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Practitioner's Guide to Assessment of Autism Spectrum Disorders in Infants and Toddlers

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Abstract

Recent advances in clinical research have made it possible to diagnosis autism spectrum disorders (ASD) as early as the second year of life. The diagnostic process early in development is often complex, and thus, familiarity with the most recent findings in clinical assessment instruments, early symptoms, and developmental trajectories of young children with autism is essential. This paper provides an empirically based practitioner's guide to issues and concerns related to early diagnosis of ASD in very young children, documentation of the course and patterns of ASD symptomatology in infants and toddlers, and current understanding in the field of diagnostic outcomes of children identified in the first and second year of life.

Keywords

Early diagnosis; Clinical evaluation; Infants and toddlers

Introduction

Research now suggests that the diagnosis of autism spectrum disorders (ASD) can be reliably made in the second year of life, and these early diagnoses appear to be relatively stable over time (Chawarska et al. 2009; Rogers 2009). However, diagnosis of very young children can be quite complex, as the presentation of children in the first 2 years of life can vary from that of an older child with autism. Rather than observing the full-blown disorder, the clinician is often observing the unfolding symptoms of social and communicative dysfunction. Because the eventual severity of the disorder is as yet unknown at these early ages, it is common for clinicians to refer to the emerging disorder as ASD (indicating the

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presence of a clinically significant disorder on the autism spectrum) rather than by the current diagnostic labels of Autistic Disorder, Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS), or Asperger's Disorder. Symptoms of ASD often present in the context of marked developmental delays (Akshoomoff 2006; Chawarska et al. 2009), making it challenging to assess how much the child's social disability may exceed delays in other domains. In this case, it can also be difficult to ascertain how behaviors that are often associated with cognitive disability, such as repetitive behaviors, contribute to the diagnostic formulation. The complexity that these factors introduce can be especially challenging for clinicians who, until now, have not been called upon to make early diagnoses. Thus, this paper translates research on early ASD symptomatology into a practical guide for psychologists, child psychiatrists, developmental pediatricians, and other practitioners faced with the growing need to provide differential diagnoses for very young children presenting with concerns for autism.

Current Diagnostic Trends

Despite advances in early identification, diagnosis of ASD is often delayed until early preschool age. While most parents report having concerns about their children in the first or second year of life (Baghdadli et al. 2003; De Giacomo and Fombonne 1998; Volkmar et al. 1985; Chawarska et al. 2007), parents report that the time between their first concerns and their child's ASD diagnosis was, on average, 1.7 years for autism, 2.1 years for PDD-NOS, and 4.6 years for a diagnosis of Asperger's Syndrome, with the average age at first diagnosis around 3.2, 3.7, and 7.2 years for Autism, PDD-NOS, and Asperger's Syndrome respectively (IAN 2010). These delays can be due to a number of factors, including inadequate screening practices (e.g., Dearlove and Kearney 1990; Sices et al. 2003), limited parental awareness of symptoms of ASD or delayed action by pediatricians after initial concerns are voiced by parents (e.g., Shevell et al. 2001), and limited availability of specialized diagnostic services for children under the age of 3 years.

However, due to increasing awareness of early symptoms of autism amongst parents (Chawarska et al. 2007, 2009; Ozonoff et al. 2009) and professionals (Johnson et al. 2007; Myers et al. 2007; Zwaigenbaum et al. 2009), development of early screening instruments (e.g., Robins et al. 2001; Stone et al. 2008; Wetherby et al. 2004), as well as very successful educational campaigns by the US Center for Disease Control and parent organizations such as Autism Speaks, the number of children under the age of 3 years undergoing comprehensive diagnostic evaluations and entering early intervention programs in the US has increased considerably in the past decade (Chawarska et al. 2008; Dawson 2008; Dawson et al. 2010; Koegel et al. 2008; Wetherby and Woods 2008; Zwaigenbaum et al. 2009).

