Recognition of Autism Before Age 2 Years

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Objectives

After completing this article, readers should be able to:

1. Characterize the deficits of a child who has autism.
2. Describe the role of observing linguistic and social development in children who have autism.
3. Implement a strategy for surveillance and screening of young children.
4. Discuss the management of autism, including the use of medication.

Introduction

Primary care physicians (PCPs) caring for 1,000 children in a general practice should expect approximately six of their patients to meet the diagnosis of an autism spectrum disorder (ASD). (1) The impact of this prevalence is illustrated by a 2004 survey of PCPs revealing that 44% cared for 10 or more patients who had an ASD. (2) The ASDs include: autistic disorder (AD), Asperger disorder, and pervasive developmental disorder—not otherwise specified (PDD-NOS), a threshold term used when a child meets some but not all criteria necessary for a diagnosis of either AD or Asperger disorder.

Research has documented the positive benefit of developmental and behavioral intervention for children who have ASDs, particularly when it is initiated prior to 3 years of age. (3) Early recognition of ASDs also is important for timely genetic counseling because younger siblings of children diagnosed as having ASDs have a 10 times increased risk of also having an ASD. The purpose of this review is to help the clinician recognize signs of ASDs that occur before age 2 years. Because the signs of Asperger disorder usually appear later, they are not discussed. The term "autism" is used in this review to represent both AD and its milder form, PDD-NOS.

Currently, autism is diagnosed infrequently in children younger than 3 years of age. The diagnostic process is prompted most often by parental concern about absent or delayed speech. However, signs of autism, especially deficits in social skills and in preverbal gestural language, are present in most children by 18 months of age. Such signs are subtle and may not be noticed by parents. New information regarding very early signs of autism is emerging through prospective studies of high-risk infant siblings. (4) Using these early "red flags," one study reported that approximately 50% of affected children could be diagnosed reliably by 14 months of age. (5) To facilitate earlier recognition through surveillance and screening strategies, the American Academy of Pediatrics (AAP) has published two guidelines: "Identifying Infants and Young Children with Developmental Disorders in the Medical Home: An Algorithm for Developmental Surveillance and Screening" (6) and "Identification and Evaluation of Children with ASDs." (7) In an effort to lower the average age of diagnosis and promote earlier access to intervention services, these guidelines recommend ongoing surveillance for autism (and general development) at every health supervision visit in all children and "heightened surveillance" in high-risk younger siblings. An increased awareness of early signs of autism can facilitate surveillance and enable PCPs to play a key role in early recognition that, when coupled with earlier access to appropriate interventions, could improve outcomes markedly.

The Importance of Surveillance

Diagnosing ASD is very difficult because there are no pathognomonic clinical signs or confirming laboratory tests. Over the past 3 decades, the DSM (American Psychiatric

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Association *Diagnostic and Statistical Manual of Mental Disorders*) criteria have served as the gold standard for making a clinical diagnosis; newer standardized tools that operationalize the DSM-IV criteria (eg, Autism Diagnostic Observation Schedule [ADOS] (8)) optimize diagnosis. However, some of the DSM-IV criteria address developmental skills that do not apply to children younger than 2 years of age, and standardized tools have not yet been validated in this age group. For example, "failure to form age-appropriate peer relationships" is not applicable to children younger than 2 years of age. Additionally, it is impossible to apply criteria relating to abnormal conversational skills and stereotypic language in a young, preverbal child. Often, ritualistic behaviors and a need for routines appear in children who have autism after the third birthday. Thus, even children who have severe AD may not meet full criteria at very young ages, making diagnosis even more challenging.

Realizing this diagnostic dilemma, especially in the face of widespread media promotion of early diagnosis and intervention, Stone and associates (9) have suggested applying only the following four of the possible DSM-IV criteria to children younger than 3 years of age:

- Lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (eg, by lack of showing, bringing, or pointing out objects of interest)
- Lack of social and emotional reciprocity
- Marked impairment in the use of multiple nonverbal behaviors, such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
- Delay in or total lack of the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)

Three of these criteria address social skills, illustrating their relative importance in defining autism in very young children. These researchers propose that all four criteria be present to make the "provisional diagnosis" of autism in the very young child; re-evaluation using the full DSM-IV-TR (10) criteria, combined with a standardized tool, is recommended after the third birthday. Because referring young children for diagnosis depends on the PCP's ability to recognize behaviors illustrating these criteria, the following discussion targets early social and prelanguage deficits. Currently, no standardized autism-specific screening tests for children younger than 18 months of age are available, although some are in development. Thus, surveillance, coupled with a high degree of suspicion, is very important and should lead to earlier diagnosis and intervention.

