



Autism spectrum disorder in primary care

Abstract: Nurse practitioners working in the primary care setting will commonly see children with autism spectrum disorder. It is important for clinicians to be vigilant for subtle developmental signs that can lead to early identification and diagnosis.

This article presents information on assessment, screening, the responsibilities of coordinating services, and ways to support families.

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Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by impaired social communication and social interactions with unusual responses to stimuli, restricted interests, and repetitive behaviors.¹ It is diagnosed through the use of behavioral assessment. With ASD occurring in 1 in 42 males and 1 in 189 females, primary care providers (PCPs) commonly encounter individuals with ASD; however, it is not always easy to identify.^{2,3}

ASD can be diagnosed as early as 15 months of age, but only 20% of children with ASD are identified before age 3, and at least one-third of children are diagnosed after age 6.^{4,7} A delay in diagnosis is concerning, as early intervention leads to the best long-term prognosis.^{8,9}

■ Case scenario

JT presents for his 2-year-old well-child visit. He has missed checkups due to transportation issues. JT still uses a bottle and

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is a picky eater, preferring only macaroni and cheese. His mother describes difficulties with toilet training and bedtime. Bedtime is an exhausting nightly ordeal, and she must lie down with him to get him to sleep. The child lives in a bilingual home, but family members have noticed only a few recognizable words, and his mother cannot think of any two-word phrases in JT's repertoire. On exam, he starts to scream and squirm, making evaluation difficult.

This scenario describes many toddlers: picky eaters, fearful, poor sleepers, and having limited language skills. These findings could be considered "typical toddler behavior," but they warrant thorough assessment. Providers who too quickly dismiss these signs as normal may overlook red flags for ASD and other developmental disabilities. How can the NP determine if this is a typically developing child or a child to refer for further evaluation?

■ Assessment in primary care

Identifying children at risk for ASD in primary care settings requires a multipronged approach, including:

- reviewing a developmental history for risk factors (low birth weight, prematurity, polyhydramnios, prenatal infections with fever, multiple births, in vitro fertilization, advanced maternal and paternal age, large discrepancy



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between maternal and paternal age, and less than 18 months between deliveries)

- reviewing family history with heightened surveillance if there are siblings with ASD (10%-25% recurrence in families) or close relatives with developmental or psychiatric diagnoses
- eliciting and paying attention to parental concerns
- standardized developmental screening
- observing the child.¹⁰⁻¹³

These components are part of every checkup. Refer children who screen positive for atypical or delayed development for a more targeted evaluation. The prevalence of ASD is the same across all racial and ethnic groups, but non-Hispanic Black children tend to be evaluated later, highlighting the critical need for universal screening.³

The American Academy of Pediatrics (AAP) recommends every child have an ASD-specific screening at 18 and 24 months of age.¹⁴ The Modified Checklist for Autism in Toddlers, Revised, with Follow-up (M-CHAT-R/F) is a free validated tool useful in primary care for ASD screening

(www.mchatscreen.com). This parent report tool takes less than 5 minutes to administer and identifies children needing more extensive evaluation.

While the M-CHAT-R/F is intended to detect children at risk for ASD, a child who screens positive needs further evaluation, as the test is 94% predictable for some type of developmental disorder.¹⁵ The 24-month M-CHAT-R/F needs to be completed even if the 18-month M-CHAT-R/F is negative because 25% to 30% of children with ASD may initially appear typical but then lose skills and regress between 15 and 24 months.¹⁶ The tool has been translated to a number of languages.

It is tempting to jump to conclusions during a brief well-child visit; however, a single behavior is not enough to make a diagnosis as demonstrated in a 2015 study by Gabrielsen and colleagues.¹⁷ Psychologists with expertise in ASD analyzed 10-minute video samples of toddler behavior in two groups of children: "typical children" who acted as controls and a group who were positive during universal ASD screening.¹⁷ The goal was to identify children who needed referral for further evaluation.

Children with ASD engaged in unusual behavior only about 11% of the time. Most of the time (89%), these children demonstrated behaviors that were reassuring as typical (for instance, turning when called). All children in the ASD group responded to their name at least once, whereas 50% of controls did not react. Focusing on a limited range of typical behaviors, experts missed 39% of children who should have received a referral for ASD

evaluation. This study demonstrates the importance of formal screening tools in addition to clinical impressions and parental concerns for proper developmental evaluation.

