

# From the CDC: Understanding Autism Spectrum Disorder

An evidence-based review of ASD risk factors, evaluation, and diagnosis.

**ABSTRACT:** Autism spectrum disorder (ASD) is a condition characterized by impaired social communication as well as restricted and repetitive behaviors. It is considered a neurodevelopmental disorder because it is associated with neurologic changes that may begin in prenatal or early postnatal life, alters the typical pattern of child development, and produces chronic signs and symptoms that usually manifest in early childhood and have potential long-term consequences. In past decades, autism was conceptualized as a strictly defined set of behaviors, usually accompanied by intellectual impairment. Today, it is recognized as a spectrum, ranging from mild to severe, in which behaviors vary substantially and the majority of children who fall on the spectrum have average to above average intellectual ability. Here, the authors discuss the risk factors for ASD, its epidemiology, common concurrent conditions, evaluation, diagnosis, treatments, and outcomes.

**Keywords:** autism spectrum disorder, epidemiology, impaired social communication, neurodevelopmental disorders, repetitive behaviors, risk factors, treatment

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that typically manifests in early childhood as impaired social communication and restricted, repetitive behaviors and falls on a spectrum ranging from mild to severe.<sup>1</sup> For example, some people with ASD are nonverbal or speak only in simple sentences, while others are verbally skilled but have problems with social communication and pragmatic language, such that they respond inappropriately in conversation, misunderstand nonverbal communication, or lack age-appropriate competency to establish friendships. People with ASD may have difficulty adapting to changes in their routine or environment. Their interests, which are typically intense, may be limited. Some display stereotyped, repetitive motor movements or unusual sensory responses. Current diagnostic criteria for ASD can be found in the *Diagnostic and Statistical Manual of Mental Disorders*, fifth edition.<sup>1</sup>

## RISK FACTORS

ASD etiology is not completely understood, but multiple factors likely contribute to ASD development.<sup>2</sup> Neurologic changes that result in ASD may begin in prenatal and early postnatal life,<sup>3</sup> and genetic factors (both rare and common variants) are a source of population variation in ASD-related behaviors.<sup>4,5</sup>

**Sibling recurrence risk.** Studies have reported that roughly 15% to 20% of younger siblings of children with ASD meet the diagnostic criteria for ASD themselves.<sup>6,7</sup> Concordance is higher among monozygotic compared with dizygotic twins.<sup>8,9</sup>

**Other prenatal and perinatal risk factors** have been identified, including

- prenatal exposure to valproic acid<sup>10</sup> or thalidomide,<sup>11</sup> and rubella infection.<sup>12</sup>
- advanced parental age.<sup>13,14</sup>
- maternal gestational diabetes and bleeding.<sup>13</sup>

# Your Child at 18 Months (1½ Yrs)★



Child's Name \_\_\_\_\_

Child's Age \_\_\_\_\_

Today's Date \_\_\_\_\_

Milestones matter! How your child plays, learns, speaks, acts, and moves offers important clues about his or her development. Check the milestones your child has reached by 18 months. Take this with you and talk with your child's doctor at every well-child visit about the milestones your child has reached and what to expect next.

## What Most Children Do by this Age:

### Social/Emotional

- ☐ Likes to hand things to others as play
- ☐ May have temper tantrums
- ☐ May be afraid of strangers
- ☐ Shows affection to familiar people
- ☐ Plays simple pretend, such as feeding a doll
- ☐ May cling to caregivers in new situations
- ☐ Points to show others something interesting
- ☐ Explores alone but with parent close by

### Language/Communication

- ☐ Says several single words
- ☐ Says and shakes head "no"
- ☐ Points to show someone what he wants

### Cognitive (learning, thinking, problem-solving)

- ☐ Knows what ordinary things are for; for example, telephone, brush, spoon
- ☐ Points to get the attention of others
- ☐ Shows interest in a doll or stuffed animal by pretending to feed
- ☐ Points to one body part
- ☐ Scribbles on his own
- ☐ Can follow 1-step verbal commands without any gestures; for example, sits when you say "sit down"

### Movement/Physical Development

- ☐ Walks alone
- ☐ May walk up steps and run
- ☐ Pulls toys while walking
- ☐ Can help undress herself
- ☐ Drinks from a cup
- ☐ Eats with a spoon