"Clinical probes" are provided in most sections of this article to assist the PCP during the surveillance process, but they should not be used as a screening test. Rather, they are only one part of the total package of developmental screening. The PCP may choose to use one or several of these procedures at each visit to monitor for autism. In addition, an autism screening tool (6)(7)(11) should be used at the 18- and 24-month visits and at any time there is concern about ASD.

Early Social Skill Deficits

Social development usually parallels overall general functioning (motor+adaptive+language+cognitive domains) in children developing typically. Social development also parallels general functioning in children who have global developmental delays (GDD). For children who have autism (with or without coexisting GDD), the development of social skills and language is more delayed and characteristically is "out of sync" with motor, adaptive, and cognitive functioning. The discrepancy between the development of social skills and general development is one of the most important defining criteria. Discrepancies may be recognized late in the first year after birth, but they become more obvious by 18 to 24 months of age in children later diagnosed with autism.

The delay in social development often is manifested by a lack of or decreased need for social and emotional reciprocity or "social relatedness." Such children have little or no inherent drive to connect with others and share feelings, thoughts, and actions. Infants who have other disabilities may have difficulties connecting with others due to their respective motor, hearing, and visual deficits, but the inherent drive to connect promotes compensatory strategies. For example, infants who have cerebral palsy establish social relatedness through eye contact, facial expressions, sounds, and conversations. Infants who have hearing loss often are visually hypervigilant and use eye contact and gestures to connect with others. Children who have autism make few, if any, attempts to compensate and relate socially to others; they often are content being alone and usually ignore other's bids for attention, affection, or connectedness. Children who have autism may seek comfort and affection when injured or anxious, but such self-initiated bids solely when in need should not be interpreted as sufficient indicators for "normal" social relatedness. Children who have autism can and frequently do form attachments with family members; sometimes these may become unusually strong and problematic. Deficits in social relatedness are manifested by several behaviors that can be observed spontaneously or prompted during an encounter.

Joint Attention

The single most distinguishing characteristic of very young children who have autism is a deficit in "joint attention" (JA) that is discrepant from overall functioning. JA is a normal, spontaneously developing behavior that demonstrates the child's joy in sharing an object or event with another person. It is triadic; the child alternates attention between an object/event and another person by looking back and forth between the two and "connecting." It is a core feature of the DSM-IV-TR criteria and a critical component of current autism screening and evaluation tools.

JA may be classified as: 1) "spontaneous" when the child looks back and forth between an interesting object/event and the caregiver to share interest, 2) "responding" when the child looks in the direction indicated by the pointing of a caregiver and connects, and 3) "initiating" when the child points to an object/event to establish a connection with another person. As with all developmental skills, JA appears to develop in graduated stages. At 8 to 10 months of age, the typically developing infant, while socially engaged with his or her mother, will follow the mother's gaze if she shifts her eyes away from the infant to look at an object or event across the room. This action leads to sharing in a mutual experience. Children who have autism often take no notice of the shift in gaze. At about 10 to 12 months of age, a child can "follow a point." For example, if a parent sees something of interest, points in its direction, and says, "Oh look!", the typically developing child looks in the direction that the parent is pointing. On seeing the object of interest, the child looks back at the parent and may smile (or frown if the object is frightening). If the object is unfamiliar or the infant does not see the object, he or she may look back quizzically at the parent. True JA requires the child not only to follow the point, but to look back at the caregiver and "connect" socially. A child who has autism often is oblivious to the parent's bid or may look briefly at the object with little interest but does not look back at the parent to connect.

Clinical Probe for the 12- and 15-month Health Supervision Visits: *The clinician might demonstrate the child's ability to follow a point by saying, "Look! See the. . . ." and point to an interesting object or picture on the wall or ceiling. If there is no response, call louder and initiate the bid with the child's name or a tap on his or her shoulder. Often, no degree of intensity is successful in getting the child who has autism to look.*

Later emerging JA skills are characterized by the child rather than the adult initiating the connection. Pointing can serve both communicative and social functions for children. As a communicative function, children point to "request" an item that is out of reach. As a social function, children point to comment or to draw another's attention to an event or object (eg, the moon) out of mere interest and simply to "share." The "requesting" point normally appears first, at approximately 12 to 14 months. Words later may accompany the pointing as language develops. While pointing to the desired object (eg, a cookie), a child looks back and forth between the object and the caregiver to be reassured that the caregiver understands his or her need. The gesture is triadic (child, object, caregiver), but it is not purely social because the cookie is the goal and the caregiver is simply a means by which the child can obtain it. Most children who have autism do not master this skill at age-expected times; some, especially younger children, may make no attempt to elicit help and just cry. Others may lead their parents by the hand to the desired object and simply wait or cry. Some may engage in primitive pointing involving an outstretched arm in the direction of the desired object and opening and closing the hand in a repetitive grasping motion. However, there is no eye contact or connection with the parent, so this gesture does not represent true JA.