Stability of Diagnosis in the Second Year of Life

Identifying a child as having an ASD diagnosis in the first or second year of life can be challenging, as development during the first years of life progresses at a rapid and sometimes uneven pace across developmental domains. However, prospective longitudinal studies of toddlers diagnosed with ASD suggest that both short- and long-term stability of

expert clinical diagnosis is very good, with the majority of children who receive an ASD diagnosis in the second year of life continuing to do so at 3 or 4 years of age (Chawarska et al. 2007; Cox et al. 1999; Turner and Stone 2007), with rates ranging from 80 to 100% (Kleinman et al. 2008; Chawarska et al. 2009; Lord et al. 2006). Children who are initially diagnosed with ASD but do not retain the diagnosis have been found to be those with milder symptoms, particularly in the social domain, as well as higher cognitive functioning (Turner and Stone 2007). However, differentiation within the autism spectrum appears to be more challenging (Chawarska et al. 2007; Cox et al. 1999; Stone et al. 1999), and marked changes in clinical presentation within the spectrum over time are to be expected as symptoms of autism evolve and verbal and nonverbal cognitive skills improve (Lord et al. 2006; Charman et al. 2005; Chawarska et al. 2007, 2009; Kleinman et al. 2008). Because research is mixed regarding differentiation within the autism spectrum in toddlers, stating that the child has an ASD is usually sufficient. Additionally, this approach is also congruent with the current direction of the DSM-V revisions, which may no longer provide separate diagnostic categories for ASDs, but rather the diagnosis of Autism Spectrum Disorder, with severity ratings based on the level of support required (for more information, see http://www.dsmv.org).

Diagnosis in the First Year of Life

The first empirical reports regarding the development of infants who later are diagnosed with ASD have begun to emerge. Based on the extant evidence it appears that most infants who go on to develop ASD appear to have relatively intact eye contact and social smile at 6 months of age (Bryson et al. 2007; Landa and Garrett-Mayer 2006; Ozonoff et al. 2010; Young et al. 2009; Zwaigenbaum et al. 2005), with the frequency and quality of these social behaviors declining between 6 and 12 months (Ozonoff et al. 2010). There is an emerging consensus across a number of studies that symptoms of social disability become more apparent, at least on the group level, around 12 months of age (see Rogers 2009 for a review). Behaviors affected at this age include responsivity to name (Nadig et al. 2007), atypical object exploration and repetitive behaviors (Kim and Lord 2010; Ozonoff et al. 2008), and language and nonverbal communication (Landa and Garrett-Mayer 2006; Paul et al. 2011; Presmanes et al. 2007; Yoder et al. 2009).

At present, diagnosis of autism within the first year of life poses many challenges, and there are not yet clear empirically-based guidelines for this process. Thus, clinicians who are asked to evaluate the presence of ASD in infants should exercise great caution. If an infant exhibits significant delays in social interaction with or without the accompanying developmental and language delays, clinicians are advised to recommend services as appropriate to address the identified delays and conduct a re-evaluation within 3–6 months. The re-evaluation should focus on social-cognitive and other developmental skills and determine their rate of progress over time, which would be helpful for clarifying the diagnosis. At present, no reliable diagnostic procedures for infants under the age of 12 months have been established, though work in this direction is in progress.

Clinical Evaluation of Autism in Toddlers

Considering both the developmental and pervasive nature of the disorder, making a diagnosis of ASD is a complex process. Many ASD symptoms such as delayed development of expressive language, diminished social responsivity, behavioral difficulties, and repetitive behavior can be found in other disorders and syndromes. The presence of intellectual disability further complicates the differential diagnostic process, especially in very young children where deficits in social engagement due to severe cognitive impairment need to be differentiated from those resulting from ASD. Under optimal conditions, diagnosis of ASD in toddlers is based on assessment of functioning in multiple areas, including verbal and nonverbal developmental skills, social communication and interaction skills, presence of atypical motor and sensory behaviors, adaptive levels of functioning, as well as careful review of the child's health, development, and family history. Genetic screening for chromosomal and other genetic abnormalities as well as neurological, vision, and hearing exams are often indicated. In addition, practitioners should be aware of possible comorbid conditions, including issues related to attention, attachment, and anxiety. Referral for an evaluation for disruptive or self-injurious behaviors is also warranted in some cases. Results of these assessment procedures are necessary not only for making differential diagnosis but also for assembling an individualized treatment plan tailored to the child's unique profile of needs and abilities (Klin et al. 2005; Zwaigenbaum et al. 2009). The practitioner may also wish to consult guidelines and handbooks for the evaluation of ASD to complement the information provided below (e.g., Chawarska et al. 2008; Klin et al. 2005; Filipek et al. 2000).

