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Clinical Assessment and Management of Toddlers With Suspected Autism Spectrum Disorder: Insights From Studies of High-Risk Infants

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ABSTRACT

With increased public awareness of the early signs and recent American Academy of Pediatrics recommendations that all 18- and 24-month-olds be screened for autism spectrum disorders, there is an increasing need for diagnostic assessment of very young children. However, unique challenges exist in applying current diagnostic guidelines for autism spectrum disorders to children under the age of 2 years. In this article, we address challenges related to early detection, diagnosis, and treatment of autism spectrum disorders in this age group. We provide a comprehensive review of findings from recent studies on the early development of children with autism spectrum disorders, summarizing current knowledge on early signs of autism spectrum disorders, the screening properties of early detection tools, and current best practice for diagnostic assessment of autism spectrum disorders before 2 years of age. We also outline principles of effective intervention for children under the age of 2 with suspected/confirmed autism spectrum disorders. It is hoped that ongoing studies will provide an even stronger foundation for evidence-based diagnostic and intervention approaches for this critically important age group. Pediatrics 2009;123:1383–1391

Autistic disorder is the most severe form of a spectrum of related disorders (autism spectrum disorders [ASDs]) characterized by impairments in reciprocal social interaction and communication and by the presence of repetitive, inflexible behavior. At an estimated prevalence of 1 in 150, the ASDs are among the most common forms of severe developmental disability. Parents of children with ASDs generally identify concerns by the age of 12 to 18 months. However, recent US data suggest that the average age of diagnosis remains at ~4 years and possibly older in socioeconomically disadvantaged groups. In part to address the unacceptably long interval between parents’ initial concerns and confirmation of diagnosis, the American Academy of Pediatrics (AAP) and other groups have recently published clinical practice guidelines on the early identification, screening, and diagnosis of ASDs. With AAP recommendations that all 18- and 24-month-olds be screened for ASDs and active public awareness campaigns by the Centers for Disease Control and Prevention and other public and private organizations, there is increasing demand for diagnostic assessment of very young children. However, unique challenges exist in applying current diagnostic guidelines for ASDs to children under the age of 2, because standard criteria of the Diagnostic and Statistical Manual, Fourth Edition and International Classification of Disease, Tenth Edition, gold-standard diagnostic tools, and even the best judgment of experienced clinicians have rarely been applied to this age group. To address these challenges and...
develop an initial framework for early diagnosis and treatment of ASD* in children younger than 2 years, we address the following questions:

1. What do we know about the early signs of ASDs?
2. Can ASDs be detected in children younger than 2 years by primary care providers?
3. What is the current best practice for diagnostic assessment of ASDs before 2 years of age?
4. What are the challenges in establishing ASDs diagnoses in this age group?
5. What interventions can be offered to children under the age of 2 with suspected/confirmed ASDs?

WHAT DO WE KNOW ABOUT THE EARLY SIGNS OF ASDs?

Although as yet there are no reliable biological markers for ASD,† much has been learned about early behavioral signs. Initially, research on the early manifestations of ASDs was mainly confined to parents’ retrospective reports and early home videos.18,19 However, retrospective findings were supplemented recently by prospective studies of infants at high risk, allowing considerable progress in understanding the early emergence of ASDs.20

Retrospective Studies

Retrospective studies involving parental recollections provide several important insights into the early development of children with ASDs. Some parents of children later diagnosed with ASDs recall developmental differences in the first few months of life, although a larger proportion became concerned during the second year.4,5,21,22 Variability exists in the types of concerns that parents recall, although concerns involving delayed speech and language development (and/or lack of responsiveness [eg, when the child’s name is called], which may be perceived as a possible hearing problem) are among the most common.4,7 Other concerns include extremes in behavioral reactivity and disruption in social-communicative, play, and motor development.21,23 Concerns also may be expressed about sleep and feeding.19 In 20% to 50% of children with autism, parents retrospectively describe a pattern of regression involving loss of speech and/or social-emotional connectedness during the second year of life, most often around 18 months of age.19,24-28