At 14 to 16 months of age, typically developing children point to "comment" or call attention to and share an interesting object/event. The same triad exists (child, object, caregiver), but the goal is reversed. The object now is the means by which the child obtains his or her goal, the adult's attention. The child looks alternatively between the object and the caregiver for reassurance that the object has been observed and appreciated. Again, depending on the child's language level, he or she may use words when pointing. Such pointing serves a purely social purpose and represents fully developed JA. Absence of this type of pointing in the 18- to 24-month-old child is a strong indicator of autism. Such JA pointing should be distinguished from pointing simply to label objects with little eye contact or sharing of affect. Although children who have autism can be adept, even advanced, at self-initiated, ritualistic labeling, they often are unable to point to the same objects on request, and the labels rarely are used in social conversation. Around 14 to 16 months of age, typically developing children also begin to "show" or bring an object to the parent, as if to say, "Look at this!" This JA act is qualitatively

different from bringing an object to the parent to request help, such as to open a container, but even this latter type of request is infrequent in autism.

Lack of or significantly delayed JA skills appear to be specific to autism. Children who have GDD without coexisting autism develop JA skills at rates similar to those of the development of other skills and begin pointing to comment when they attain a mental age of 14 to 16 months. Thus, it is necessary to know the child's overall level of functioning as well as his or her progress in JA development to help distinguish autism from GDD. If there is a discrepancy between the two domains in which JA skills are significantly more delayed than general skills, autism is likely. If the delays in all domains are similar, GDD is more likely. JA appears to be a pivotal skill; that is, functional language is dependent on its mastery. Studies have shown that once JA is mastered, functional language begins to emerge within 1 year. (3) This insight has stimulated the development of curricula that have resulted in successful promotion of both JA and language development. This accomplishment underscores the urgency of an early diagnosis and prompt access to intervention.

Social Orienting

Social orienting is the ability to orient to verbal stimuli, in particular, turning to respond to one's own name being called. At about 8 to 10 months of age, most typically developing children consistently turn their heads toward the speaker when their names are called. Children who have autism usually do not, prompting parents to call louder or tap their child's shoulder in an effort to get a response. Parents may begin to have concerns that their child cannot hear. With greater vigilance, they realize that the child seems to hear well in other situations. This dichotomy occurs because children who have autism often attend to environmental sounds extremely well but ignore human voices. Retrospective evaluations of birthday videos of 1-year-old children later diagnosed as having autism have demonstrated that "blinded" viewers could recognize autism with accuracy at this age. (12)(13) The best distinguishing factor was failure to orient to name because JA pointing skills may not be universally present in typically developing children at 1 year of age. Newer prospective studies of high-risk infant siblings later diagnosed with autism have confirmed this finding. (14)

Clinical Probe for the 12-month Health Supervision Visit: *The PCP can evaluate the child's ability to orient socially by calling out his or her name and noting whether the child turns and connects. If he or she does not, the PCP should try again. Typically developing children usually respond by the second trial.*

Pretend (Symbolic) Play

Lack of or delayed symbolic play (using pretend actions with objects) appears to be a relatively reliable distinguishing feature of autism. Play usually evolves in a highly predictable manner in typically developing children. Once children can grasp and retain objects (approximately 4 months of age), sensory-motor play begins. They mouth and manipulate objects (eg, blocks). Children who are 8 to 10 months old may throw the blocks, bang them on the table, or bang them together. Oral-motor play evolves into a more functional type of play as children become aware of the intended use of objects. At 12 to 14 months of age, using this new understanding as well as evolving imitation skills, children begin to build towers with the blocks. Pretend play subsequently emerges and, with time, increases in complexity and imagination. "Simple pretend play" usually begins at approximately 16 to 18 months of age, when children begin to use miniature representative items, such as a tiny plastic bottle or spoon to feed a doll or a toy telephone to "talk" with parents. At 18 to 20 months of age, children begin to engage in "complex pretend play." They use generic items (eg, blocks) rather than miniatures to represent food and pretend to feed the doll or pretend that a banana or stick is a phone. Pretend play also becomes more complex as children engage in several successive pretend scenarios (feeding, changing clothes, and putting the doll or teddy bear to sleep).