Behavioral assessment. Well-child visits provide an excellent opportunity for behavioral assessment. Social smiles are expected by 2 to 3 months of age, and children should have consistent eye contact. A hallmark of an ASD diagnosis is a lack of spontaneous shared enjoyment; therefore, evaluation for "joint attention" (a child's desire to share an event or object with another) should begin by the 9-month visit.¹⁴ At this age, an engaged child with typical development performs a three-point gaze shift: looks at the parent, follows the parent's gaze to look at the object the parent looks at, and looks back at the parent with shared effect.

Joint attention can also be obtained by playing "peek-a-boo." The child responding when his or her name is called is expected at this visit as well.¹⁴ Parents of children with ASD often express concern about their child's hearing due to the lack of response by the child when his or her name is called (see *Well-child visit: Key observations*). Conversely,

these children seem overly sensitive to loud sounds, such as crying children or vacuum cleaners. It is the unusual responsiveness that is concerning.

Children with hearing loss do not respond to any sounds, while children with ASD preferentially ignore human sounds. Any child with hearing concerns or language deficits should be sent to an audiologist. Additionally, behavioral extremes (either constant crankiness or exceptional passivity) warrant further investigation.^{14,18} In typically developing children, separation anxiety emerges around 9 months, and crying when taken away from the parent is a reassuring sign that the child is developing attachments. Children with ASD may be inconsolable in a novel environment or with a change in routine, even in the presence of parents.

By the 12-month visit, a child should be able to “follow a point.” Have the parent exclaim, “Look” and point to an interesting picture or object; a typically developing child will look at the image, but a key component is that the child will respond next by looking back at the pointer and smile, indicating a joy in sharing this experience. A child with ASD will not have a mutual connection. By the 15-month visit, the typically developing child should gesture to “request” an object and attempt to engage the caregiver to obtain a desired item. As child gets older, pointing and speech add to this social connectedness.

At the 18-month visit, the child should interact with the provider. Be prepared to act unexpectedly: blow bubbles, wind up a toy, and watch the toddler’s reactions. Even most shy children should demonstrate delight. Imaginative play and pretending with objects (such as talking on phone and feeding the baby) are expected activities at 18 months of age. Hand the child a baby doll along with a bottle or spoon and suggest to “feed the baby.” If a male child will not perform, parents may justify their son’s lack of imaginative play and say, “he cannot do that because he does not play with dolls.” Educate parents that this is a developmental milestone regardless of gender. The child has experienced over 1,500 meals by this age and has observed the social behavior of eating.

Communication. Speech/language delay is frequently found in children with ASD, and this is often the parents’ first developmental concern. Careful speech/language evaluation identifies red flags pointing to developmental delay and a possible ASD diagnosis. Look for babbling by 12 months, one word other than “mama,” “dada,” or family names by 16 months, and two-word phrases, such as “help me” and “that’s mine” by age 2 years. Receptive competencies, such as understanding words (commands such as “give

Well-child visit: Key observations^{14,18}

Visit in months	Skill	How to elicit and assess
2	Social smile	Does the child smile back at the NP?
	Eye contact	Does the child look the NP in the eyes?
9	Joint attention	Play “peek-a-boo”
	Responds to name	Call name from across the room/behind the child
12	Follow point, look back at pointer with joy	Point to picture, exclaim “Look at that!”
15	Gesture to request	Observe interactions between the child and caregiver
18	Pretend play	Give child toys and say, “Feed or bathe baby”

me”) or picking up cues from the environment (putting on a coat means going to the park), provide a window into social capabilities, such as joint attention, social reciprocity, and sharing.¹⁷ Loss of acquired language and consistent echolalia (repeating words or phrases) are also red flags. Development is progressive, and regression at any age is concerning and a reason to consider an ASD diagnosis.

Although some young children have trouble with articulation, children with ASD have trouble with speech prosody: intonation (monotone), rhythm, rate, pitch,

Refer all children diagnosed with ASD for OT to assist with sensory differences and deficits in play and self-help skills.



volume, and quality of sound (nasal).^{19,20} As they get older, children with ASD may have difficulty having a conversation, defined as a four-point volley, with each person adding information, allowing the next person to continue.²⁰ Language of children with ASD often sounds scripted (using phrases from favorite cartoons) or restricted to their preferred interests (trains or dinosaurs). While families wait for an ASD-specific diagnostic evaluation, the NP should send a family for speech/language evaluations to begin treatment.

Sensory. Refer all children diagnosed with ASD to occupational therapy (OT) due to sensory differences as well

as deficits in play and self-help skills. These differences are either hypo- or hyperreaction to information received via the senses, including vestibular (sense of balance) or proprioceptive (body awareness in space).¹ To modulate sensory input, children with ASD have stereotypic, repetitive, or atypical behaviors, such as hand flapping, finger flicking, rocking, or twirling. These behaviors appear intense and serve a self-regulating or self-soothing purpose. OT helps determine the function of these behaviors, and strategies should then be developed to meet a child's sensory and adaptive needs.