## You Know Your Child Best.

Act early if you have concerns about the way your child plays, learns, speaks, acts, or moves, or if your child:

- ☐ Is missing milestones
- ☐ Doesn't point to show things to others
- ☐ Can't walk
- ☐ Doesn't know what familiar things are for
- ☐ Doesn't copy others
- ☐ Doesn't gain new words
- ☐ Doesn't have at least 6 words
- ☐ Doesn't notice or mind when a caregiver leaves or returns
- ☐ Loses skills he once had

**Tell your child's doctor or nurse if you notice any of these signs of possible developmental delay and ask for a developmental screening.**

If you or the doctor is still concerned

1. Ask for a referral to a specialist and,
2. Call your state or territory's early intervention program to find out if your child can get services to help. Learn more and find the number at [cdc.gov/FindEI](http://cdc.gov/FindEI).

For more information, go to [cdc.gov/Concerned](http://cdc.gov/Concerned).

**DON'T WAIT.**  
**Acting early can make a real difference!**

### ★ It's time for developmental screening!

At 18 months, your child is due for general developmental screening and an autism screening, as recommended for all children by the American Academy of Pediatrics. Ask the doctor about your child's developmental screening.



[www.cdc.gov/ActEarly](http://www.cdc.gov/ActEarly)  
1-800-CDC-INFO (1-800-232-4636)



Download CDC's  
Milestone Tracker App



**Learn the Signs. Act Early.**

- neonatal complications, including low birth weight and small size for gestational age.<sup>15</sup>
- preterm birth.<sup>16</sup>

Although there's evidence that any of these factors, which can negatively affect prenatal and perinatal health, may increase the risk of ASD, no single prenatal or perinatal factor has been found to have more than a modest association with ASD.<sup>15</sup> Additional reviews and meta-analyses of research on ASD risk factors have been published.<sup>17-19</sup> Research into risk factors for ASD is ongoing, including through such case-control studies as the Centers for Disease Control and Prevention (CDC) Study to Explore Early Development (SEED; see [www.cdc.gov/ncbddd/autism/seed.html](http://www.cdc.gov/ncbddd/autism/seed.html)). Studies have shown that there is no link between receiving vaccines and developing ASD, as is discussed in the evidence-based meta-analysis of case-control and cohort studies by Taylor and colleagues.<sup>20</sup> Additional information on vaccine safety is available from the CDC at [www.cdc.gov/vaccinesafety/concerns/autism.html](http://www.cdc.gov/vaccinesafety/concerns/autism.html).

has narrowed in recent years, possibly as a result of more effective outreach directed at racial and ethnic minority communities and improved access to diagnostic services.<sup>23, 26</sup>

**Other contributing factors** to the recent rise in estimated ASD prevalence include

- changes in ASD diagnostic criteria, clinical practices for identifying and diagnosing children with developmental delays, and reporting practices.<sup>27-30</sup>
- improved access to ASD services through better insurance coverage.<sup>30-32</sup>
- the inclusion of children with high intellectual ability and few or mild ASD symptoms.<sup>26, 28</sup>

The heterogeneity of ASD prevalence estimates across geographic areas<sup>26</sup> further supports the premise that regional differences in evaluation, diagnosis, clinical and reporting practices, and service access may affect calculated prevalence.

**Prevalence trends by state.** The CDC has recently introduced an Autism Data Visualization Tool (see [www.cdc.gov/ncbddd/autism/data](http://www.cdc.gov/ncbddd/autism/data)), which

## Diagnosing ASD can be challenging.

To date, there is no biomarker or medical test that can distinguish those with ASD from those without.