Children who have severe autism and coexisting GDD often remain in the sensory-motor stage throughout their lifespans; they continue to mouth, throw, twirl, and bang objects stereotypically. Some progress to functional play, but true pretend play is rare before the age of 2 years. Furthermore, they often have little interest in toys, frequently preferring everyday items such as string, sticks, rocks, dirt, chains, strings of beads, and books or videos (simply for carrying, not for reading or viewing). When they are interested in typical toys, they may play with them in unusual ways. For example, rather than typical play with a miniature truck, a child who has autism might turn it upside down and spin the wheels repeatedly. Some children, particularly those who have normal

nonverbal intelligence, may be proficient in constructive play (eg, using objects in combination to create a product, such as stacking blocks, nesting cups, putting puzzles together) and in computer games because mastery of such play depends on independent trial and error problem-solving rather than on observation and imitation of others. Children who have autism excel in tasks learned through trial and error. Occasionally, parents may state that their child is unusually good, that is, he or she is content to play alone for hours, requiring little attention or intervention from the parent. Such solitary play usually is sensory-motor and ritualistic or constructive. Many children who have autism appear to enjoy "roughhouse play" (being tickled or tossed in the air) and show fleeting moments of social relatedness.

Although additional social deficits are associated with ASDs, they often are not recognized until later, especially in children who have Asperger disorder. Other deficits are more difficult to assess in a clinical setting and impractical for use in surveillance.

Clinical Probes for the 12- and 18-month Health Supervision Visits: *The PCP should ask the parents about the child's favorite toys and manner of play. The PCP also might ask the parents to describe a "typical day" to ascertain the amount of time spent in solitary play. If parents report that their child enjoys playing with other children, probe further. If play consists solely of chasing and roughhousing, the enjoyment simply may reflect sensory-motor pleasure rather than social engagement.*

Early Language Skills Deficits

Historically, delays and deviancies in language development have been the most common presenting signs in children later diagnosed with autism. However, parents usually did not raise such concerns until after their child's second birthday, making such concerns less helpful in reaching an early diagnosis. Because autism has been spotlighted frequently in the lay media, more parents are aware of the condition and may ask about it regardless of the nature of their child's symptoms. To reassure the parents or to suggest appropriate referrals, the PCP should be familiar with the more subtle and earlier appearing prelinguistic language deficits associated with autism. Parents may not be able to verbalize such subtle deficits, requiring the PCP to probe with specific questions. Parents may be unable to answer some questions decisively at the time of questioning, but once the subtle characteristics are brought to their attention, they often become very vigilant. In such cases, the PCP should encourage the parents to report back once they have had the opportunity to observe their child systematically.

Prelinguistic Language Abnormalities

Prelinguistic abnormalities characterizing autism during the first postnatal year became apparent when investigators began retrospective evaluation of infant videos of children later diagnosed with autism. (12) Recent prospective studies of high-risk infant siblings have confirmed video findings and revealed new "red flags." (4) Some infants later diagnosed with autism are unusually quiet and make few vocalizations. Other infants make atypical vocalizations; instead of cooing and babbling, they may hum or grunt for extended periods, squeal stereotypically, or laugh inappropriately. Others may be very irritable, cry for long periods of time, and be difficult to console. Infants later diagnosed with autism demonstrate fewer gestures. They may not wave "bye," extend their arms to be picked up, or engage in interactive hand movements associated with children's songs and nursery rhymes.

Normal babbling usually begins by 6 months of age, with repetition of the same phoneme ("ba ba") in a monotone voice. Later, the babbling advances in complexity, with utterances becoming longer and including several phonemes ("ba da pa pa da da"). Typically developing infants develop a to-and-fro pattern of babbling and silence (or apparent listening) that is coordinated with the caregiver's speech and is similar to true turn-taking conversation seen in older children. For example, many infants babble to themselves playfully on awakening. When the mother approaches the bedside to say "good morning," the infant, on seeing the mother, smiles and stops babbling at the sound of her voice. When the mother turns away to get diaper supplies and pauses in her speech, the infant may resume babbling but stop again when mother speaks. High-risk siblings who are later diagnosed with autism often continue to vocalize as if they are not aware of their mother's speech; thus, vocalizations overlap and do not resemble the "proto-conversations" described previously. Often, there is less eye contact, and the mother may feel that her child does not recognize her voice or notice when she enters or leaves the room. At 10 to 12 months of age, the typical infant adds inflection to utterances in an attempt to tell a story (jargoning), causing his or her speech to sound like animated gibberish. Children who have autism often do not add inflection at this age.

