

5 Pervasive Developmental Disorders: The Autism Spectrum Disorders

Essential Concepts

- The essential feature of the pervasive developmental disorders is abnormal social development and relatedness.
- Early detection and early intervention improve prognosis.
- Some children may be diagnosed before 1 year of age.
- Treatment should be multimodal and multidimensional.
- Anxiety and other psychiatric disorders may complicate prognosis and treatment.

Pervasive developmental disorders (PDDs) are also referred to as the autism spectrum disorders (ASDs). There are five disorders in this category: autism, Asperger disorder, Rett disorder, childhood disintegrative disorder, and pervasive developmental disorder, not otherwise specified (PDD, NOS). The core feature of the PDDs is an abnormal relatedness and social development. Although cognitive and motor development are often also affected, it is the manner of relating and communicating that is the sine qua non of the disorders.

These are tragic yet fascinating disorders. Although the movie *Rainman* was fiction, individuals with ASDs may be high functioning in many ways, but experience an extreme need for sameness and routine, lack of flexibility, inability to read social cues and interact in a reciprocal manner, and odd speech. The term "spectrum" is sometimes used to denote the fact that the level of impairment and disability can be quite variable.

At one time, individuals with ASDs were thought to be resistant to intervention. We now know that early multimodal and multidimensional treatment may markedly improve prognosis. Support for the family, in addition to the child, is also required. Psychiatrists work within a system of care to provide needed services for a child and his or her family.

BASIC PRINCIPLES

From the earliest description of autism by Leo Kanner in 1943, the disorder has been studied widely to ascertain the etiology and effective treatments. Some initial descriptions of "refrigerator mothers" as the cause has been long since dispelled. A neurological insult of multiple etiologies (genetic, intrauterine, neurotransmitter, or neurophysiological abnormalities) is posited. Parenting patterns do not cause autism. However, high parental skill level in working with his/her child may improve prognosis.

Although many children with more severe forms of ASD may be identified early, I find that schools commonly request consultation for children with mild "spectrum" disorders in the early elementary years. These children often present with tantrums as a key concern. The demands of school (for socially appropriate and conforming behavior) and the exquisite sensitivity of an ASD child to overstimulation, poor social and coping skills, and extreme need for sameness may overwhelm these youngsters. Oppositionality, obsessive-compulsive behaviors, and behavioral outbursts may be the primary complaint. A full evaluation followed by recommendations that allow the child to feel comfortable and not overwhelmed in the educational setting is often crucial to the child's ability to learn and the school's ability to provide for him or her.

Diagnostic Criterion and Epidemiology

It is estimated that 1% of the population may have a diagnosable autistic spectrum disorder. The number of children diagnosed with ASD has increased rapidly in the last 10 years, probably due to increased rate of detection of milder forms of the disorder, as well as potentially genetic or environmental contributors. The prevalence is greater in boys (except for Rett disorder). Girls with the disorder tend to be more severely affected. ASDs present in equal prevalence across race, ethnicity, and nationality. Table 5.1 summarizes the diagnostic criteria and epidemiology of the PDDs. Tables 5.2 and 5.3 summarize the etiology and differential diagnosis.

Comorbid Mental Disorders

Comorbidity is common with the ASDs. It is estimated that up to 80% of children with autism also have mental retardation. Anxiety disorders, obsessive-compulsive disorder, and

Childhood disintegrative disorder	Estimated at 0.11 per 10,000 Boys, 8; Girls, 1	Normal development for at least 2 years Severe loss of developmental skills before 10 years of age	Skill loss usually occurs over a 6-9 month period, then plateaus Poor prognosis Mental retardation
Rett disorder	0.44 to 2.1 per 10,000 Girls only (rare reports of boys)	Normal at birth, but onset by 2 years Deceleration of head growth Loss of motor skills with hand-wringing movements; gait disturbance Loss of language Loss of social engagement	Severe regression in skills by age 2 Physically and socially debilitating High mortality (1.2%/yr)
PDD, NOS	2-16 per 10,000 Boys > Girls	"Atypical autism" Does not meet criteria for autism because of late age of onset, atypical symptoms, or subthreshold symptoms	Life-long, but variable outcome Frequent comorbid psychiatric disorders