Stereotypies can include self-injurious behaviors (SIBs), including head-banging or head-hitting, biting, and pinching. SIB is often a communicative act and requires a behav-

tight heel cords, or habit, but since some children with ASD toe walk, suspicion for ASD should be considered in a child with this behavior.

■ Management of the child with ASD

After a formal ASD diagnosis, NP responsibilities are just beginning. A child with ASD benefits from a multidisciplinary approach with the PCP managing this chronic disorder (see *Management considerations for children with ASD*). In collaboration with the family, the NP has a central role in promoting positive routines, creating a calm environment, and coordinating with specialists to improve quality of life.

Effective therapies. Applied behavioral analysis (ABA) by a BCBA has been demonstrated to be effective treatment

and helps improve long-term outcomes.^{23,24} Through reinforcement, new positive behaviors (such as learning to talk or acquiring social skills) are encouraged, and challenging behaviors are reduced. A child learns, "If I do this, then I get that." ABA training is intense;

the AAP recommends at least 25 hours each week, year round, with a one-to-one child-to-teacher ratio.⁸ NPs should know where to obtain high-quality ABA in the community because it is a cornerstone of ASD treatment.

Sleep. PCPs have a critical role in helping families of children with ASD manage sleep issues. Two-thirds of children with ASD have chronic insomnia.^{25,26} Insomnia is defined both as trouble falling asleep (taking more than 30 minutes) and/or prolonged night waking, which decreases optimal functioning during the day.²⁷ Research indicates that in children with ASD, sleep disturbances—particularly night waking—lead to daytime behavioral issues, such as aggression and hyperactivity.²⁸ Many children with ASD have additional comorbid high-arousal disorders, including attention-deficit hyperactivity disorder (ADHD) or anxiety, which make sleeping difficult.

The high prevalence of insomnia in ASD demands routine screening. Evaluate children with sleep issues to assure that there are no comorbid medical conditions, such as nighttime seizures, reflux, sleep disordered breathing, asthma, sinusitis, eczema, toothaches, or other pain that inhibits optimal sleep.²⁹ There are two types of behavioral insomnias: sleep-onset association, in which the child depends on very specific situations (people, objects, or settings) to fall asleep or stay asleep (when they wake in the night, those same interventions need to be repeated); the second is limiting setting difficulties created by caregivers who cannot stick to a consistent bedtime routine.^{25,26}

The Autism Treatment Network's (ATN) Sleep Committee created a Sleep Tool Kit that contains evidence-based



Sleep disturbances in children with ASD—particularly night waking—lead to daytime behavioral issues, such as aggression.

ioral therapist, such as a board-certified behavior analyst (BCBA) to evaluate the function of the behavior. SIB should be addressed early, as it may become dangerous, causing injuries such as detached retinas and cauliflower ears.

Motor. Children with ASD usually meet their gross motor developmental milestones on time. Children with ASD often have hypotonia, joint laxity, or poor coordination.^{21,22} Toe-walking can be a sign of cerebral palsy, a symptom of

Management considerations for children with ASD

Family support

- Screen siblings for ASD
- Safety check: fire, water, elopement, and identification
- Help parents find ways to enjoy spending time with the child (play, music, and the like)
- Affirm parents' hopes for future

Initiate evaluations and coordinate services

- Audiology evaluation
- Speech/language therapy
- Genetic testing
- Applied behavior analysis by a BCBA
- OT with sensory integration
- Physical therapy if coordination problems are present
- Special education/individualized education program
- Adaptive physical activity
- Mental health counseling for child and family
- Dental care by ASD-sensitive provider
- Transition planning by age 14 years

Table courtesy of the authors.

best practice strategies to improve sleep (see *Strategies to enhance sleep*). If these behavioral and environmental modifications are ineffective, regular or extended-release melatonin may improve sleep in some children with ASD. Use pharmaceutical grade melatonin and dose 30 minutes prior to the desired sleep time.²⁶ Some families find melatonin loses effectiveness in their child over time.