Other prelinguistic language deficits overlap with social skills. For example, not responding to one's name being called (previously discussed in the section on social orienting) traditionally has been considered a receptive language deficit, but recent studies of infants later diagnosed with autism have revealed that the deficit is linked more closely to the infant's lack of social relatedness than to a deficit in receptive language or auditory comprehension. (14) The possibility of a hearing loss also must be considered, but as noted previously, children who have hearing impairments fail to orient to *both* social and environmental sounds.

Clinical Probes for the 9- and 12-month Health Supervision Visits: *Does your baby . . .*

- *Look at you when you are speaking?*
- *Babble? If yes, also ask . . . Does he or she take turns vocalizing back and forth with you? Are vocalizations becoming more varied in terms of types of sounds and length of sequences? Are these associated with a growing repertoire of gestures?*
- *Wave "bye bye"? Raise his or her arms to be lifted?*
- *Hear your voice as well as he or she hears environmental sounds?*
- *Make any unusual vocalizations or laugh inappropriately?*

Absent or Delayed Speech

Absent or delayed speech has been the most common presenting sign in children who have autism. Although most parents sense something is wrong by the time their child is 18 months of age, they often do not share such concerns with the clinician until much later, when they realize that the child has little or no speech. Sometimes they may rationalize that the delays are due to the child's temperament (shy) or to a lack of opportunity to interact with peers if the child is an only child. Although these and other environmental factors may cause delayed speech, receptive language is normal. Because parents sometimes overestimate their child's receptive language abilities and think that their child "understands everything," referrals to audiology to evaluate hearing status and to a speech and language pathologist to evaluate both expressive and receptive language are indicated. If hearing and receptive language are normal, a "wait and see" approach may be appropriate. However, the PCP should recommend strategies (by means of developmental handouts, attendance at a preschool with verbal peers, etc) to stimulate speech.

If hearing is normal but receptive language is delayed, the PCP must consider the possibility of autism or GDD; a "wait and see" approach is not appropriate. In such a case, the PCP should refer the child immediately to a developmental or autism specialist (or a team of specialists) and to an early intervention program. (7) The clinician should not delay; a definitive diagnosis is not necessary to begin intervention. Staff can begin an intervention program that is tailored to the child's deficits, and if necessary, the strategies can be revised after a diagnosis becomes evident. Unfortunately, parents of children who have autism sometimes complain that they received inappropriate reassurance regarding their concerns, the clinician counseled a "wait and see" approach, and referrals were delayed.

Clinical Probes for the 12- and 15-month Health Supervision Visits: *The AAP has recommended formal screening for developmental skills at 9, 18, and 24 or 30 months of age using standardized tools that include social and language milestones. These tools also can provide useful probes for the 12- and 15-month visits. Developmental tools are described in both the AAP Developmental Surveillance and Screening (6) and ASD (7) guidelines. Selected tools are included in the AAP ASD Toolkit (11).*

Language Regression

Approximately 25% to 30% of children later diagnosed with autism seem to develop normally and then regress. Parents may report that children stopped talking and gesturing (eg, pointing, waving bye) and made less eye contact. Such "autism regression" usually occurs between 15 and 24 months of age, with most occurring between 18 and 21 months. Although children appear to develop normally until the regression, recent prospective studies have revealed that some children have subtle social and language deficits prior to regression. (13) (15) Loss of language skills is not pathognomonic for autism; it occurs in Rett syndrome and other neurodegenerative disorders. Loss of speech associated with seizures is characteristic of Landau-Kleffner syndrome, but regression is later (usually older than 3 years) and not associated with loss of eye contact or social skills. Sleep electroencephalographic tracings in affected children often are characteristic.