Analysis of early home videos also suggests that some, but not all, children later diagnosed with ASDs show signs of atypical development by their first birthday or shortly afterward.14 Again, the evidence implicates disruptions in multiple developmental domains. Social-communicative findings include atypical patterns of social orienting, joint attention (ie, sharing attention with others), imitation, and affect regulation, as well as increased negative affect and ambiguous affective expressions and reduced use of gestures.26-35 Infants later diagnosed with ASDs show decreased flexibility, variety, and appropriateness of object-oriented play compared with infants later diagnosed with mental retardation.36

Prospective Studies

Studies of infants with an older sibling with ASD constitute a promising new approach to investigating early development in ASDs.20,37-38 These infants have an increased risk of developing ASDs, currently estimated at 5% to 10%,3 20-fold higher than the risk in the general population.19-41 Complementing this approach is recent research aimed at identifying features that differentiate ASDs from both other developmental disorders and typical development among 12- to 24-month-olds who have failed a screen for communication deficits.42,43 In both types of prospective studies, systematic measures of behavior are collected early in development, with assessments for ASDs conducted at 24 to 36 months when more reliable diagnoses are possible.

To date, prospective studies have shown that by 12 to 18 months of age, infants later diagnosed with ASDs are distinguished from other infants at high risk by impairments and/or delays in 1 or more of the following domains: (1) visual (atypicalities in visual tracking and fixation on objects44,45 and prolonged visual inspection of objects41-46); (2) motor (decreased activity levels,45 delayed fine and gross motor skills,47,48 and atypical motor mannerisms45,49); (3) play (delays in the development of motor imitation,41 limited toy play,41,50 and repetitive actions with toys43,45,46,49); (4) social-communication (atypicalities in eye gaze, orienting to name, imitation, social smiling, reactivity, and social interest and affect, with reduced expression of positive emotion17,43-45,50,51); (5) language (delays in babbling [especially back-and-forth social babbling], verbal comprehension and expression, and in gesturing, as measured with standardized assessments17,43,44,48,52,53); and (6) general cognitive development (at least 2 groups have reported slower acquisition of new skills [associated with declining standard score] in a subset of toddlers subsequently diagnosed with ASDs48,45,46). Most studies have focused on differences between infants subsequently diagnosed with ASDs and those with typical development, although many features (including atypical exploration of toys, repetitive motor behaviors, and reduced social-communication and shared positive emotion) also distinguish ASDs from language and other developmental delays at this early age.42,43,50 There may be a subgroup of infants with impairments consistent with an ASD diagnosis as early as 13 to 14 months.50,54 Even in those with a later onset, symptoms are generally apparent by the age of 18 to 24 months.43,44,50

Prospective studies of ASDs have demonstrated the feasibility of detecting emerging signs of the disorder through intensive monitoring of high-risk cohorts.45,50 Although there is variability in the onset and precise

*In this article, “ASDs” refers to both autistic disorder and pervasive developmental disorder not otherwise specified; Asperger syndrome would rarely be diagnosed in a 2-year-old, because the criteria include language skills (presence/absence of phrase speech) at 33 months. We also acknowledge that some clinicians would delay diagnostic subtyping (and use the broader term “ASD”) because of variation in symptom trajectories after age 2 and difficulty applying some criteria in very young children, such as quality of peer relationships.

†Accelerated head growth early in life is an intriguing candidate (Cody-Hazlett et al, 2006; Courchesne et al, 2005; Dawson et al, 2006), but there have been no published prospective data to assess its potential predictive validity.
nature of these signs, children with ASDs generally show atypical development across cognitive and speech/language, social-communicative, and/or sensory-motor domains by 2 years of age. There is overlap between these signs (summarized in Table 1) and previously published lists of “red flags” for ASDs.13 As we shift our focus from infants at high risk to those in the general community, we must consider how knowledge about early signs can be applied in a surveillance and/or screening context.