### EPIDEMIOLOGY

Recent national surveys suggest that 2% to 3% of children ages three through 17 have a current or previous diagnosis of ASD.<sup>21, 22</sup> A review of data from 2000 through 2014 on eight-year-old children in selected U.S. communities by the CDC's Autism and Developmental Disabilities Monitoring (ADDM) Network showed that ASD prevalence estimates rose more than 150% over this period, from 6.7 per 1,000 in 2000 to 16.8 per 1,000 in 2014.<sup>23</sup>

**Sex differences.** CDC data show that ASD prevalence is four times greater in boys than in girls.<sup>23</sup> Higher prevalence among boys may be related to differences in biological susceptibility to ASD<sup>24</sup> or to less frequent or incomplete identification of ASD in girls because girls with ASD have less well-recognized symptom profiles or higher intellectual ability, better language skills, and perceived better social skills.<sup>25</sup>

**Racial/ethnic differences.** Historically, CDC surveillance reports had estimated higher ASD prevalence among white children compared with both Black and Hispanic children, possibly because of a failure to identify ASD among children across all racial and ethnic groups. Although ASD prevalence estimates continue to be higher among white than among Black and Hispanic children, the disparity

provides information about trends in ASD prevalence by state.

**ASD in adults.** The CDC does not collect prevalence data on ASD in adults; however, a population-based survey of adults in the United Kingdom (UK) estimated that approximately 1% met the study criteria for ASD, though most had never been formally diagnosed.<sup>33</sup> A follow-up analysis of these data combined with data collected from participants in the Intellectual Disability Case Register study found a similar combined prevalence rate (1.1%) among adults.<sup>34</sup> While ASD is usually diagnosed in childhood, some people first seek diagnosis in adulthood. Another UK study reported on 255 adults referred to the Autism Diagnostic Research Centre for neuropsychological assessment, 100 of whom were subsequently diagnosed with ASD.<sup>35</sup> Only four of these had a learning disability, as identified through the education system or a recent diagnosis (intelligence quotient [IQ] below 70). It's not clear why those found to have ASD were not diagnosed earlier in life, but the researchers suggest that comorbid psychiatric diagnoses, which affected 58%, may have been factors, as psychiatric conditions such as anxiety and depression may have concealed ASD traits, delaying appropriate referral.

## COMMON CONCURRENT CONDITIONS

Although the proportion of children identified with ASD and concurrent intellectual disability has declined over time,<sup>26</sup> suggesting improved identification of ASD in children with a high level of intellectual ability, a substantial proportion of people with ASD have concurrent intellectual disability. The most recent CDC data indicate that nearly one-third of eight-year-old children with ASD had an IQ within the range of intellectual disability (70 or below).<sup>23</sup>

Other conditions that commonly occur with ASD include the following<sup>36</sup>:

- motor abnormalities, up to 79%
- attention deficit–hyperactivity disorder (ADHD), 28% to 44%
- gastrointestinal problems, 9% to 70%
- sleep problems, 50% to 80%
- aggressive behavior, up to 68%
- anxiety, 42% to 56%
- depression, 12% to 70%

**Associated pediatric conditions** may include language delay, which occurs in up to 87% of three-year-olds with ASD,<sup>37</sup> or language regression (for example, children's loss of their first few words or the development of severely impaired receptive–expressive language).<sup>38</sup> The risk of children with ASD developing epilepsy is greatest before the age of five and around the time of puberty and is greater in children with concurrent intellectual disability.<sup>38</sup>

**Neuropsychological and medical conditions**, like the core features of ASD, may interfere with health, functioning, and relationships with family members and peers. The complex health care needs of people with ASD are best addressed through the medical home model of care, which is defined by the American Academy of Pediatrics (AAP; see <https://medicalhomeinfo.aap.org/overview/Pages/Whatisthemedicalhome.aspx>). It is important to consider the person's ASD and concurrent symptoms when conducting the history and physical evaluation, weighing treatment plans, and coordinating referrals for medical evaluation and care.<sup>39</sup>

**Risky behavioral issues.** In addition to associated medical conditions, people with ASD, particularly those with intellectual disability, may display risky behaviors such as self-injury (up to 50%)<sup>36</sup> and wandering, which has been reported by parents to occur in 37.7% of children who have both ASD and intellectual disability and 32.7% of children who have ASD without intellectual disability.<sup>40</sup> These behaviors may pose safety risks and generate considerable stress for both people with ASD and their families. The CDC provides safety information and resources on these and other potential dangers facing children with special needs at [www.cdc.gov/ncbddd/disabilityandsafety/index.html](http://www.cdc.gov/ncbddd/disabilityandsafety/index.html). Families may also find toolkits from Autism Speaks to help them address challenging behaviors

at [www.autismspeaks.org/family-services/tool-kits/challenging-behaviors-tool-kit](http://www.autismspeaks.org/family-services/tool-kits/challenging-behaviors-tool-kit).