Clinical Probe for the 15-, 18,- and 24-month Health Supervision Visits: *Although most parents are keenly aware when regression occurs and are likely to raise a concern, others may rationalize that some event in the child's life (birth of a sibling, move to a new house, death of a family member) caused the loss of skills. Because they may not raise a concern spontaneously, a probe might be needed to highlight the importance of skill loss.*

Atypical Language

In addition to delays, children who have autism may demonstrate atypical language during their second year. Such atypicalities sometimes can give the false impression of "advanced speech," but the speech is echolalic, ritualistic, and not functional. Most typically developing children pass through a stage where they imitate or echo another's speech (echolalia). They repeat the last one or two words of the sentence they have just heard. Echolalia usually is a temporary phenomenon and occurs when toddlers are rapidly gaining new words. Echolalia is classified as immediate (parroting that occurs immediately after the partner's vocalization) or delayed (parroting that occurs at a later time). Whereas typically developing children demonstrate the immediate variety, children who have autism demonstrate both types. Autistic echolalia also is much more pervasive and enduring and includes larger "chunks" of verbal material. Children who have autism may demonstrate exceptional verbal memory linked to echolalia and recite nursery rhymes, advertisement jingles, or the ABCs at much younger ages. The complexity of such speech utterances can mask true deficits in both functional speech and receptive language skills. For example, a child may be able to recite the ABCs or sing a television advertisement jingle but be unable to ask the parents for a drink or follow a command. Unless this discrepancy between echolalic and functional language is recognized, a speech deficit may be overlooked.

Some children who have autism, especially those who are of normal intelligence, may become obsessed with labeling colors, shapes, and numbers, yet they are unable to use the terms in conversation or point to them on request. Unlike typically developing toddlers, they demonstrate less interest in common everyday objects or pictures in books and rarely point to them to request new words. They may use overlearned gestalt pseudophrases or sentences spoken as a single "giant word" (eg, Whatisit? Idontknow.), yet they lack the ability to combine words in novel phrases that convey meaning. Some children who have autism say "pop-up" words that seem to be out of context and have no communicative intent to the listener. As suddenly as these words pop up for no apparent reason, they also disappear. Occasionally, the parent might discover the stimulus for the verbalizations. For example, on seeing his mother dressed in a pink striped blouse, a toddler who has little if any functional speech exclaims "elephant." No elephants (real or stuffed) or pictures of elephants are within sight. He repeats this several times that day but not the next. The next time mother wears the same pink blouse, he again begins saying "elephant" repeatedly. The mother may recall that she wore the blouse the day they went to the zoo and he saw his first elephant.

Clinical Probes for the 15,- 18,- and 24-month Health Supervision Visits: *The clinician may ask the parent about echolalia, pop-up words, and exceptional labeling skills. Formal screening is recommended at the 18- and 24-month visits.*

Restrictive Interests, Stereotypies, and Repetitive Behaviors

Although many typically developing children form attachments to a stuffed animal, a special pillow, or a "blankee," children who have autism often prefer hard items (ballpoint pens, flashlights, a piece of rope, or particular action character). Moreover, the attachment is much more robust; they may insist on holding the object most of the day, even during meals. Many children develop stereotypies (hand flapping, twirling, finger movements, rocking, head nodding, toe walking, licking, sniffing). Although stereotypies are distinctive and obvious, they usually appear later than 2 years of age. They are not specific for autism; children who have severe GDD or visual impairment also may demonstrate stereotypies. Even typically developing children can flap their hands briefly when excited. Some children engage in repetitive behaviors (eg, lining objects up) and may protest violently when they are directed to a new activity. The anger and protest may escalate quickly to a prolonged temper tantrum and aggression or self-injurious behaviors.

Clinical Probes for 9,- 12,- and 15-month Health Supervision Visits: *Note gait and whether the child carries a comfort item into the office. Probe for unusual attachments, stereotypic movements, self-injurious behaviors, and unusually severe temper tantrums with transitions or for no apparent reason.*

Additional Early Autism Characteristics Not Included in the DSM-IV-TR Criteria

Children who have autism may demonstrate simultaneous hyposensitivities and hypersensitivities for different stimuli within the same sensory modality. For example, the slightest sound of water dripping may irritate the child, yet the child may seem oblivious to his mother calling his name loudly. Affected children may explore toys visually in unusual ways, holding objects very close to their eyes, looking at objects out of the corners of the eyes, or demonstrating an unusual head tilt. Others may have oral aversions and intolerance to certain food textures that contribute to self-imposed restricted diets. Additionally, children may demonstrate "tactile defensiveness" and not tolerate soft touch, garment labels, or certain textures. On the other hand, they may be indifferent to noxious stimuli and injuries that typically are painful to others. The dichotomy is puzzling, but it is believed to be due to an abnormal arousal level or sensory gating system.