**CAN ASDs BE DETECTED IN CHILDREN YOUNGER THAN 2 YEARS BY PRIMARY CARE PROVIDERS?**
The AAP recommends ASD screening at ages 18 and 24 months as part of developmental surveillance during regular health visits.13 The Checklist for Autism in Toddlers (CHAT)55,56 is the only ASD screen that has been assessed in a geographically defined cohort with comprehensive long-term follow-up of diagnostic status in both screen-positive and screen-negative children, needed to accurately estimate sensitivity and specificity.57 Using its original criteria (absence of gaze monitoring, pointing to show, and pretend play, as measured by parental questionnaire and direct observation by a health professional), only 18% of the children who were diagnosed with ASD between 20 and 24 months were detected by the CHAT at 18 months.57 Even with less stringent criteria (absence of pointing to show), the sensitivity of the CHAT was only 38%.

The Modified Checklist for Autism in Toddlers (M-CHAT)§58,59 is a parent questionnaire that includes items from the CHAT but covers a broader range of signs and a wider age range (16–30 months). The M-CHAT includes a follow-up interview in which the parent is asked in more detail about symptoms endorsed on the questionnaire. This interview increases the specificity of the M-CHAT and is highly recommended.§ The sensitivity of the M-CHAT is reported to be as high as 85%, but in research to date, assessment for ASDs has mainly been limited to screen-positive children (screen-negative children were only evaluated if identified by health care providers as being at risk or if identified through re-screening 2 years later). Thus, available data allow the positive predictive value (PPV) of the M-CHAT to be more accurately estimated than its sensitivity. The follow-up interview markedly increases the PPV of the M-CHAT in toddlers screened by community physicians, from 11% with the questionnaire alone to 65% among toddlers who exceed the screening cutoff on both the questionnaire and interview.90 Thus, ASD screening may be most informative when used as a starting point for active and repeated discussion regarding parental concerns. Recent data suggested that the PPV of the M-CHAT may be lower in children aged 16 to 23 months than those older than 24 months (PPV estimated at 28% and 61%, respectively, in a large community sample60), which emphasizes the importance of repeated assessment.

There are other measures that may help detect early behavioral indicators of ASDs. The Infant Toddler Checklist (ITC)61 was designed to screen for communication delays (recommended in the AAP guidelines for toddlers younger than 18 months).12 Repeated screening of 9- to 24-month-olds by using the ITC in a recent community sample of 5385 toddlers identified >90% of those with ASDs, although further evaluation was needed to distinguish ASDs from other communication delays.62 The Screening Tool for Autism in Two-Year-Olds (STAT) is an interactive assessment designed to identify signs of ASDs in referred or at-risk children (ie, “level 2” screener). The STAT, previously designed to assess children >2 years old, may also be informative in the second year (sensitivity and specificity recently estimated at 95% and 73%, respectively, in a sample of 71 infants aged 12–23 months and at high risk).63 Instruments designed to detect behavioral signs of children as young as 12 months have also shown promise, including an interactive measure, the Autism Observation Scale for Infants,54,64 and a parent questionnaire, the First Year Inventory,56,67 although additional research is needed to assess their clinical utility.

**Overall, community-based screening provides a means to systematize ASD surveillance and maximize**
opportunities to detect early signs. The degree to which available tools accurately classify children as being at high or low risk is still being evaluated, although substantial progress is being made. Community pediatricians and other health care providers play an essential role in identifying very young children who are at risk for ASDs through active surveillance for early signs. Nevertheless, uptake of ASD screening into general pediatric practice remains limited,64 and further efforts to understand and address barriers to implementation are needed.65,70 Notably, ASD-specific screeners can potentially identify toddlers with ASD who are not flagged by either parents or clinicians in a general surveillance context,58,62 suggesting that incorporating such tools provide “added value” to routine surveillance. Some community physicians may be reluctant to raise concerns about early signs of ASDs to avoid upsetting parents,71 but numerous studies have indicated that parents would prefer to be informed and that delaying discussion can create much greater distress.72 Ultimately, careful attention to parents’ concerns and atypical patterns of toddler behavior in the child’s “medical home,” supported and complemented by ASD-specific screening tools, may help reduce the time between initial emergence of ASD symptoms and referral for specialized assessments. Siblings of children with ASDs are at increased risk of both ASDs and other developmental concerns (eg, language delay) and may warrant heightened vigilance.12

WHAT IS CURRENT BEST PRACTICE IN THE DIAGNOSTIC ASSESSMENT OF CHILDREN YOUNGER THAN 2 YEARS?

