Autism Spectrum Disorder

Description/Etiology
Autism spectrum disorder (ASD) is a pervasive developmental disorder that manifests in early childhood and is characterized by severe impairment in socialization and communication, stereotypical and rigid behavior, and restricted interests. The Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) definition of ASD combines the previous edition’s (i.e., DSM-IV-TR’s) diagnoses of autism, Asperger syndrome, childhood disintegrative disorder, and pervasive developmental disorder not otherwise specified. To be diagnosed with ASD, an individual must exhibit deficits in communication and social interaction and restricted and repetitive behaviors, interests, and activities. Signs and symptoms of ASD usually appear in early childhood—typically during the second year of life—as delayed language development, lack of social interest or interaction, intense interest in a limited range of activities, and a rigid response to change. In order for a diagnosis of ASD to be made, these manifestations must cause clinically significant impairment in functioning and must not be better explained by intellectual disability or global developmental delay.

Although there is no known cause, it is thought that genetic influences and gene-environment interactions play a central role in the development of ASD, and that these factors result in deficits in inhibitory control and executive function activities (e.g., planning, selective attention, working memory, problem solving, cognitive flexibility) that produce the characteristic behavioral patterns of ASD. Cognitive function in patients with ASD ranges from developmentally delayed to high functioning. Some individuals with ASD exhibit irritability and aggressiveness toward self or others. Many are frequently hypersensitive to touch, pain, light, and other stimuli. Language deficits are common and some patients lack speech completely. Patients with ASD are at increased risk for seizures.

Diagnosis of ASD is clinical based on patient assessment. ASD can be diagnosed as early as 12 months in cases of severe developmental delay and as late as kindergarten or first grade in children without intellectual or language impairment. ASD is a lifelong disorder that requires continued medical surveillance and intensive, individualized, structured educational and behavioral interventions with an interdisciplinary team and family involvement. Interventions from a variety of disciplines such as speech, behavioral, and occupational therapy are necessary to increase adaptive behavior and self-reliance skills. Medication can be prescribed if appropriate for management of severe behavioral manifestations (e.g., aggressiveness, irritability) or if comorbid conditions (e.g., anxiety, seizures, depression) are present. It is important that all members of the treatment team focus on the patient’s individualized needs and treatment regimen to build structure and maintain consistency. Inconsistency in routine can cause anger or rage in some individuals with ASD. Prognosis is largely determined by the patient’s IQ.

Facts and Figures
ASD affects an estimated 1% of the population of the United States. It is unclear whether the actual incidence of ASD has been rising over the past two decades or if the increased rate of diagnosis is more correctly attributed to earlier diagnosis, broadened diagnostic criteria, and/or heightened public and parental awareness of ASD. It is four times more common in males than in females, but females tend to have more severe intellectual disability. An estimated 30–70% of patients with ASD have comorbid psychiatric disorders.
Seizure disorders affect 25% of patients with ASD. About half of patients with ASD have severe or profound intellectual disability, 35% have mild to moderate intellectual disability, and 20% have IQs in the normal range. Just 20% of children with ASD are diagnosed before three years of age and at least 33% are diagnosed after six years of age.

**Risk Factors**

Although the etiology of ASD is unknown, genetic, infectious, toxic, and traumatic factors might be associated with increased risk. Heritability estimates for ASD range from 37% to > 90%. Risk of recurrence in siblings is reported to be as high as 25%. An estimated 15% of cases are the result of a known genetic mutation and ~ 1,000 gene mutations have been identified as being possibly contributory to ASD. A few cases are associated with genetic syndromes, including fragile X syndrome, tuberous sclerosis, Down syndrome, Rett syndrome, and Angelman syndrome. Additional risk factors include parental schizophrenia, maternal depression, nonpsychotic personality disorders in a parent, advancing age of either parent, closer spacing of pregnancies, premature birth before 26 weeks’ gestation, and maternal use of valproate during pregnancy. Viral infection during the first trimester of pregnancy is a suspected cause of ASD; maternal rubella during pregnancy is associated with significantly increased rates of ASD. Women with diabetes mellitus, hypertension, or obesity might be at increased risk of having a child with ASD.

**Signs and Symptoms/Clinical Presentation**

› Impaired social functioning: lack of facial expressions and eye contact, lack of interest in participating in age-appropriate activities, inability to share or acknowledge the needs of others, failure to point to objects of interest, refusal to hug or cuddle, and preference to be alone
› Impaired language development: delayed or lack of expressive language, poor receptive language, failure to initiate conversation, and parroting of the spoken words of others
› Stereotypical behaviors: insistence on a routine without interruptions or changes, extreme food fussiness, repetitive play with the same toy in the same fashion, preoccupation with or attachment to one or more specific objects, walking on tiptoes, echolalia (i.e., unsolicited, meaningless repetition of noises or phrases), palilalia (i.e., a complex tic, characterized by involuntary repetition of syllables, words, or phrases), and repetitive clapping, hand flapping, finger flicking, body rocking, or hand banging