Although standardized diagnostic assessments for infants and toddlers yield informative data that influence diagnostic decision making, the gold standard for diagnosis continues to be "expert clinical opinion" (Chawarska et al. 2007; Bishop et al. 2008; Volkmar et al. 1994); however, what constitutes an expert clinician is not well delineated. For the purposes of this guide, it is proposed that an expert clinician is one who holds the professional credentials necessary for providing a diagnosis of ASD and is knowledgeable regarding (a) the typical course of development, (b) developmental deviations associated with a myriad of non-ASD disorders of infancy and childhood, and naturally, (c) the onset and course of ASD symptomatology. Considering the complexity of the clinical presentation of toddlers with developmental disorders, a multidisciplinary assessment is often necessary to clarify diagnostic ambiguities and to design an individualized treatment program (Klin et al. 2005). Such multidisciplinary teams often include a licensed clinical or developmental psychologist, speech pathologist, occupational or physical therapist, social worker, and developmental pediatrician. Often, teams are comprised of professionals who work in the same clinical practice within a university, medical, or community setting; however, as the need for early diagnostic evaluations increases, many practitioners who work in solo practices will be called upon to evaluate young children for whom ASD is suspected. Working independently, however, should not limit the practitioner's ability, or motivation, to glean information from other disciplines. Instead, an experienced diagnostician working in private practice should be mindful to work collaboratively with the child's primary care

physician and make referrals to professionals from other disciplines whenever the limits of their professional competence result in a less thorough evaluation than what is indicated.

Many structured and semi-structured assessments, as well as observational and parent report measures, are available for the purposes of informing the diagnostic assessment of infants and toddlers suspected of having ASD. While it is beyond the scope of this guide to review every available measure relevant to this age group, a selection of psychometrically sound measures used in both research and clinical practice is provided in Table 1. The reader is encouraged to familiarize themselves with the measures noted and select those measures that best fit the needs of the child being evaluated. Outside of a structured research evaluation, it is quite reasonable that different measures will be utilized for different children, and different clinics will rely on measures that best meet the needs of their practice. Thus, no standard assessment battery is recommended; however, the assessment should be comprehensive and tap every area of development relevant to making an informed differential diagnosis. Additionally, it is essential that the instruments used for diagnostic and assessment purposes with young children are standardized, reliable, and norm-referenced (e.g., developmental or language tests), and, in the case of screening and diagnostic instruments, have established sensitivity, specificity, as well as predictive positive and negative values. Heightened awareness regarding the diagnosis of ASD in very young children has generated increased interest in developing screening and diagnostic instruments that target this age range. Two promising instruments include the Autism Observation Scale for Infants (AOSI; Bryson et al. 2008), which is an observational measure for infants from 6 to 18 months of age, and the First Year Inventory (FYI; Reznick et al. 2007), which is a parent questionnaire for one-year-old children, although these measures are currently primarily used in research settings.

Although standardized assessment measures are designed to elucidate key areas relevant to an ASD diagnosis, the practitioner should view the entire evaluation, from the initial meeting in the waiting room through the diagnostic conference, as an opportunity for behavioral observation. Oftentimes children with ASD (and children in general) may be more or less likely to exhibit particular behaviors within the context of structured activities (e.g., a developmental assessment) compared to free play activities (e.g., the waiting room, or play-based assessment such as the ADOS-G). For children with ASD, the comparison between these types of settings is often essential for treatment planning and determining the level of support a child may require to engage in developmentally appropriate behavior. For example, if the child displays significantly greater social interaction skills during very structured activities (e.g., completing a clear task at a table) than during free play, this speaks to need for intervention to focus more heavily on spontaneous initiation and generalizing these skills to unstructured settings. Thus, a comprehensive evaluation is more than the administration of multiple assessment measures, it is the combining of information from multiple sources, settings, and time points.