Nurses can help facilitate the coordination of treatment and safety approaches to challenging behaviors across home and community settings, including schools. Children with disabilities, including ASD, may be at increased risk for maltreatment, including neglect and physical abuse due to caregiver stress. Nurses should be prepared to recognize the signs of maltreatment and intervene when necessary. The Child Welfare Information Gateway, a service of the Children's Bureau of the Administration for Children and Families at the U.S. Department of Health and Human Services (HHS), provides several resources for health care professionals on child maltreatment at [www.childwelfare.gov/topics/preventing/developing/collaboration/professionals](http://www.childwelfare.gov/topics/preventing/developing/collaboration/professionals).

## EVALUATION AND DIAGNOSIS

Diagnosing ASD can be challenging. To date, there is no biomarker or medical test that can distinguish those with ASD from those without.

To make a diagnosis, health care professionals rely on

- developmental history.
- parent, caregiver, or self-reported responses to questions about ASD-related behaviors.
- direct observations of behavior.

Concerns initially reported by parents or caregivers of children with ASD often include

- language delays or unusual language usage.
- atypical social responses, such as difficulty initiating and sustaining interactions with other children or not responding to their name being called.
- repetitive behaviors, such as resistance to change.
- emotional and behavioral reactivity.

**High-risk infants.** Data suggest that in high-risk infants (such as those who have an older sibling with ASD), the characteristic signs of ASD, such as social communication difficulties and repetitive behaviors, would usually become apparent between the ages of 18 and 36 months if they too have ASD.<sup>3</sup> However, there may be prodromal behaviors that emerge in the first year, including difficulties with emotional regulation,<sup>41</sup> lack of response to bids for attention, inconsistent face gazing, and impaired motor control.<sup>3</sup> These signs may occur before the more easily recognized signs of ASD are apparent and may go unrecognized by parents, caregivers, and health care providers as potential indications of ASD. If, however, parents or caregivers raise concerns about these features with health care providers, it is important that providers take such observations seriously.

**Early identification.** Efforts by public, pediatric, and other health organizations have focused on identifying children with ASD as early as possible to facilitate prompt treatment and behavioral intervention. CDC data have indicated that, for nearly

all children with ASD, developmental concerns were documented by age 36 months, though there has been little progress in lowering the age of first ASD evaluation.<sup>23</sup> In some cases, ASD can reliably be diagnosed by age two,<sup>37</sup> though the stability of early diagnoses depends on the experience of the diagnosing clinician. Because children with ASD display both typical and atypical behaviors, an average health visit may not allow enough time to observe a child's atypical behavior.<sup>42</sup>

**Recommendations of the AAP.** To address the complexity of identifying ASD at an early age, the AAP recommends that all children receive ASD-specific screening with a standardized ASD screening test at ages 18 months and 24 months, or whenever concerns arise, and that developmental surveillance occur at each health visit.<sup>39</sup>

**Developmental surveillance,** a flexible, ongoing process of assessment that continues as the child grows, involves the following steps<sup>39</sup>:

- asking parents or caregivers about concerns they may have regarding their child's development, and listening and responding to these concerns
- obtaining and documenting the child's developmental history

- noting findings based on informed observation of the child
- identifying potential risks, strengths, and supportive factors in the child's medical and life history
- maintaining an accurate record of the surveillance and screening activities
- seeking input from and sharing observations and opinions with other health care professionals and educators outside the medical home (for example, with specialty providers or preschool teachers), with the consent of the patient or caregiver

Several online resources are available to assist health care providers in conducting developmental surveillance and to help parents of children in their practice track their child's developmental milestones (see *Developmental Surveillance Resources*).