The unique challenges of evaluating ASD symptoms in children younger than 2 years require the diagnostic process to be led by a clinician experienced in developmental disabilities assessment in this age group. Notably, many of the early behaviors associated with ASDs overlap with those associated with language delay and intellectual disability.18,73 Thus, a thorough appraisal of the child’s development in multiple domains, including cognitive, language, communication, and adaptive functioning, as well as social interaction, communication, and play skills, is essential for differential diagnosis12,74 (see Table 2).

Social and communication development are optimally assessed by using a combination of direct observation and parent interview. Low-frequency behaviors are more likely to be identified on the basis of parental report,75 whereas professionals may have a better frame of reference from which to judge some classes of behaviors (eg, joint attention) relevant to detecting ASDs in the first 2 years of life.76,77 Standardized diagnostic instruments such as the Autism Diagnostic Observation Schedule (ADOS)78 are informative and can help guide clinical decision-making. The ADOS performs best in children with a nonverbal mental age of ≥15 months.78,79 In younger children, sensitivity of the instrument is excellent, but its specificity is lower.78,80 Recent modifications to the ADOS, including a revision of the diagnostic algorithms81 and the development of a toddler module with a downward extension of tasks and behavioral codes, may enhance stability of earlier diagnoses. However, with reduced specificity in very young children, interpretation of ADOS scores (at least until the toddler module is widely available) requires careful consideration of the overall clinical context, particularly the child’s cognitive development.

Two of the most widely used standardized measures for assessing cognitive development in infants and toddlers are the Bayley Scales of Infant Development62 and the Mullen Scales of Early Learning.82 These instruments help determine if children are meeting expected milestones in targeted domains (including verbal, nonverbal, and motor skills), identify important intraindividual discrepancies (eg, in nonverbal cognitive versus social skills), and provide a developmental context for interpreting observed behaviors. Some of the cardinal symptoms of ASDs, such as impairments in joint attention behaviors, must be considered relative to developmental level (eg, using eye gaze to direct parents’ attention does not emerge until 10 months in typical development).64 Observing the child’s problem-solving strategies can also provide insights into attentional, motivational, and cognitive factors that may affect his or her performance and influence his or her response to treatment. Assessment of adaptive skills (eg, by using the Vineland Adaptive Behavior Scales II)63 provides the clinician with information regarding the child’s ability to translate his or her cognitive assets into successful functioning in day-to-day situations.86–89 Assessment of language and communication skills is also essential. In addition to standardized measures of expressive and receptive language (eg, the Preschool Language Scales-IV),90 informative measures in this age group include the Communication Symbolic Behavior Scales Developmental Profile (CSBS DP),61 which involves observation of the child’s communication during semistructured play tasks, and the MacArthur-Bates-Communication Development Inventory,91 which is a parent-report inventory of words and

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**TABLE 2 Recommendations for Diagnoses of Possible ASDs in Infants and Toddlers**

The diagnostic process must be led by a clinician experienced in working with infants and toddlers with ASDs.

Information should be obtained from

A standardized observation with predetermined tasks and scoring
Parent report and developmental history concerning social and communication skills, repetitive interests/behaviors, as well as sleep, feeding, and sensorimotor issues
Structured observation of social, communication, and play skills and repetitive interests/behaviors
Standardized assessment of cognitive, language, and adaptive/daily living skills
Judgment of an experienced clinician, in consultation with a multidisciplinary team, based on interpretation of all available results

Treatment recommendations should be tied to specific symptoms, although often these are symptoms having to do with delays or lack of behaviors rather than the presence of abnormalities
Parents should be given clear information about what will happen next
What are appropriate treatment goals?
What are the options for treatment now and later?
How can they monitor progress?
When should the child be reevaluated?
gestures. Normal hearing should always be confirmed for children with language delay, including those at risk for ASDs. Other assessments should be determined by specific concerns (eg, motor delays, sleeping or eating problems).44,93

As in older children, ASD diagnoses for toddlers should be based on clinical best judgment (in consultation with a multidisciplinary team), taking into account all available information regarding the child’s developmental and medical history, observed and parent-described ASD symptoms, and cognitive, language, and adaptive skills using standardized measures. The added challenges of clinical decision-making in children younger than 2 years of age emphasize the importance of a close partnership between community physicians and specialized diagnostic teams. Although access to such teams varies between communities, such expertise and associated training is needed to build system capacity to ensure appropriate follow-up for children identified through surveillance and screening initiatives.