There are many factors that influence how an evaluation unfolds for a particular child, but perhaps the most significant factor pertains to where the child is in the diagnostic process. For practitioners working in an ASD specialty clinic, it is likely that someone in the child's life (e.g., a primary care, early intervention, or daycare provider) has already expressed

concern regarding ASD symptoms and/or performed primary or secondary screening for ASD. In such situations, parents are typically aware that their child may present with concerning features, and they may have already obtained an evaluation at another site (e.g., a non-specialty clinic, a local early intervention agency) and are subsequently seeking diagnostic clarification with regard to ASD. However, the reality is that although screening is recommended by the American Academy of Pediatrics (Johnson et al. 2007), screening for ASD does not occur for all children. These children typically arrive at a general, early childhood practice due to non-specific concerns about language delays or "sensory issues." In such cases, it may be useful to employ secondary screening measures as a method for triaging cases and informing the selection of assessment procedures and referrals (please see the article related to screening in this special issue for more information).

Developmental Assessment

In order to understand the child within a developmental framework, it is recommended that practitioners begin evaluations with a comprehensive examination of a child's overall development, including cognition, motor skills, and a survey of language abilities. Approximately 60–70% of children with ASD suffer from cognitive impairments (Fombonne 2005), and their cognitive profiles are often marked by presence of deficits as well as strengths (Joseph et al. 2002; Tsatsanis 2005). Thus, it is essential to evaluate profiles of developmental skills in young children with ASD not only for diagnostic purposes but also for designing individualized treatment programs that address their deficits and capitalize on their strengths (Chawarska and Bearss 2008). Similar to older children with ASD, a majority of toddlers presenting for differential diagnosis evidence delays in one or more developmental domains. For instance, Akshoomoff (2006) found that almost 75% of toddlers with ASD (mean age of 30 months) presented with significant delays in at least one domain assessed by the Mullen Scales of Early Learning (Mullen 1995). Chawarska et al. (2007) reported that although less than 20% of infants diagnosed with autism prior to the second birthday had significant delays (2 standard deviations below the mean) in nonverbal functioning on the Mullen Scales, almost 70% had significant delays in expressive and receptive language skills. In comparison, those later diagnosed with PDD-NOS tended to have relatively spared nonverbal skills, but more than 40% had verbal scores in the very low range.

While a majority of toddlers with ASD present with delays across multiple areas of development (e.g., non-verbal problem solving skills, motor skills, as well as receptive and expressive language skills), their developmental profiles are often uneven with some domains of development being more affected than others (Chawarska et al. 2009). In toddlers with autism, verbal skills are often more significantly delayed than nonverbal skills (e.g., Akshoomoff 2006; Carter et al. 2007; Chawarska et al. 2009; Charman et al. 1997, 2003; Eaves and Ho 2004; Landa and Garrett-Mayer 2006; Wetherby et al. 2004), and deficits in their understanding of and responsivity to language are typically more pronounced than delays in production of speech-like vocalizations and verbalizations (Paul et al. 2007; Chawarska et al. 2009). This type of profile is particularly prevalent in toddlers with autism where developmental delays and the discrepancy between verbal and nonverbal

functioning is more dramatic than in those with a milder form of the disorder, PDD-NOS (Chawarska et al. 2009).

In summary, studies of developmental profiles of toddlers diagnosed with autism converge to suggest that a prototypical profile consists of relatively spared nonverbal skills (e.g., visual discrimination and memory, visual-motor coordination) and more significantly affected verbal skills. In a large proportion of toddlers with autism, the ability to respond to speech (orient to speech sounds, recognize elementary words, follow simple directions) is more delayed than their ability to produce speech sounds and words. Developmental profiles of children with a milder form of ASD (i.e., PDD-NOS) might reveal lesser impairment as they tend to have better developed language skills and lower levels of developmental delay (e.g., Chawarska et al. 2009).

