including social interaction skills, communication abilities, and the presence of restricted and repetitive behaviors, interests, or activities. These impairments are atypical relative to the individual's developmental level, usually appear early in life, and are often associated with mental retardation.

AUTISTIC DISORDER (AUTISM)

Autism is characterized by significant impairments in social interaction and communication as well as restricted and stereotypic patterns of behavior, interests, and activities (e.g., resistance to change, repetitive nonfunctional motor mannerisms, and preoccupation with parts of objects). Additionally, unusual responses to the environment (e.g., insensitivity to pain, over-reactivity or under-reactivity to noise) may be present. Social deficits include impairments in the use of multiple nonverbal communicative behaviors (e.g., eye contact, use of gestures), reduced awareness of others, inability to form appropriate relationships, failure to spontaneously share enjoyment and experiences, and lack of social and emotional reciprocity (e.g., not actively participating in social games; not noticing another's distress).

Language comprehension is significantly impaired (sometimes worse than expression) in people with autism, and nearly half are never able to communicate verbally. Those with verbal skills often have odd intonation, reverse pronouns, make up terms (neologisms), rarely use idioms correctly, and may repeat phrases heard on previous occasions (echolalia). Individuals without functional spoken language can benefit from training in the use of sign language, picture exchange, or other forms of augmentative communication (e.g., computers). Pretend play, imitation, and joint attention (seeking attention for the purpose of sharing interest or pleasure) are also impaired.

Both the cognitive deficits and behavioral sequelae of autism can range from mild to severe. Some individuals engage in disruptive behaviors, including self-injury, aggression, and property destruction. Most adults with autism require varying degrees of caregiver support throughout their lives; only a minority of autistic adults achieve independent living. The two most common predictors of better outcome in people with autism are higher intelligence quotient (IQ) and greater functional language.

Recent prevalence estimates range as high as 60 in 10,000 individuals. The disorder is four to five times more common in males, although females are more likely to exhibit more severe mental retardation. Because there is a genetic component in the development of autism, there is an increased risk (3%–10%) for the disorder in siblings of affected people and an even higher risk for partial forms of the disorder appearing in siblings. Although several environmental factors, such as toxins and vaccination, have been suggested to cause autism, there is no evidence supporting this. Neuropathological and neurochemical studies suggest a wide range of possible abnormalities, with the most consistent evidence focusing on the serotonin system (neurochemically) and the limbic system (anatomically).

CHILDHOOD DISINTEGRATIVE DISORDER

Childhood disintegrative disorder (CDD) is probably the rarest and least understood of the ASDs. CDD occurs when a child develops typically for at least 2 years before experiencing a marked overall decline in previously learned skills. Some early signs that may be present before skill regression include irritability, anxiety, increased activity, and a loss of interest in the environment. The decline in skills occurs before the age of 10 (usually between the ages of 3 and 4) and results in significant impairment in the areas of play skills, language, social skills, and adaptive behavior. Additionally, children with CDD may develop stereotypies (nonfunctional repetitive motor or vocal behaviors) and restricted interests. Many individuals with CDD exhibit disruptive behaviors, including aggression and self-injury. The overall presentation of children after regression is nearly indistinguishable from children with autism, and the prognosis is usually poor. Children usually function in the severely mentally retarded range and are at higher risk for developing seizures. Often, subtle neurological impairments and abnormalities are present.

Although a specific cause has not been found, it appears as though CDD occurs as the result of damage to the developing brain. Prevalence data are lacking, although CDD may be underdiagnosed because of the symptom overlap with other ASDs, including Rett's syndrome and autism, as well as degenerative neurological disorders. It is important to conduct a thorough medical and neurological examination to rule out other causes of the developmental regression. This is especially important when regression occurs at a later age (e.g., after age 5). Additionally, individuals with CDD should be monitored closely for seizure disorders. Epidemiological data are limited, but recent data suggest that CDD is more common in males. CDD is a lifelong condition, although the loss of skills usually stabilizes, and some limited improvement occurs.

RETT'S DISORDER

Rett's disorder (also called Rett's syndrome) is a severe neurodevelopmental disorder primarily affecting females that impairs all aspects of development. It causes severe to profound mental retardation, severe communication impairment, loss of functional hand use and other physical disabilities, and impaired social skills, as well as medical problems such as seizures, feeding difficulties, cardiac abnormalities, scoliosis, and autonomic nervous system dysfunction. With an estimated prevalence of 1 in 10,000 to 15,000 female births, it is among the most common causes of mental retardation in young girls. A major research advance came in 1999 when a specific genetic abnormality was identified in most girls with Rett's syndrome. This gene *(MECP2*) encodes for a protein that plays a role in the regulation of gene expression and is found on the Xq28 region of the X chromosome.

Rett's syndrome is unique among the ASDs in that stages of illness progression have been delineated. The first stage begins after a period of typical development that generally lasts between 6 and 18 months. During Stage 1 (early-onset stagnation), deceleration of head growth and reduced interest in playing become apparent. In addition, the appearance of odd hand-waving behaviors and reductions in eye contact and language abilities may be observed. Severe regression occurs in Stage 2 (developmental regression), which occurs between 1 and 4 years of age. In this stage, the characteristic hand-washing or hand-wringing behaviors result in a loss of hand skills; gross motor impairment and clumsiness are present; the ability to speak is lost; and the capacity to understand language is seriously impaired. Furthermore, cognitive abilities deteriorate, breathing is often irregular, seizure activity often begins, and episodes of laughing during the night may occur. It is during this stage that symptoms may mimic those of autistic disorder, and thus differential diagnosis can be challenging.