The motor skills of children who have autism may seem advanced, especially running, climbing, and jumping skills. However, some children may have subtle deficits in fine motor skills, coordination, and motor planning and sequencing of movements. Most affected children exhibit fewer imitative motor patterns, although this may be linked more closely to social rather than pure motor deficits. In addition to abnormal quality of motor actions, children who have autism may demonstrate abnormal amounts of activity. Some may appear to be hyperactive and motor driven, with an exterior focus of attention. Others may be hypoactive or withdrawn, move little, and seem to have an interior focus of attention.

Clinical Probes at the 12,- 15,- 18,- and 24-month Health Supervision Visits: *The clinician should ask the parents about diet, unusual visual behaviors, hypo- or hypersensitivities, and activity level. A neurologic examination or gross and fine motor screening tools can be helpful in evaluating coordination and motor skills.*

Autism is not associated with a classic physical phenotype. More than 90% of children who have ASDs have "idiopathic" ASD, meaning that the cause currently is unknown and that there is no known associated syndrome. Such children look normal and have few, if any, dysmorphic features (eg, posteriorly rotated ears). Some of these children demonstrate accelerated head growth beginning at about 6 months of age and continuing for the next few months, sometimes to the point of macrocephaly. However, the head size often normalizes in late childhood. The remaining 5% to 10% have an associated syndrome (eg, fragile X syndrome, tuberous sclerosis, phenylketonuria) and manifest the physical signs that are characteristic of that syndrome. (16)

Clinical Strategy for All Health Supervision Visits: *Generally, most dysmorphic features are recognized at birth; some may be subtle and require ongoing surveillance as they become more prominent. The PCP should measure the head circumference and plot head growth to monitor for growth acceleration. Although the PCP should consider the possibility of hydrocephalus, computed tomography scan or magnetic resonance imaging often is not indicated unless there are associated neurologic signs or other indicators for neuroimaging studies.*

Screening and Evaluation

In addition to conducting surveillance at every health supervision visit, the AAP has recommended formal screening of all children with a standardized ASD-targeted tool at the 18- and 24-month visits and whenever a concern is raised. (7) To operationalize this recommendation, the AAP has developed an ASD toolkit. (11) Although the toolkit contains several screening tools, no screening tool is ideal, and most target children 18 months and older. Screening tools for younger children are in development and should be available soon. The AAP tool kit also can serve as a resource guide for both clinicians and parents because it contains related policy statements, clinical reports, surveillance and screening algorithms and tools, and two collections of fact sheets (one set for physicians and one for parents) that address major health, developmental, educational, transition, and family support issues associated with ASDs.

The ASDs surveillance and screening algorithm published in the AAP ASD clinical report (7) states that if a child has two or more risk factors (eg, older sibling having an ASD, parent concern, other caregiver concern,

or physician concern), the PCP should make three simultaneous referrals: to an early intervention or school program (depending on the child's age), to an ASD specialist or team of specialists for a comprehensive evaluation, and to audiology (if this has not already been done). If the child has only one risk factor, the PCP should use an appropriate screening tool based on the age of the child. If the result is positive, the PCP should make the same three referrals. If the results of the screening are negative, an additional appointment should be made within 1 month to monitor progress and address any residual concerns the parent or PCP might have in spite of a negative screen. If there are no risk factors, the child should be screened with an ASD-specific tool at 18 and 24 months. This process is described in much greater detail in the AAP ASD clinical report. (7)

Whereas surveillance and screening are the responsibility of the PCP, the comprehensive evaluation may be accomplished best by a specialist or team of specialists who have expertise in autism. Such evaluations usually include the following components: 1) a thorough history, including a three-generation pedigree; 2) a detailed physical examination focusing on neurologic signs and dysmorphic features; 3) thorough developmental or psychometric evaluations, depending on the child's mental age and level of cooperation; 4) assessment with a standardized ASD tool (eg, ADOS) that operationalizes the DSM-IV-TR criteria, or if not available, assessment of the criteria themselves using clinical judgment; 5) an assessment of the family's strengths and needs; and 6) an etiologic search based on results of the first five components. The PCP may be involved in this process to varying degrees, depending on the available local resources and his or her level of comfort.