WHAT ARE THE CHALLENGES IN ESTABLISHING ASD DIAGNOSES BEFORE THE AGE OF 2 YEARS?

Although there have been significant advances in our ability to detect early signs of ASDs, diagnostic evaluation in children younger than the age of 2 remains challenging.

First, some children with ASDs, particularly those with more intact language and intellectual development, may have more subtle symptoms at an early age.44,50 Speech delays are often the concern that parents first report, so for children without marked delays, early symptoms may be less apparent. As well, a proportion of children with ASD symptoms may show “plateauing,” deceleration, or frank losses in cognitive and social development or functioning in the second year.44,48,50 Thus, mild symptoms and even an absence of symptoms at 18 months do not “rule out” a later diagnosis of ASDs. Ongoing surveillance and follow-up are essential, particularly for children who are referred because of early concerns but do not initially receive an ASD diagnosis.

Second, it may be difficult to distinguish between ASDs and other atypical patterns of development at an early age. This may be especially true among infant siblings of children with ASDs, who are at risk not only for ASDs but also for a broader spectrum of delays including but not limited to the areas of emotion expression and referential communication.20,53,94–98 Similarly, early diagnosis of ASDs may be difficult in toddlers with severe developmental delays, or impairments in vision and/or hearing, for whom standardized diagnostic instruments have shown limited specificity.80,99

Third, there is still uncertainty about the stability of ASD diagnoses in children younger than 2 years of age. Numerous studies have shown that an autism spectrum diagnosis at age 2, based on the clinical judgment of experienced clinicians using information from standardized instruments, is generally reliable and stable well into the school-age years. However, there have been few published studies of the stability of ASD diagnoses for those under the age 2. Chawarska et al77 reported that within a referred sample of 31 toddlers first assessed at the age of 14 to 25 months, 27 were diagnosed with ASDs, all of whom retained the diagnosis when reassessed 1 year later. However, Kleinman et al100 reported that 20% of 61 children aged 14 to 35 months initially diagnosed with ASDs no longer met ASD criteria 2 years later, and although some of the children who moved off the ASD spectrum continued to have another type of diagnosis, more than one third of them had typical development at follow-up.

Thus, questions remain about how to best apply and interpret ASD diagnostic criteria and symptom measures in this age group. Even very experienced diagnosticians may be faced with clinical uncertainty in children younger than 2 years, which can be difficult for both the clinical team and families seeking answers and help for their children.77,92,101 However, with due consideration to the burden of uncertainty and concerns regarding over-diagnosis,102 it bears emphasizing that providing “false hope” and postponing a diagnosis also incurs costs to children and families. In our experience, the most critical issue in mitigating the negative impact of uncertainty is timely access to appropriate intervention services for the child and supports for the parents. Hence, in addition to sensitive feedback regarding diagnosis, parents must be given clear information about treatment options and referred to appropriate interventions and supports.

WHAT INTERVENTIONS CAN BE OFFERED TO CHILDREN YOUNGER THAN 2 YEARS OF AGE WITH SUSPECTED/CONFIRMED ASDs?

As our ability to identify toddlers with suspected or confirmed ASDs improves, the need for research on the efficacy of very early intervention approaches becomes critical. Although a variety of evidence-based interventions exist for preschool-aged children with ASDs,103 it is not clear that such interventions would be equally beneficial, or even appropriate, for children younger than 2. In this section, we comment on the unique developmental characteristics of children with ASDs younger than 2 years of age and the implications for the design of effective interventions.