Disorder	Prevalence	Clinical Findings	Clinical Course
Autism	2-15 per 10,000 Boys, 4; Girls, 1 Girls generally with more severe disorder	Severe impairment of social interaction and communication Restricted, repetitive, and stereotypic patterns of behavior, interest, and activities Onset before age 3 Mental retardation common	Life-long course Seem to improve as they mature Superior IQ improves prognosis 1/3 develop comorbid psychiatric disorders
Asperger disorder	Estimated 10-36 per 10,000 Boys, 5; Girls, 1	Impairment in social interactions Preoccupation with one or more restricted patterns of interest No delay in language or cognitive development Nonverbal learning disability cognitive profile common Motor clumsiness	Life-long course Seem to improve as they mature Superior IQ improves prognosis 1/3 develop comorbid psychiatric disorders

TABLE 5.1. Characteristics of the Pervasive Developmental Disorders

attention deficit hyperactivity disorder are all quite common. Tic disorders and psychotic symptoms are also notable comorbidities. Of note, a number of chromosomal disorders (especially fragile X and tuberous sclerosis) present with autistic-like features. Intrauterine viral infections, phenylketonuria, and seizure disorders are also associated. Table 5.4 summarizes components of a thorough evaluation.



When you sit with a child, try to imagine the world through the child's eyes. If you find yourself feeling frustrated by your inability to engage the child, ignored or used as a tool, or having a very difficult time helping the child with a transition or reciprocal play, consider an ASD. More subtle forms of Asperger disorder may present in a very bright and interesting child who spends a session telling you about the details of his Yu-Gi-Oh cards (or any other area of fascination). It is the lack of flexibility, pedantic or professor-like speech, one-way nature of the communication, and high level of persistence in the topic that distinguish the child with mild Asperger disorder from a child who loves a hobby. Adults may delight in learning about the topic. Peers typically do not.

Treatment

Early detection and developmentally sensitive multimodal treatment are required for optimal outcomes. The primary intervention for ASDs is educational. Another key element is parent guidance and support, as this disorder is quite devastating to parents and families. Public Law 94-142, the Individuals with Disabilities Educational Act (IDEA), stipulates that every child, regardless of his or her disability, has a right to a free and appropriate public education in the least restrictive environment. For the eligible child, public educational services are mandated to begin at age 3. Key elements of early interventions include: 1) teaching the child to pay attention to other people, imitate others, use preverbal and verbal communication, play and socially interact; 2) a teaching environment that is highly supportive of the child's learning needs and involves systematic teaching of skills in a one-to-one setting with trained personnel; 3) a program that is predictable and routine; 4) a functional approach to problem behaviors; 5) a thoughtful strategy

TABLE 5.2. Etiology of Autism Spectrum Disorders

Type	Description
Hereditary	Multiple genes involved 50 times more prevalent in siblings High concordance (60–90%) in monozygotic twins Associated with other known genetic syndromes (e.g., fragile X and tuberous sclerosis)
Early alterations in embryonic development	Prenatal damage due to infection, toxins, and substance abuse, or etiology unknown
Neuroimaging findings	Ventricular enlargement/abnormal symmetry Cerebellum with hypoplasia Increased serotonin synthesis in dentate nucleus Lack of activation of fusiform gyrus (brain area that recognizes faces) Larger brains than typically developing children
Neurotransmitter abnormalities	Abnormalities in glutamate, serotonin, dopamine, opioid, and GABA neurotransmitters Autoimmune disorders/antibodies to serotonin-1A receptors

TABLE 5.3. Differential Diagnosis for Autism Spectrum Disorders

Disorder	Differentiating Features
Selective mutism	Child is able to speak and does so with family History of normal relatedness with family
Developmental language disorders	May use nonverbal cues; generally well related
Verbal apraxia/dyspraxias	Communicates nonverbally
Reactive attachment disorder	History of severe abuse/neglect
Attention deficit hyperactivity disorder	Able to make friends, even if their behavior makes sustaining friendships difficult
Mental retardation	Eye contact and relatedness spared
Childhood schizophrenia	Bizarre or unusual thoughts, hallucinations, loss of reality testing