The rate of progress over time is highly variable (Bryson et al. 2007; Chawarska et al. 2009), with some toddlers making very slow progress and others evidencing remarkable gains over time. Thus, while mild delays at the time of diagnosis constitute a good prognostic sign, the presence of significant delays around the child's second birthday does not necessarily mean that the child will continue to have marked delays later on. The sources of such heterogeneity in presentation and rate of progress is not known, but research on identifying factors that are responsible for such diversity of syndrome expression is underway. Therefore, it is difficult for the clinician to make predictions regarding the child's long-term developmental potential based upon these early results. In addition, while there appear to be emerging developmental profiles which may correspond to range of severity within the autism spectrum, further research is needed before clinicians can utilize this information to aid in prediction of autism severity. What is clear, however, is that clinicians can anticipate that infants and toddlers presenting with ASD will likely present with developmental profiles marked by significant delays in receptive and expressive language, as well as possible delays in nonverbal skills.

To assess overall developmental level, there are a variety of tools available to the clinician. These tools vary in terms of the time required to administer the test, the domains of development that are measured, and the ratio of direct assessment, behavioral observation, and parental report associated with each measure. Most commonly seen in the literature for young children with ASD are the Mullen Scales of Early Learning (Mullen 1995) and the Bayley Scales of Infant and Toddler Development, Third Edition (Bayley-III; Bayley 2005). Some clinicians may also choose the Battelle Developmental Inventory, Second Edition (BDI-2; Newborg 2005) which involves more parental report and observation, whereas the Bayley and Mullen both rely heavily on direct assessment. In addition, clinicians looking for an assessment that covers all domains of development as mandated by IDEA may also utilize the Developmental Assessment of Young Children (DAYC; Voress and Maddox 1998) or the Bayley, as these include not only cognition, language, and motor, but also adaptive and social-emotional development scales. The Peabody Developmental Motor Scales (PDMS-2; Folio and Fewell 2000), often administered by the occupational or physical therapist on the team, can also be utilized for a more detailed view of the child's motor capabilities. For a further description of these measures please refer to Table 1.

Additionally, it is important to note that structured developmental assessment can be challenging given that many children with autism present with decreased attention to tasks, poor imitation skills, and difficulty following the lead of an interactive partner (for further discussion of assessment challenges, see Chawarska and Bearss 2008). However, even toddlers with the most challenging behavioral features (e.g., inattention, stereotyped behaviors, extreme anxiety, arousal dysregulation) are capable of completing a standard assessment given appropriate modifications to accommodate their attentional and learning style as well as motivational factors. Unlike typically developing children or children with other developmental concerns, children with autism are not often motivated by their desire to please the examiner; thus, practitioners have to offset the potentially negative effects of decreased social motivation and other testing difficulties by allotting more time for testing, utilizing a testing room that is small and void of distracting materials, carefully planning the child's seating and orientation in the room, facilitating transitions between tasks (often interspersing preferred tasks such that motivation to continue participation does not wane), and using extraneous reinforcement to promote good test-taking behavior such as attention, compliance, and attempts to manipulate the presented objects (but not specifically for task success as this would violate standardization of most developmental assessments). Also, it is often helpful to include parents in the assessment process by soliciting their advice regarding what might motivate their child or what aspects of the testing they might find aversive (e.g., loud sounds associated with deflating a balloon). Furthermore, parents can provide valuable feedback on the representative nature of the child's performance during the assessment, which is essential for diagnostic purposes and for treatment planning.

