A n estimated 1% of U.S. children have been diagnosed with Autism Spectrum Disorders (ASDs). During the past year, ASDs have garnered considerable interest from the medical community, the public and the media. Prominent examples include the debunking of the fraudulent research reported by Dr. Andrew Wakefield associating autism with childhood vaccines; and recent approval by the American Psychiatric Association of new criteria for ASDs (eliminating Asperger’s syndrome as a separate category) that will be published in the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5).

To help shed some light on ASDs, we devote this CD Summary to describing (briefly) what is known about their diagnosis and screening; epidemiology; and pathogenesis. 

**DIAGNOSIS AND SCREENING**

Diagnosing ASDs is difficult at best. The specific criteria for the diagnosis of ASDs is beyond the scope of this CD Summary; but we can suggest some resources.

The American Academy of Pediatrics (AAP) guidelines for the evaluation and management of children with ASDs recommend developmental screening of all children at 18 and 24 months of age, because these are critical times for early social and language development, and because of the relative effectiveness of early intervention. Early screening is thought to be important because the brain circuitry that supports social and language behavior is rapidly developing and is shaped by early childhood experience. Although there is no cure, early intervention may improve the skills of children with ASD. One of the first autism symptoms to emerge during infancy is a lack of attention to other people. Early intervention is designed to draw the child’s attention to others and to engage him in pleasurable interactions, thereby increasing the opportunities for learning and for a more normal trajectory of brain development.

The Oregon Pediatric Society’s Screening Tools And Referral Training (START) project attempts to increase standardized health screening so that primary care providers can identify children and families who need services earlier; and to enhance coordination of care and communication, so that children and families receive appropriate health care. START recommends the 23-item Modified Checklist for Autism in Toddlers (M-CHAT) to screen for autism (according to AAP recommendations) at all 18- and 24-month well-child visits. M-CHAT should be administered during a face-to-face interview with a parent.

**EPIDEMIOLOGY**

Although no reliable Oregon-specific data exist, the reported prevalence of ASD in the United States and other developed countries has increased since the 1970s, and particularly since the late 1990s. Most studies from the mid-1990s found a prevalence of approximately 1 per 1,000 for autism and 2 per 1,000 for ASD, compared with 0.5 per 1,000 in previous decades.

More recently, the CDC Autism and Developmental Disabilities Monitoring Network (which identified ASDs through screening and abstraction of existing health and education records that documented behaviors consistent with the DSM-IV criteria), found an ASD prevalence of 11 per 1,000 at age eight in 2008.

Although the prevalence of children diagnosed with ASD has been increasing, it is unclear whether this is a true increase, or is attributable to changes in care seeking, diagnostic criteria, or study methodology. Systematic reviews of the epidemiologic studies of autism have found evidence that changes in case definition and increased awareness account for much of the apparent increase in prevalence.

Other factors that may play a role include earlier detection, availability of more specialized developmental services, and “diagnostic substitution.” The latter implies that, if services are available for children diagnosed specifically with ASD as opposed to another condition, providers may be more likely to report the ASD diagnosis.

Boys are 4.6 times as likely as girls, and non-Hispanic White children more likely than Hispanic and non-Hispanic Black children to be diagnosed with ASD. The risk of having a child with ASD increases with each 10-year increase in maternal age (adjusted relative risk [RR]=1.3; 95% confidence interval [CI]=1.1–1.5) and paternal age (RR=1.3; 95% CI=1.1–1.5).2

**PATHOGENESIS**

The pathogenesis of ASD is incompletely understood. The general consensus is that ASD has a genetic etiology, which alters brain development, affecting social and communication development and leading to restricted interests and repetitive behavior. Environmental factors, acting through epigenetic mechanisms (i.e., genetic transmission of characteristics not in the genes themselves, but rather by gene modification), may turn on abnormal genes early in fetal development, thereby predisposing to ASD. Much more work is needed to evaluate factors that affect ASD prevalence over time. CDC and the Autism and Developmental Disabilities Monitoring Network investigators continue to explore these factors, focusing on understanding disparities in the identification of ASDs among certain subgroups and evaluating the increase in reported ASD prevalence.

**CHANGES IN DSM-5**

The American Psychiatric Association recently voted to release the DSM-5, its first major revision in 17
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years. The psychiatrists’ association is wrestling with the question of where to draw the line between unusual and abnormal, and its decisions are sure to be wrenching for some families. Under the current criteria, a person can qualify for the diagnosis by exhibiting any six of 12 specified behaviors; under the proposed definition, the person would have to exhibit three deficits in social interaction and communication, and at least two repetitive behaviors. The proposed changes may exclude some higher functioning persons who meet current diagnostic criteria.

RESOURCES

The Oregon Commission on Autism Spectrum Disorder (see www.orcommissionasd.org/) was established to develop appropriate, collaborative and timely supports and services across the lifespan. The Commission has made recommendations to the Governor regarding the diagnosis of autism, the lack of consistency between the medical diagnosis and educational classification for autism, and the importance of closer communication and collaboration between healthcare and education professionals for the benefit of children with autism and their families.

CDC, in collaboration with the American Academy of Pediatrics and the American Academy of Neurology, developed the autism “A.L.A.R.M.” initiative to establish standard practices among physicians, to simplify the screening process, and to ensure that all children receive routine and appropriate screenings and timely interventions (see www.medicalhomeinfo.org/downloads/pdfs/AutismAlarm.pdf, www.cdc.gov/autism and www.aap.org/autism). The acronym admonishes as follows:

• Autism is prevalent: One of every six children is identified with a developmental disorder or behavioral problem; approximately one of every 88 children is diagnosed with an ASD.

• Listen to parents: Early signs of autism are often present before 18 months of age; parents usually do have concerns that something is wrong; and generally do give accurate and quality information.

• Act early: Make screening and surveillance an important part of your practice; know the subtle differences between typical and atypical development; learn to recognize red flags; use validated screening tools, and identify problems early.

• Refer to: Early intervention or a local school program (do not wait for a diagnosis); an autism specialist, or team of specialists, for a definitive diagnosis; audiology to rule out a hearing impairment; local community resources for family support.

• Monitor: Schedule a follow-up appointment to discuss concerns; look for other conditions (e.g., seizures) known to be associated with autism; educate parents and provide them with up-to-date information; advocate for families with local early intervention programs, schools, respite care agencies, and insurance companies; watch for additional signs of autism or other developmental disorders; continue to provide a medical home.

REFERENCES