TABLE 5.4. Evaluation Essentials for Children with Autism Spectrum Disorders

- **Clinical History**—focus on pre- and perinatal history, early social and language development, sensory sensitivities (to sounds, light, tactile stimulation, smells), odd or repetitive behaviors, ability to make transitions and changes in routine, family history, medical history.
- **Evaluation of the child**—focus on eye contact, ability to relate, ability for reciprocal and imaginative play or talk, note perseveration on a topic or activity, tone of voice and intonation, unusual movements or stereotypes, delayed or unusual speech, need for doing things in a particular manner or to be in charge of how things are done in a persistent and intense manner. Observation of the child's functioning in school or at home may also be helpful.
- **Medical and neurological assessment and hearing evaluation** (may require brainstem audiometry if child is not able to fully cooperate).
- **Psychological testing** to determine cognitive level, adaptive functioning scales (needed to diagnose mental retardation), academic testing to rule out learning disabilities and assist in academic planning. Speech and language testing, hearing testing, and occupational therapy and physical therapy evaluations should be completed as appropriate for the child. A young child should receive a developmental evaluation.
- **Diagnostic scales:** Autism Diagnostic Observation Schedule (ADOS) and Autism Diagnostic Interview (ADIR).

for transitioning from a specialized preschool classroom to the kindergarten class; 6) family involvement.

Treatment is multidisciplinary (teachers, mental health professionals, medical professionals, speech and language, occupational, and physical therapists, parents) and multimodal (utilization of multiple approaches targets the specific areas of disability, such as social, cognitive, motor, and academic). Table 5.5 summarizes the common educational, psychosocial, and pharmacological treatments used in providing for the complex child with ASD.

KEY POINT

Despite considerable attention in the lay press, there is no evidence that links the mumps, measles, and rubella (MMR) vaccine to autism. Due to the severity and chronicity of the disorder, a plethora of unproven therapies have gained attention.

Many families will ask about these. They include nutritional supplements, elimination diets to combat food allergies, infusions of immune globulins, secretin, chelation therapy to remove mercury, facilitated communication (support of child's arm and hand while typing on keyboard), and others. There is lack of empiric support for all of these therapies, and education of families about the data and research is an important component of treatment.

CLINICAL VIGNETTE

Bob presented at age 3 with multiple problems including decreased and unusual language (echolalia, pronoun reversal), play which included lining up cars in pristine rows or spinning the wheels, and sensory sensitivities (tactile and loud noises). Bob's mother noted that his new 5-month-old sister smiles and looks at her more than Bob ever did. She assumed that Bob's difficulties had been due to his recurrent ear infections and lack of hearing. You refer for hearing testing, routine medical and neurological examinations, and developmental testing. Language and social skills deficits are notable, and you diagnose autism. Bob is enrolled in a preschool program, then half-day kindergarten. You consult again when he is 6 years old and in first grade.

Bob is very difficult to manage at school and at home. Bob throws tantrums for 1 to 2 hours if not allowed to do as he pleases. He is very active, inattentive, and does not sit still in school. The parents, initially resistant to medications, requested a medication assessment due to Bob's inability to comply with even simple commands and his lack of educational progress. A low dose of the stimulant methylphenidate was initiated for treatment of attention deficit hyperactivity symptoms. However, Bob demonstrated increased irritability and sleep disturbance with even very low doses of methylphenidate. Thus, it was discontinued, and a trial of the atypical antipsychotic risperidone was initiated for treatment of his inflexibility and agitation. Bob's motor activity decreased to a more age-appropriate level, tantrums decreased dramatically, and he was amenable to the school curriculum. In fact, the family was able to take a vacation together and enjoy it for the first time, with only minor outbursts by Bob.