Social Interaction Skills

Subsequent to assessing overall development, the clinician now has a context in which to understand the child's use of social interaction skills and make the determination if these skills are significantly delayed relative to other areas of development (providing more evidence for an ASD), or are on par with other developmental skills (perhaps indicating a diagnosis of global developmental delay). In many children, symptoms of ASD reach clinical levels around or before their second birthday (Chawarska et al. 2007, 2009). The most consistently reported symptom at this age is the limited or declining interest in the social world. Toddlers with ASD show limited interest in purely social face-to-face interactions, seek physical comfort with parents infrequently, and make fewer initiations and responses to bids for social interaction from others (Garon et al. 2008; Landa et al. 2007; Mitchell et al. 2006; Wetherby et al. 2004; Yoder et al. 2009). Specific deficits in responsivity to social bids include the lack of a social smile (i.e., smiling in response to another person's smile) and limited response to name (Nadig et al. 2007). Additionally, toddlers with ASD spend less time monitoring others' faces and facial gestures including eye contact, affective expressions, and gaze cues (Bryson et al. 2007; Chawarska et al. 2007; Wetherby et al. 2004, 2007) as well as less time monitoring other people's activities (Shic et al. 2011). These children also show atypical affective responses, with a specific reduction in the expression of positive affect (Bryson et al. 2007; Garon et al. 2008; Wetherby et al. 2004). Eye contact is often fleeting and not well meshed with the flow of social and communicative exchanges (Chawarska et al. 2007), and children appear to have limited

interest in social games and reciprocal face-to-face interactions, unless they are accompanied by a physical component such as tickling, rocking, tossing in the air, etc.

The most frequently used assessment instrument of social interaction delays and abnormalities in ASD is the Autism Diagnostic Observation Schedule-Generic (ADOS-G; Lord et al. 2000). Although the ADOS-G is highly effective in identifying children with prototypical, or severe cases of autism, the measure has several limitations when applied to very young children (Bishop and Norbury 2002; Chawarska et al. 2007; deBildt et al. 2004; Lord et al. 2000). Specifically, when employed in children under the age of 2, the ADOS-G might over-diagnose children with low levels of functioning (i.e., a child with global developmental delay might score in the range indicating high likelihood of ASD on the ADOS-G), and under-diagnose higher functioning and verbal children with milder forms of ASD, such as PDD-NOS (Chawarska et al. 2007, Lord et al. 2000). To address these concerns, new algorithms have been developed to improve sensitivity and specificity of the instrument by dividing participants into smaller, more homogeneous groups (Gotham et al. 2007, 2008). These revisions include separate scoring algorithms in Module 1 for children who use words and those who do not, and separate algorithms in Module 2 for children who are under and over 5 years of age. The new algorithms have been found to increase ADOS-G sensitivity to milder forms of ASD in young nonverbal children as well as older children who have not developed beyond phrase speech (Gotham et al. 2007).

To further improve sensitivity and specificity of the ADOS-G as a diagnostic instrument for very young children, the creators of the ADOS-G developed a new module for younger children, the ADOS-Toddler (ADOS-T; Luyster et al. 2009). The ADOS-T is designed for children under the age of 30 months who have non-verbal mental ages of at least 12 months and motor skills that allow them to at least cruise their environment. The Toddler module follows the same structure as other modules of the ADOS-G, in which the examiner presents semi-structured and motivating activities for the child and observes the child's responses as well as attempts to maintain the interaction. The ADOS-T module was enriched with several new toddler-appropriate activities, and the scoring system was modified to better capture performance of children between 12 and 30 months. Two diagnostic algorithms have been derived for the ADOS-T: (1) for children between 12 and 20 months and nonverbal children who are 21–30 months, and (2) for verbal children between 21 and 30 months. Scores on the ADOS-T fall into categories reflecting overall level of concern regarding the likelihood that the child has ASD: little or no concern, mild or moderate, moderate or severe. The ADOS-T demonstrated very good sensitivity and specificity in the validation sample (Luyster et al. 2009). The ADOS-T is currently only available for research purposes, but it is anticipated to be published in 2012 as part of the new ADOS-II.