Management

The PCP's most important role is to provide a medical home, as with all children (www.medicalhomeinfo.org). This can be more challenging for several reasons: 1) Many children who have ASD are unable to describe their symptoms or localize their pain; 2) Care coordination can be complex due to a variety of practitioners (developmental and behavioral specialists, pediatric subspecialists, therapists, teachers, social workers, vocational staff); 3) Parents may be stressed and require more family supports; 4) Children may have ongoing pica and continue to need lead screening; 5) Children may have coexisting medical (seizures, nutritional), psychiatric (anxiety, obsessive compulsive disorder), and behavioral (aggression, sleep disorders) problems; and 6) Infant siblings need heightened surveillance. The PCP also may need to counsel parents regarding the 10 times increased risk of ASDs in subsequent siblings if this information was not conveyed by the specialist making the diagnosis.

Management of autism generally is the province of early intervention specialists, educators, therapists, and behavior management specialists. Early intervention programs and schools should provide services that are individualized, appropriate, and intense and should address development, social skills, behavior, and academic issues.

Although all children should receive appropriate developmental, therapeutic, and educational services, not all require medication. There is no medical "cure" for autism, and only one medication (risperidone) has been approved by the United States Food and Drug Administration for use in children who have ASDs. Medication is an adjunctive treatment that often is used to address challenging behaviors after behavioral and environmental interventions have failed. However, medications never should be started without first performing a physical examination to rule out a medical cause (eg, tooth abscess, ear infection, abdominal discomfort) for a new-onset behavior. When the child puts him- or herself or others in danger, medication may become a "front-line" intervention.

Medications should be considered when behaviors (eg, self-injurious behaviors, aggression, anxieties, obsessions, stereotypies, sleep disorders) negatively affect or prevent the progress of educational, therapeutic, and behavioral interventions. When choosing a medication, the clinician should target the most challenging behavior. When prescribing the medication, the clinician should "start low and go slow." Children should be monitored for adverse effects, especially when they are unable to verbalize symptoms. Parents of children who have ASDs often seek complementary and alternative medicine (CAM). (17) Although it is difficult to be familiar with the myriad CAM possibilities, the PCP should be familiar with some of the most popular local options and provide the family with evidence-based information, if available. The AAP clinical report provides a detailed review of the management of children who have ASD. (18)

Conclusion

A growing body of evidence is revealing that many children who have autism can be recognized before age 2 years. PCPs now have the opportunity to play a pivotal role in early recognition. PCPs who recognize autism in children younger than 2 years of age, refer them for comprehensive evaluations, and help them enroll in appropriate early intervention programs benefit the chil-

dren, their families, their future schools, and society. Early diagnosis and intervention improve outcomes, empower families, decrease the need for special education services in later years, and increase the child's chance for independence and gainful employment as an adult, especially if there are no coexisting cognitive deficits. A high degree of suspicion and a solid understanding of early deficits in social and preverbal skills are critical to early recognition. The AAP ASD clinical reports and toolkit should help guide and support PCPs in this endeavor.

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Suggested Reading

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Autism Speaks, a nonprofit organization dedicated to increasing awareness of autism and raising money to fund autism research, together with First Signs, the leader in early identification and intervention of children with developmental delays and disorders, and Florida State University, have developed a first-of-its-kind web-based video glossary to help professionals learn more about the early warning signs of ASDs. The glossary, available free of charge at www.autismspeaks.org, www.firstsigns.org, and http:// firstwords.fsu.edu, contains more than 100 video clips that illustrate both typical and atypical development.

PIR Quiz

Quiz also available online at www.pedsinreview.org.

5. Differences in pointing behaviors, as an example of "joint attention," may be used in descriptions of children believed to have autism. Mature joint attention is demonstrated *best* with a point whose purpose is to:

   A. Comment.
   B. Direct.
   C. Distract.
   D. Label.
   E. Request.

6. Of the following, the *best* DSM-IV-TR-based criteria to identify children younger than 2 years of age who have autism are:

   A. Abnormal conversational skills and stereotypic language.
   B. Delay in achieving speech and language milestones.
   C. Failure to form age-appropriate peer relationships.
   D. Impairment in use of nonverbal behaviors.
   E. Ritualistic behaviors and need for routine.

7. The failure of an 18-month-old child who has autism to respond to his or her name when called is believed to be the result of deficits in:

   A. Auditory processing.
   B. Cognition.
   C. Hearing.
   D. Receptive language.
   E. Social relatedness.

8. Careful observation of typical children at play has been important in better understanding children who have autism. Children who have autism rarely demonstrate evidence of:

   A. Constructive play.
   B. Oral motor play.
   C. Roughhouse play.
   D. Sensory motor play.
   E. Symbolic play.