TABLE 5.5. Treatment Essentials for Children with Autism Spectrum Disorders

Comments	Reference
Applied behavioral analysis (ABA) Plan that teaches appropriate behaviors that are to be generalized to all domains of a child's environment	Rosenwasser and Axelrod; Lovaas
Discrete trial training (DTT) Teaching skills in specific situations	Grindle and Remington
Structured teaching A system for organizing the environment and optimal conditions under which the child should be taught	Schopler
Developmental individual difference relationship model (DIR) "Floortime"—uses relationship method to help the child relate and attend to the social setting	Greenspan and Wieder
Treatment and education of autistic and communication-handicapped children Collaboration between mental health and educational professionals with parents to formulate an effective education and treatment plan	Schopler
Social stories The use of stories to problem solve social dilemmas	Gray and Garand

Psychosocial Approach	Comments
Family support	Support groups, individual supportive counseling
Parent psychoeducation	Teaching parents about the disorder and collaboration in treatment planning
Parent behavioral management training	Use of behavioral specialist to help parents learn to employ behavior management protocols to help their child learn appropriate behavior
Public education system	Special educational services individualized to the needs of the child
Referral for special therapies (speech, occupational, physical)	Speech therapy for delays and to teach social speech OT for sensory processing and fine motor deficits PT for coordination deficits
Referral for disability services and support	Ensure that the child and family gain appropriate entitlements commensurate with the child and family's needs
Psychopharmacological Interventions	Target Symptoms
Antipsychotics: risperidone, aripiprazole, ziprasidone, olanzapine, quetiapine, haloperidol, thioridazine	Aggression, agitation, irritability, hyperactivity, and self-injurious behavior
	The primary medications used to treat behavioral disturbances in ASD are weight gain, hyperlipidemia, hypertension, and increased prolactin

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Attention Deficit
Hyperactivity Disorder

Essential Concepts

- ADHD is a clinical diagnosis based on careful history taking, clinical examination, and information from multiple sources and multiple settings (school, home, community).
- The clinician must differentiate the core symptoms of ADHD from the secondary effects of other psychiatric disorders.
- Hyperactivity does not need to be present during the mental status exam to make the diagnosis of ADHD.
- Concomitant learning disabilities and comorbid psychiatric disorders should be evaluated.
- Baseline and follow-up rating scales are helpful in monitoring the effectiveness of treatment interventions and medication regimens.

Attention deficit hyperactivity disorder (ADHD) is the most commonly diagnosed psychiatric disorder of childhood and is characterized by deficits in attention, concentration, activity level, and impulse control. ADHD tends to run in families and is often associated with significant comorbidity with other psychiatric disorders, both externalizing (such as oppositional defiant disorder and conduct disorder) and internalizing (such as depression and anxiety), as well as bipolar disorder. The impact of ADHD on the child, his or her family, schools, and society is enormous, with billions of dollars spent annually for school services, mental health services, and increased use of the juvenile justice system. In contrast with historic notions, children do not typically "outgrow" ADHD. Morbidity and disability often persist into adult life. It is not infrequent that I have revealed the

TABLE 5.5. Treatment Essentials for Children with Autism Spectrum Disorders (continued)

Psychopharmacological Interventions	Target Symptoms	Comments
SSRIs: fluoxetine, sertraline, citalopram, escitalopram, fluvoxamine	Anxiety, perseveration, compulsions, depression, and social isolation	Positive response may be correlated with family history of affective disorder
Stimulants: methylphenidate, dextroamphetamine, amphetamine salts	Hyperactivity and inattention	Potential adverse effects are restlessness, insomnia, mania
Alpha-2 agonists: guanfacine, clonidine	Hyperactivity, aggression, and sleep dysregulation	Variable response More positive response with Asperger disorder May increase agitation and stereotypic behaviors
Anticonvulsants and lithium	Aggression and self-injurious behavior (SIB)	Clonidine is the more sedating Need for blood monitoring may limit use
Naltrexone	Self-injurious behavior (SIB)	May help mood lability May be useful, but not robust data
Amantadine	Hyperactivity, irritability, and aggression	Need hepatic monitoring Need more studies
Melatonin	Sleep dysregulation	May be first line for insomnia