Chapter 6: Autism Spectrum Disorder

# 6.1 Description & Epidemiology

## What is ASD?

* ASD is a disorder that emerges in early childhood and is characterized by deficits in social communication and the presence of restricted or repetitive behaviors, interests, or activities.
* Deficits in social communication include (1) a lack of reciprocity in social interactions, (2) problems with nonverbal communication such as poor eye contact, and (3) a lack of interest in interpersonal relationships or problems making and keeping friends.
* Restricted or repetitive behaviors include (1) stereotyped speech or movement, (2) resistance to change, (3) fixated interests, and (4) unusually high or low sensitivity to sensory stimulation.
* When diagnosing ASD, clinicians can specify the severity of social communication deficits and restricted/repetitive behaviors, the presence of language impairments, or coexisting medical or mental disorders.

## What Disorders Frequently Occur with ASD?

* Approximately 44% of school-age children with ASD may meet criteria for Intellectual Disability.
* Approximately 25% of children with ASD are mute and approximately 50% more youths with ASD show problems with speech or language.
* Children who show deficits in social communication, but do not display restricted or repetitive interests or behaviors would be diagnosed with Social (Pragmatic) Communication Disorder rather than ASD.
* Approximately 70% of youths with ASD have a comorbid mental disorder (e.g., ADHD, anxiety) and 70% have an associated medical problem (e.g., GI problems, seizures).

## How Common is ASD?

* The ADDM Network, which reviews medical and school records, estimates the prevalence of ASD as 1 in 68 youths. Data from the NHIS, which is based on parent report alone, estimates prevalence at 1 in 48 youths. These data suggest that ASD occurs in 1.5% to 2.1% of children.
* Boys are 4.0-4.5 times more likely than girls to be diagnosed with ASD. Girls with ASD, however, tend to show greater impairment than boys.
* ASD is disproportionately diagnosed in non-Latino, White youths and children from higher-SES families. Recognition of ASD and access to high-quality educational and medical services may partially explain these demographic differences in prevalence.

## What is the Prognosis for Children with ASD?

* Approximately two-thirds of children eventually diagnosed with ASD display signs of the disorder by age 18 months. Early indicators include a lack of eye contact, failure to initiate social interactions, and problems with joint attention and imitation.
* Approximately one-third of children eventually diagnosed with ASD do not show signs of the disorder until after age two years, when social communication deficits and restrictive, repetitive behaviors or interests emerge.
* Prognosis for youths with ASD is variable. The best outcomes are seen in children with (1) higher intellectual ability, (2) better language skills, and (3) greater initial social engagement.

# 6.2 Causes

## Is ASD Heritable?

* ASD is heritable; approximately 40% of the variance in children’s ASD symptoms are attributable to genetic causes.
* The Autism Gene Project has identified several leading causes for ASD: deletions or duplications of genetic material (15%), specific genetic mutations (7%), and metabolic disorders (2-3%). Unfortunately, the causes of 75% of cases of ASD cannot be determined at this time.
* Older men are at greater risk for offspring with ASD than younger men. Explanations for their increased risk include genetic mutations in sex cells, exposure to environmental toxins over the man’s lifespan, and epigenetic effects.

## What Brain Abnormalities are Associated with ASD?

* The growth dysregulation hypothesis indicates that youths with ASD show abnormal maturation of the cortex, large head circumference, and high synaptic density in early childhood. In contrast, youths with ASD show poor neural connectivity, especially in brain regions responsible for social communication and language.
* Some youths with ASD show smaller and less active functioning of the amygdala, a portion of the limbic system responsible for emotional processing.
* Children with ASD often show underactivity of the right fusiform gyrus, a region responsible for processing human faces and interpreting social behavior.
* Many youths with ASD display underactivity of the prefrontal cortex, a region responsible for executive functioning. These youths have difficulty attending to important social information and adapting to changes in their environment.

## What Problems with Social Cognition Characterize ASD?

* Infants later diagnosed with ASD often show delays in joint attention and social orientation during the first 24 months of life. These deficits cause them to miss out on social and linguistic information and can lead to later deficits.
* Toddlers diagnosed with ASD often show delays in symbolic (pretend) play. Symbolic play is a precursor to later language development.
* Many 3- and 4-year-olds later diagnosed with ASD show deficits in theory of mind, that is, the ability to appreciate the mental states of others. These deficits likely contribute to problems interpreting others’ social behaviors and problems with empathy.

# 6.3 Identification, Prevention, & Treatment

## How Is ASD Identified and Diagnosed?

* Although deficits in social communication usually emerge between 6 and 12 months, most parents become concerned when their children show delays in language and symbolic play between 12 and 18 months of age.
* The American Academy of Pediatrics recommends universal screening for ASD at 18 months. The M-CHAT-F/F is a frequently-used screening instrument.
* Formal ASD diagnosis is made by an interdisciplinary team of professionals. Clinicians tend to rely on interviews, such as the ADI-R, and observations, such as the ADOS-2.

## What Treatments Are Effective for Preschoolers and School-age children?

* EIBI relies on discrete trial training to improve the social communication and language skills of young children with ASD. EIBI is associated with increases in IQ and social functioning when administered by well-trained therapist on a frequent basis.
* Pivotal response training is a behavioral intervention in which parents use direct reinforcers to increase children’s motivation to engage in social interactions. A secondary goal of the program is to improve children’s ability to direct and manage their own behavior.
* TEACCH is a classroom-based intervention that relies on a highly structured environment and scaffolding to improve social communication and reduce challenging behaviors in children with ASD.

## What Treatments Are Effective for At-risk Infants and Toddlers?

* Prevention programs seek to improve the early social communication skills of children at-risk for ASD.
* Reciprocal Imitation Training (RIT) teaches imitation skills. Therapists imitate children’s play and reinforce children for imitating gestures and other actions.
* JASPER relies on discrete trial training to improve joint attention and symbolic play in young children at-risk for ASD. Treatment can be delivered by therapists, parents, or teachers.

## Is Medication Effective for Youths with ASD?

* Medication is typically used to reduce challenging behaviors and comorbid mental disorders experienced by youths with ASD.
* The atypical antipsychotic medications aripiprazole (Abilify) and risperidone (Risperdal) are useful in reducing irritability, self-injury, and physical aggression in children with ASD.
* Medications are also sometimes prescribed to children with ASD for comorbid ADHD or sleep problems.

## How Can Clinicians Improve Communication in Youths with ASD?

* AAC systems can be used to supplement the communication skills of children with ASD as they acquire spoken language or compensate for spoken language in children who are mute.
* PECS is a low-tech system in which children communicate by pointing to or exchanging cards with symbols or pictures that represent actions, feelings, ideas, or objects.
* Many youths with ASD and language deficits rely on speech generating devices or visual scene displays to communicate with others.

## What Interventions have Limited Empirical Support?

* Many popular interventions for ASD, such as special diets, sensory integration training, and equine assisted therapy have limited empirical support.
* Parents may select therapies with limited support because they are not familiar with the research literature, because their child did not improve in traditional therapy, or because evidence-based treatments are not available in many communities.
* Best practices for ASD include early identification and treatment, intensive and structured learning opportunities, low student-to-teacher ratios, and high parental involvement.