Nurses often play a critical role in surveillance, coordination, and championing the efforts of the health care team through the following actions:

- taking the developmental history
- eliciting parents' concerns
- sharing observations of the child with the primary care provider
- distributing and scoring age-appropriate screens
- informing the primary care provider of screening results for discussion with the family
- submitting and following up on ordered referrals
- recognizing a pattern of early childhood development consistent with ASD in older children and adults, whose difficulty in developing and maintaining friendships, communicating, and understanding what behaviors are expected in school or on the job may suggest undiagnosed ASD
- identifying concurrent conditions that often affect people with ASD
- referring parents of children with ASD, or adults with ASD that was undiagnosed in childhood, to services and specialists

#### **Early intervention and special education.** If

ASD risk is indicated on a validated screening tool, or if the provider or parent is concerned the child might have ASD despite normal screening results, the child should be referred promptly for further developmental and medical evaluation as the screening tool may have produced a false negative or the child may have other developmental delays that should be addressed.<sup>43</sup> Children under age three can be referred to the state's early intervention program (see [www.cdc.gov/ncbddd/actearly/parents/states.html](http://www.cdc.gov/ncbddd/actearly/parents/states.html) for information on early intervention). Patients ages three through 21 can, through the Individuals with Disabilities Education Act (IDEA), receive evaluations and services through their local school district's special education program. Referral for further developmental evaluation, audiological testing, and assessment for early intervention or special education services can all occur simultaneously. Developmental

### **Developmental Surveillance Resources**

- *Developmental Surveillance: What, Why and How*, a video for health care providers from the American Academy of Pediatrics (AAP), available at: [www.youtube.com/watch?v=sceYLUHhgnU&feature=youtu.be](http://www.youtube.com/watch?v=sceYLUHhgnU&feature=youtu.be)
- Milestone Tracker, a free app from the Centers for Disease Control and Prevention (CDC) that helps parents identify their children's developmental milestones and provide support at every stage: [www.cdc.gov/MilestoneTracker](http://www.cdc.gov/MilestoneTracker)
- "Learn the Signs. Act Early" materials from the CDC, which include checklists and videos that can assist providers with developmental surveillance by encouraging parents to monitor their child's development between health care visits and discuss any concerns: [www.cdc.gov/ncbddd/actearly/milestones/index.html](http://www.cdc.gov/ncbddd/actearly/milestones/index.html)
- Autism Diagnosis Criteria: DSM-5 from Autism Speaks, available at: [www.autismspeaks.org/autism-diagnosis-criteria-dsm-5](http://www.autismspeaks.org/autism-diagnosis-criteria-dsm-5)
- Identifying and Caring for Children with Autism Spectrum Disorder: A Course for Pediatric Clinicians from the AAP, available at: <https://shop.aap.org/identifying-and-caring-for-children-with-autism-spectrum-disorder-a-course-for-pediatric-clinicians>



evaluations may be completed by developmental and behavioral pediatricians, child neurologists, child psychologists, and child psychiatrists.

### TREATMENT

Currently, there is no curative treatment for ASD, but interventions may reduce troubling symptoms, improving cognition and function, thereby maximizing the ability of people with ASD to participate in the community. Treatment plans are usually multidisciplinary and tailored to the person's unique strengths and challenges. Some interventions are parent mediated. Behavioral intervention strategies often include social skills training for children and adults and focus on reducing restricted interests and repetitive or challenging behaviors. Occupational, speech, and sensory integration therapy may also be helpful.

Resources, then Topic Areas). Children with ASD may be taught in a self-contained or general education classroom, be placed in an inclusion classroom that combines elements of both, or spend part of the school day in a general education classroom and part in a self-contained or inclusion classroom.

**School nurses** may play a role in a child's treatment plan. For example, they may need to administer medication or assess health problems. School nurses should be aware that children with ASD who experience health problems may have difficulty reporting symptoms of illness or maltreatment and may be challenged by changes in routine such as visiting the nurse's office, undergoing physical examination, and interacting with unfamiliar staff.

**Support for parents.** Providers can direct parents of children with ASD to their state's free parent

## Currently, there is no curative treatment for ASD, but interventions may reduce troubling symptoms, improving cognition and function.