**Assessment**

› **Patient History**
  • Ask about prenatal and early neonatal history; medical, mental health, feeding, behavioral, and medication history; hearing and speech disorders; and family history of medical, mental health, and developmental disorders
  – Children with ASD are more likely than typically developing children to be overweight and to have gastrointestinal disorders (e.g., chronic constipation or diarrhea)
› **Physical Findings of Particular Interest**
  • Physical examination can identify nonspecific neurologic manifestations (e.g., primitive reflexes, delayed development of hand dominance), abnormal motor movement (e.g., clumsiness, awkward gait, hand flapping, tics), small head circumference at birth, dermatologic anomalies (e.g., aberrant palmar creases), and indications of self-injurious behaviors (e.g., picking at skin, self-biting, head slapping) or abuse by others
› **Laboratory Tests**
  • Although there are no laboratory tests specific to the diagnosis of ASD, laboratory tests can be performed to assess for other causes of manifestations (e.g., lead poisoning)
  • Chromosomal microarray can be performed to evaluate for chromosomal abnormalities associated with ASD
› **Other Diagnostic Tests/Studies**
  • The Autism Diagnostic Observation Schedule-Generic (ADOS-G), Autism Behavior Checklist (ABC), Childhood Autism Rating Scale (CARS), and Checklist for Autism in Toddlers (CHAT) can be administered to assess the patient, and IQ testing can be ordered
  • Speech, language, and audiologic assessment is performed to assess for hearing deficits
  • Neurologic examination is important to evaluate for seizure disorders

**Treatment Goals**

› Increase Adaptive Behavior and Promote Physical and Emotional Well-Being
• Assess patient emotional and physiologic status and provide 1:1 staffing and nurse continuity whenever possible; ask parents/other family members for information about how to successfully communicate and about the patient’s preferred routines and particular dislikes
  – Establish patient trust and provide praise for patient responses
  – Use short, concrete directives and make one request at a time (e.g., “sit on the bed”)
  – Limit patient choices and provide repeated advance information about changes in routine
  – As appropriate, use distraction such as singing, counting, or offering a toy
  – Decrease auditory and visual stimuli to limit distractibility
  – Have parents/caregivers hold the patient during the provision of care or a procedure, as appropriate, and give rewards/tokens after care and procedures
• Request referral to a mental health clinician, speech-language pathologist, dietitian, and/or occupational therapist, as appropriate
  – Because of inflexibility to change and sensory regulatory difficulties, many individuals with ASD have limited food preferences; systematically introduce new foods assisted by dietitian and/or parents, as appropriate
• Administer prescribed anticonvulsants for seizures and antidepressants (e.g., selective serotonin reuptake inhibitors [SSRIs]; e.g., FLUoxetine, citalopram, escitalopram), antipsychotics (e.g., risperiDONE, ARIPiprazole, ziprasidone), and stimulants (e.g., methylphenidate) for depression, aggression, anxiety, self-injury, and irritability. Monitor closely for adverse effects and educate parents/caregivers about potential adverse effects

Provide Emotional Support and Educate Patient/Patient’s Family
• Assess patient/parent anxiety and for knowledge deficits regarding ASD; provide emotional support and educate about ASD, potential complications, treatment risks and benefits, and individualized prognosis
• Request referral to a social worker for identification of local resources for educational support, behavioral modification or other programs for treating ASD, family therapy家长 support groups, a psychologist for cognitive testing, and outpatient occupational therapy to increase patient independence with ADLs

Food for Thought
› Vaccines are not a risk factor for ASD. In fact, the 1998 Lancet article that first suggested an association between the measles, mumps, and rubella (MMR) vaccine and autism has been retracted
› The U.S. Preventive Services Task Force cites insufficient evidence to recommend screening of young children (ages 18–30 months) for ASD when concerns have not been raised by the child’s parents or clinician
› Maternal antidepressant use during pregnancy appears to be associated with increased risk of ASD in children. Researchers in Sweden conducted a prospective study of 254,610 children and found that maternal use of antidepressants during pregnancy was associated with a 1.45-fold increased risk of ASD, particularly ASD without intellectual disability. Paternal antidepressant use during the mothers’ pregnancy did not affect ASD risk (Rai et al., 2017)
› Prenatal vitamin use might reduce risk of ASD in children. In another study in Sweden, investigators evaluated 273,107 children and concluded that in utero exposure to multivitamin supplementation reduced risk of ASD with intellectual disability by 31%, but did not affect risk of ASD without intellectual disability (DeVilbiss et al., 2017)
› Use of complementary and alternative medicine (CAM) is common in children and adolescents with ASD. The authors of a recent systematic review identified 20 studies with a total of 9,540 participants and found that the prevalence of CAM use was 28–95% in these studies. Special diets and dietary supplements were the most commonly used CAM treatments used (Höfer et al., 2017)

Red Flags
› Children with ASD are at increased risk for physical and sexual abuse and often lack the language skills necessary to report inappropriate behavior
› Early signs of ASD in infants include poor eye contact and failure to respond to their name
› Children with ASD experience extreme anxiety and can react negatively or with aggressiveness to noisy environments or new situations; hypersensitivity to tactile, visual, or auditory stimuli can cause resistant behavior and/or outbursts toward medical personnel

What Do I Need to Tell the Patient/Patient’s Family?
› Encourage parents to enroll the child with ASD in a behavioral modification program at school or in the community, learn about behavior modification in the home, join a support group to benefit from social support systems and resources, and
continue treatment with a speech-language pathologist and an occupational therapist who are knowledgeable about sensory integration

For more information, contact the Autism Society at http://www.autism-society.org

References