The onset of stage 3 (pseudostationary period) is variable. During this time, skills do not deteriorate as much, and the girls begin to interact more with their environments. Although not able to communicate verbally, many with Rett's syndrome are able to make basic communicative efforts using eye gaze and intense staring. Gross motor skills continue to deteriorate, seizures are common, and this period usually lasts years to decades. Stage 4 (late motor deterioration) is typified by complete loss of the ability to walk and increased physical rigidity. Severe scoliosis and progressive muscle wasting are characteristic, and most individuals must use wheelchairs by adulthood. However, seizure activity tends to decrease, the deterioration of cognitive abilities stabilizes, and contact with others improves.

ASPERGER'S DISORDER

Asperger's disorder (or Asperger's syndrome) is characterized by significant and long-standing impairments in social interaction, restricted and repetitive activities and interests, and stereotyped behaviors. Unlike individuals with other ASDs, those with Asperger's syndrome do not display clinically significant delays in language or cognitive development. Because of typical (or nearly typical) cognitive and language development, Asperger's syndrome is often not diagnosed until the late preschool or early elementary school years when social deficits become more apparent. People with Asperger's have narrow interests (e.g., clocks, hotels, or train schedules) with which they are single-mindedly preoccupied to the exclusion of other developmentally appropriate activities. These restricted interests often interfere with social and academic development. For example, individuals with Asperger's often attempt to engage others in conversation related to these stereotyped interests without regard for the other's interest in the topic.

Individuals with Asperger's syndrome generally excel in academic subjects that entail rote memorization but tend to fare more poorly in tasks requiring

flexible thinking, creativity, and higher-order cognitive processes such as abstract thinking. Similarly, these individuals tend to apply social rules in a rote manner and evidence a strict adherence to routines. Anecdotally, people with Asperger's syndrome are clumsy or physically awkward and often have difficulty with tasks that require fine motor abilities such as writing.

People with Asperger's often have a distinctive vocal quality characterized by overly formal speech, atypical prosody, and reduced use of gestures. Furthermore, these individuals have difficulty interpreting nonverbal cues (e.g., diverted gaze as indicative of loss of interest) and the more subtle nuances of language. Additionally, an overly literal interpretation of language that results in a lack of understanding of idioms, common expressions, and the like is generally found. Appropriate pragmatic language skills (e.g., eye contact, body proximity during conversation, and appropriate social greetings) are often lacking. Together, these impairments result in social difficulties, peer rejection, and alienation. People with Asperger's are typically aware of their social deficits and may make unsuccessful efforts at social contact, which can result in distress and elevate risk for the development of affective disorders such as depression.

Asperger's syndrome is diagnosed at least five times more frequently in males, and there is often a family history of social difficulties or other ASDs. As Asperger's syndrome has come to be more widely recognized among clinicians and parents, controversy has arisen surrounding accurate and appropriate diagnosis. Some professionals tend to loosely apply diagnostic criteria, and many in the field think that this condition may be overdiagnosed. Thus, reliable prevalence data are currently unavailable. In addition, questions exist regarding how Asperger's fits into the autism spectrum and whether it is actually distinct from high-functioning autism (HFA). Differential diagnosis is complicated by the fact that there is symptom overlap with other disorders (e.g., schizoid and schizotypal personality disorders, nonverbal learning disability, and attention deficit hyperactivity disorder), although there are separate and unique diagnostic criteria that should differentiate Asperger's from these other conditions.

A better prognosis is associated with Asperger's than with the other ASDs because these individuals do not have the cognitive impairments or communication deficits that are associated with autistic disorder, Rett's disorder, and childhood disintegrative disorder. People with Asperger's syndrome may be able to pursue careers related to their restricted interests, although they often fail to achieve occupational status commensurate with their level of cognitive and academic functioning.

PERVASIVE DEVELOPMENTAL DISORDER, NOT OTHERWISE SPECIFIED

A diagnosis of pervasive developmental disorder, not otherwise specified (PDD-NOS) is indicated in individuals presenting with significant impairments in social interactive skills in conjunction with one or both of the following: (1) verbal or nonverbal communication deficits; and (2) stereotyped behavior, interests, or activities. This category includes "atypical autism," or symptom presentations that do not meet full diagnostic criteria for autistic disorder. Clinically, the PDD-NOS diagnosis is often applied rather liberally and inappropriately in cases in which a diagnosis of various behavior disorders, mental retardation, autistic disorder, or one of the other ASDs would be more appropriate.

TREATMENT INTERVENTIONS

Interventions include the use of applied behavior analysis to teach academic, self-care, and adaptive skills and to reduce disruptive behavior. People with autism learn best when skills are broken down into small steps, when provided with many opportunities to practice skills, and when success is rewarded. They may need additional assistance in generalizing skills to different environments. Children with ASDs benefit from structured interactions with their typically developing peers. In individuals without functional spoken language, the use of alternative communication methods such as sign language or picture systems, including the Picture Exchange Communication System (or PECS), is recommended. For some people with autism, medications may be beneficial in reducing disruptive behaviors and stereotypy; improving attention, mood, and sleep; and reducing anxiety and depression, although they are generally considered not to address the core social and communication symptoms. In addition, parents and physicians should be aware of the higher risk of seizure disorders in this population. Parents and professionals should also be

cautious when considering unproven interventions. Because the PDDs are severe and not well understood, myriad nontraditional treatments have been introduced, including chelation, facilitated communication, special diets, and secretin. The effectiveness of these interventions is questionable, and some may be harmful.

—Hilary C. Boorstein, Deborah A. Fein, and Leandra B. Wilson

Further Readings and References

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