Infants and toddlers differ from preschool-aged children in the nature of their social relationships, in their cognitive and communicative processes, their learning characteristics, and their daily routines. Infants depend on observational and experiential learning within their natural social and physical environments. Interactions that are rooted in social play, as well as those that occur within the context of caregiving, represent fundamental learning opportunities for infants. The qualities of responsibility and sensitivity in key family members (eg, attending to and joining in with the child’s current focus of interest) can have a tremendous impact on the learning and development of infants and young children with and without disabilities.102,104 In contrast, a directive style of relating can be deleterious to language, emotional/behavioral, and social development in infants and toddlers.105

As the ability to initiate joint attention consolidates around 12 months, toddlers begin to communicate in-
tentionally by using spoken language and gestures. Between 12 and 24 months, toddlers also progress from sensory-motor exploration to using objects symbolically (eg, by pretending to drink from a cup). The capacity to imitate observed acts also increases markedly in the second year of life. Social and learning characteristics of infants and toddlers contrast sharply with those of preschool-aged children, whose experience involves higher levels of autonomy, less contextualized learning and memory, and greater facility with symbolic thought and communication. Interventions that address the learning characteristics of infants and toddlers should focus on natural learning environments, child initiative and sensory-motor exploration, and the development of nonverbal intentional communicative acts and reciprocal play with social partners.

Twelve empirically based principles in a position statement by the National Association for the Education of Young Children emphasized children as active learners, the importance of play for learning, the social contexts of learning, and the interdependence of physical, cognitive, and emotional activity. Responsive and sensitive care-taking, enriched language environments using responsive rather than directive interaction styles, environments that provide opportunities for toddlers to have an active role in their own learning, and interventions that are individualized and targeted to specific skills in children and parents characterize efficacious infant/toddler interventions. Developmental principles of intervention for all infants and toddlers should characterize interventions for those with ASDs unless empirical evidence indicates differently. Moreover, although interventions designed for older children can certainly inform research and practice, existing programs cannot simply be extrapolated from older to younger children.

Published intervention research for children with ASDs younger than the age of 2 is limited to descriptive studies and recent studies that used quasi-experimental designs. Several randomized treatment trials are currently underway, including trials involving the Early Start Denver Model, Responsive Teaching, Hanen’s “More Than Words,” the Early Achievements model, and the Social Communication, Emotional Regulation, and Transactional Supports (SCERTS) model. Although ASD-specific interventions for children younger than 2 years of age are not yet widely available outside of a research context, there are existing services from which toddlers and their families can benefit, including more general early intervention services (in the United States, these would be administered through the state “birth-to-three” system) and speech and language interventions. Parents should also be informed about support groups and other community resources available to families of children diagnosed with ASDs and/or other developmental delays. There are also excellent on-line resources available to assist parents in learning about publicly funded services (eg, www.autismspeaks.org/community/family.services/100.day.kit.php).

SUMMARY AND CONCLUSIONS

Efforts directed toward earlier detection of ASDs, recently formalized in the AAP practice guidelines, have heightened the need for evidence-based approaches for infants and toddlers identified as being at increased risk. Fortunately, advances in the recognition of early signs of ASDs and increasing experience with the assessment of infants and toddlers at high risk have helped establish the feasibility of ASD diagnoses under the age of 2. Despite the challenges of clinical uncertainty, there is much that can be offered to toddlers identified as being at increased risk of ASDs. First, identification of concerns should lead to further assessment of the child’s social, communication, and play development in the office. This should include discussion of parents’ concerns (including the follow-up interview to the M-CHAT questionnaire, if administered) and observation of how the child interacts with his or her parents, uses eye contact to regulate interactions, responds to simple social games such as peek-a-boo, and/or attempts to draw others’ attention to interesting things in the environment. Whenever possible, information about how the child interacts in other social environments (eg, day care) should also be gathered. The child’s hearing should be assessed by an audiologist. If concerns remain, the child should be referred to early intervention services and for further evaluation by professionals experienced in developmental disabilities assessment in this age group. Interventions should ultimately be directed toward specific functional concerns and be informed by key developmental principles, including the child’s role as an active learner, the social contexts of learning, and the pivotal role of the parent-child relationship. It is important to support the parents throughout the process, acknowledging uncertainty where it exists but also emphasizing that we now know more than ever before about how to care effectively for toddlers with suspected ASDs. Ongoing studies (particularly those implemented within a broad range of community contexts) will continue to provide an even stronger foundation for evidence-based diagnostic and intervention approaches for this critically important age group.

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