Feeding and nutrition. Many young typically developing children are picky eaters, but feeding that feels intense, problematic, and a constant struggle is a red flag for ASD. Sensitivity to textures, severe food selectivity, or overeating preferred foods make meal times disruptive for many families who have children with ASD. This and other factors lead nearly 30% of ASD children to be overweight compared with 24% of typically developing children.³⁰

For children who are underweight, it is essential to find nutritious ways to increase calorie density. Instant breakfast powder mixed with milk is palatable to many children. Adding cheese, increasing butter, or encouraging dips (such as ranch dressing for vegetables) increases calorie consumption as well. Children with severe food selectivity issues benefit from the interdisciplinary approach found in specialty feeding clinics. These clinics can often be found in regional children's hospitals or affiliated with developmental pediatric centers. It is not uncommon for children with ASD to have extreme pica (eating nonfood items), which leads to dental and gastrointestinal (GI) problems. If a child has significant pica, a lead level should be obtained.

GI disorders. Children with ASD frequently have GI disorders, including constipation, encopresis, diarrhea, and abdominal pain.³¹ A combination of low muscle tone and other motor problems, sensory processing difficulties, excessive self-stimulatory behaviors resulting in hyposensitivity to rectal sensations, and extreme food selectivity predispose children with ASD to constipation. Constipation and other GI symptoms can be manifested as behavioral changes in nonverbal children.³² Parents should monitor for regular bowel patterns and promptly treat constipation to decrease sleep disturbances, aggression, tantrums, or SIBs caused by discomfort.

As with typically developing children, most cases of constipation are functional; dietary and medication treatments that are suggested for typically developing children are effective for children with ASD.³³ Providers are referred to ATN guidelines and the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition's clinical practice guidelines for further suggestions.^{33,34} For older children with ASD who are not fully toilet trained, insurance will cover large size pull-up underwear with a prescription.

Strategies to enhance sleep^{26,46,47}

Set the stage

- Daily exercise early in day
- No caffeine
- Exposure to morning sunlight to help set the biological clock. Immediately upon awakening, expose the child to natural light: open curtains, walk dog, go to bus stop earlier; if possible, have natural light in the classroom.

Calming activities

- Qigong massage: 15 minutes to calm and balance energy flow
- Yoga (www.namastekid.com)
- Deep breathing stimulates the vagus nerve and slows the heart rate.
- Progressive muscle relaxation
- Rocking in rocker/swinging
- Warm bath (helps shed heat with the cool-down after the bath, and increases melatonin production)
- Weighted blanket or swaddle: increases proprioceptive input, calms nervous system (available online)

Environmental

- Sleep should be the only purpose of the bedroom.
- No TV in the bedroom
- Decrease the number of toys/objects in the bedroom.
- Lower house temperature by 4° F (2.2° C); create a cool, dark room, which increases melatonin production.
- Decrease blue light; no screen time before bed. Play board games or read a book instead.
- Quiet or white noise to drown out background noises
- Address sensory issues: textures/clothing too tight or loose
- Create a nest to feel safe, such as placing the bed against the wall.

Bedtime routines

- Consistent, short bedtime routine (never more than 1 hour; set appropriate time limits)
- Calming activities (see above) before bed
- Appropriate bedtime when "irresistibly sleepy"
- Routines: waking, eating, keeping the same bedtime every day
- Use a visual schedule for concrete cues and consistency.
- Teach the child to fall asleep alone.
- Bedtime pass: if the child wakes in the night or needs the parents, the bedtime pass is relinquished. If the child stays in his or her room all night and has pass in morning, agreed-upon reward is given.

Additional information: Sleep Education Toolkit at www.autismspeaks.org/atn.

Safety. Safety concerns for all children with ASD are significant but more problematic for children with poor communication skills and/or low IQ. These children should have their name and phone numbers in their clothes, on a necklace/bracelet, or on a temporary tattoo. Notify local authorities of their presence in the community. Instead of

a traditional alarm, smoke detectors that allow parents to record a message such as “Leave house and meet at tree” are useful. The Autism Speaks website provides a tool kit with a wide range of safety suggestions.

Exercise. Children with ASD tend to have weak core strength, hypotonicity, and decreased coordination.^{21,22} These conditions can be improved with carefully chosen

often impair children with ASD; physical therapy (PT) and prescriptions for shoe inserts are helpful.