For providers who are inexperienced in treating patients with ASD, especially adult patients, it's important to consider the patient's ASD diagnosis as one of many variables that affect an individual and to learn how to adapt treatment to accommodate the patient's strengths, challenges, and differences.<sup>44</sup>

**ADHD medications.** Although no medications have proven effective in treating the core symptoms of ASD, some may be helpful in reducing concurrent conditions. Medications used to treat ADHD, including methylphenidate (Ritalin and others), atomoxetine (Strattera), and guanfacine (Intuniv), have shown benefit in treating children who have ASD and concurrent ADHD, though they may be less effective and have more adverse effects in these children than in those with ADHD alone.<sup>45-47</sup>

**Two atypical antipsychotic medications,** risperidone (Risperdal) and aripiprazole (Abilify), have been shown in randomized controlled trials to reduce irritability or agitation in children and adolescents with ASD, but patients taking these drugs should be monitored for adverse effects, including weight gain and sedation.<sup>48,49</sup>

**Individualized education programs (IEPs).** Children with ASD often have an IEP or a 504 plan through which they may receive behavioral, speech, or occupational therapy, and other services in the school setting. For information about IEPs, visit the IDEA website at <https://sites.ed.gov/idea> (go to

support organization, which can be found on the website of Family Voices, a national organization and grassroots network of families of children with special health care needs (see <http://familyvoices.org/affiliates>). These state- or territory-based organizations link parents with local resources as well as other parents of children with special needs or developmental disabilities who reside in their community. Families may seek out complementary and alternative therapies and should be encouraged to share and discuss these with their child's provider.

### OUTCOMES IN ADOLESCENCE AND ADULTHOOD

Relatively little is known about how ASD affects outcomes in adulthood, such as level of independence, education, employment, social relationships, community integration, and health status. While for some with ASD, symptom severity decreases over time,<sup>50</sup> studies suggest that outcomes are often poor, especially in the domain of social functioning.<sup>51</sup> A 2012 analysis of data from a nationally representative survey of young adults with ASD, as well as parents and guardians, found that the overall rate of paid employment following high school among young adults with ASD was 55.1%.<sup>52</sup>

**Poorer health and shorter life spans.** There is evidence that adults with ASD have poorer health and shorter life spans than adults without ASD.<sup>53</sup> A medical record review conducted at a large

northern California health care system reported that adults with recent ASD diagnoses had higher frequencies of seizures, hypertension, dyslipidemia, sleep disorders, and psychiatric conditions than sex- and age-matched controls.<sup>54</sup> Another study conducted in the same health care system reported that while utilization of health care services was higher for adults with ASD compared with adults with ADHD and adults with neither condition, women with ASD were less likely to receive gynecologic care and be screened for cervical cancer.<sup>55</sup> Premature mortality among adults with ASD is associated with a variety of medical conditions, including epilepsy, particularly in those with concurrent intellectual disability.<sup>53,56</sup> Substantially higher mortality from suicide has been found in people with ASD, especially women and those without concurrent intellectual disability.<sup>53,57</sup>

#### Adolescent transition to adult medical care.

Pediatric nurses can help adolescents with ASD prepare for the transition to adult medical care. Adult primary care nurses should be aware of the increasing numbers of people diagnosed with ASD as children who are coming into adult medical care, in addition to those who were diagnosed in adulthood, both of whom will require assistance with medical management and referrals, as well as anticipatory guidance regarding health conditions. Got Transition ([www.gottransition.org](http://www.gottransition.org)) provides toolkits and other resources for adolescents, young adults, parents, caregivers, and health care providers helping families with this transition.

#### BEYOND PATIENT CARE: NECESSARY NURSING RESEARCH

ASD is more commonly diagnosed today than it was in the past. In addition to teaching parents and caregivers about developmental milestones, conducting surveillance and screening, assisting with referrals, advocating for appropriate diagnoses, monitoring the effectiveness of treatment plans, and helping families navigate the complex systems of services and resources available for people with ASD, nurses should realize that there is a pressing need for research into interventions and services that can help support people with ASD in securing postsecondary education or training, participating in the workforce, obtaining housing, accessing transportation, and managing their health. A broad range of topics related to ASD outcomes and other issues relevant to primary care are covered in a recent HHS report to Congress, available at [www.hhs.gov/sites/default/files/2017AutismReport.pdf](http://www.hhs.gov/sites/default/files/2017AutismReport.pdf). ▼

For three additional continuing nursing education activities on the topic of autism, go to [www.nursingcenter.com](http://www.nursingcenter.com).

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