Genetic testing. Approximately 10% of patients with ASD have an identifiable genetic syndrome.³⁵ The American College of Medical Genetics recommends genetic testing for all children with ASD.³⁵ Due to its diagnostic power, chromosomal microarray, which identifies abnormalities in chromosomes, is the first line for testing.³⁶ Evaluation for fragile X, the most common known single gene cause of ASD, is recommended for males and select females with ASD. Yield for genetic syndromes is higher in patients with ASD who also have additional findings, and



Adjunctive therapies are effective to treat comorbid conditions, such as anxiety, ADHD, depression, or insomnia.

physical activities. Individual sports are a better choice than team sports due to the child’s poor social skills. Children with ASD often enjoy running or repetitive activities; poor coordination makes throwing sports difficult. The calming effect of water makes supervised swimming an excellent activity. Horseback riding increases core strength and may be reimbursed with a prescription. Flat feet and weak ankles

these individuals benefit most from testing (see *Probability of genetic disorder*).³⁵

While it will not change the ASD diagnosis, testing can improve care for children with genetic disorders that cause heart, kidney, neurologic, or immunologic problems. Testing is expensive, but many insurance companies will pay with a prior authorization. Medical procedures, including phlebotomy, are challenging in children with ASD. Souders and colleagues discuss strategies to increase success (for example, making sure the family brings tokens for the reward system proven to be successful at home).³⁷

Comorbid conditions. Autism is truly a spectrum with broad heterogeneity of features among children with this condition. Comorbid conditions may initially overshadow the ASD diagnosis. Depending on how testing is performed, 32% to 60% of children with ASD will be categorized as having cognitive impairment as well.^{3,38} At least one-third of children with ASD are estimated to have ADHD as a comorbid condition.³⁹ The repetitive behaviors of ASD may cause a child to be mistakenly diagnosed with obsessive compulsive disorder. For children with ASD, particularly those with lower cognitive abilities, 26% develop seizures in adolescence.⁴⁰ Since 40% of children with ASD have anxiety, screening all children with ASD (when possible) using the Screen for Childhood Anxiety Related Emotional Disorders scale or another anxiety scale is standard of care.^{41,42}

No medications treat the core deficits of ASD. Adjunctive therapies are effective to treat comorbid conditions, such as anxiety, ADHD, depression, or insomnia. Risperidone and aripiprazole are FDA approved for ASD to treat irritability.⁴³ Methylphenidate is FDA approved for ADHD with children, and the AAP clinical practice guideline specifies its effectiveness for children with ADHD who have autism.⁴⁴

Transition services. IQ level and diagnostic severity are the best predictors of future function.⁴⁵ By age 14 years, families must begin planning for young adulthood and

Probability of genetic disorder³⁵

ASD plus any one of the listed items increases the probability of a genetic disorder.

- Microcephaly
- Seizures
- Congenital anomalies
- Dysmorphic features
- Family history of developmental disabilities
- Family history of psychiatric problems

Resources

Autism Speaks/ATN tool kit:
www.autismspeaks.org.

AAP: Autism: Caring for Children with Autism Spectrum Disorders: A Resource Toolkit for clinicians (digital download: \$94.95):
<https://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/pages/Caring-for-Children-with-Autism-Spectrum-Disorders-A-Resource-Toolkit-for-Clinicians.aspx>.

CDC Autism spectrum disorder resources:
www.cdc.gov/ncbddd/autism.

AAP: Is your toddler communicating with you?
AAP.org \$25/50 pamphlets.

Autism Speaks: ATN/AIR-P Blood draw tool kit:
<https://www.autismspeaks.org/science/resources-programs/autism-treatment-network/tools-you-can-use/blood-draw-toolkits>.

beyond. The Autism Speaks Transition Tool Kit is a useful resource to help families get organized and prepare for the future needs of the child (www.autismspeaks.org/family-services/tool-kits/transition-tool-kit/getting-organized).

■ Case revisited

Returning to the case: Does JT have typical development? Do not be quick to dismiss his case as “just toddler behavior.” He warrants the careful investigation that is done for every 2-year-old: further questioning of developmental and family history (Was he premature? Do his siblings have ASD? How old are his parents?); eliciting parental concerns (Does he have sensory food issues? Is he constantly cranky, or only fearful in the office? How much sleep does he get? Is he chronically constipated?). Administer the M-CHAT-R/F as would be done for every 24-month-old. The NP should engage him, possibly in the waiting room where he will not feel so threatened because a comprehensive observation of his interactions and behaviors is necessary. Results of this investigation will determine the plan. This child should not be discharged as a “typical toddler” without further exploration. Obtaining a definitive diagnosis is not necessary prior to beginning speech, OT, or PT interventions in a child with developmental delays.

NPs should know their community resources for referral. (See *Resources*.) Autism evaluations are done by many different specialists depending on their expertise and interest: developmental pediatricians, NPs in specialty practice (neurology/developmental pediatrics), psychiatric NPs, psychologists, or psychiatrists, can all provide a diagnosis. 

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