Given few time constraints, the clinician may elect to use a common companion to the ADOS-G, the Autism Diagnostic Interview-Revised (ADI-R; Rutter et al. 2003). However, the validity of the ADI-R in younger children has been problematic (Chawarska et al. 2007; Rutter et al. 2003). The ADI-R tends to over-diagnose nonverbal children with mental ages below 18 months, and under-diagnose verbal high-functioning children (Chawarska et al. 2007; Cox et al. 1999; Lord 1995; Rutter et al. 2003). A study of the ADI-R in 16–30 month-old toddlers indicated that nearly 90% of the children determined to have an ASD by

clinical expert did not meet the ADI-R diagnostic cutoff due to too few restricted and repetitive behaviors (Ventola et al. 2006). Lowering or eliminating the cutoff for restricted and repetitive behaviors on the ADI-R may help to improve its utility in young children (Chawarska et al. 2007; Wiggins and Robins 2008), though further work is needed to fully adapt the instrument for children under the age of 3 (Bishop et al. 2008). The work on development of a toddler version of the ADI, with corresponding modified algorithms, is currently underway (Kim and Lord 2009), although these are not yet publicly available. It must be noted that the developers of the ADOS-G and the ADI-R have training guidelines that are encouraged to be met prior to the use of these instruments in clinical practice, and further training is required for research use. For clinical use of the ADOS-G, the developers suggest a 2-day training session, and for the ADI-R, home-based training materials are available.

If the clinician does not elect to use the ADI-R, they are strongly encouraged to use other sources of parental report to corroborate observations of social interaction within the context of the assessment. These include clinical interview focused on a comprehensive developmental history, social-communication domains from adaptive behavior measures that rely on parental report (e.g., Vineland-II), or developmental assessment that includes caregiver report of social-emotional development and adaptive functioning associated with social-communication skills (e.g., Bayley-III, DAYC).

Communication

While delays in development of vocalizations may be apparent very early in infants with autism (Paul et al. 2011), by the child's second birthday, the deficits in the ability to understand gestures and verbal communication of others, as well as to initiate communicative acts, becomes very pronounced. The ability to understand and to respond to language is often more impaired than the ability to produce speech sounds (Chawarska et al. 2009; Ellis et al. 2010; Hudry et al. 2010). Significant delays in the development of expressive language are also consistently seen in children with ASD by the age of 2 years (Chawarska et al. 2007; Chawarska and Volkmar 2005; Paul et al. 2008). Toddlers with ASD direct their vocalizations to others less frequently and also show atypical intonation of their speech (Brian et al. 2008). They also produce fewer consonants as well as word combinations, and produce a reduced inventory of words overall (Landa and Garrett-Mayer 2006; Landa et al. 2007; Paul et al. 2011). It is notable that children with ASD do not attempt to compensate for lack of spoken language with the use of gaze and conventional gestures as is often seen in children whose language delays are not associated with social disability. Thus, gestures such as pointing and showing or giving objects to others for the purpose of sharing interests or making requests are either absent or very infrequent (Chawarska et al. 2007; Chiang et al. 2008). As language begins to emerge in the second and third year, the unusual linguistic features, including echolalia (i.e., verbatim repetition of utterances of others) and scripting (i.e., repetition of phrases previously heard from books, movies, or other sources), and abnormal pitch or intonation become apparent (Oller et al. 2010; Schoen et al. 2010).

Although overall developmental measures typically provide general information regarding a child's receptive and expressive language, more detailed information regarding a child's verbal and nonverbal communication development is often useful for diagnostic purposes and treatment planning. Thus, the inclusion of a speech-language pathologist on the assessment team is recommended. For very young children, clinicians may use the Communication and Symbolic Behavior Scales Developmental Profile (CSBS-DP; Wetherby and Prizant 2002) as a measure focused on both nonverbal and verbal communication. Additionally, the Rossetti Infant-Toddler Language Scale (Rossetti 1990) and the new edition of the Preschool Language Scales (PLS-5; Zimmerman et al. 2011) may be used, as they are both appropriate for infants and toddlers. The clinician may also take a language sample and utilize structured and unstructured observation to provide a more thorough picture of the child's communication skills. Parental report can also be used to get a broader sense of the child's abilities, and measures such as the MacArthur-Bates Communication Development Inventory (CDI; Fenson et al. 2007) can be used for this purpose. Moreover, the ADOS-G also provides a wealth of information regarding the child's functional use of verbal and nonverbal communicative skills in more naturalistic settings. Thus, clinicians can select from a number of instruments to create a complete picture of the child's communication abilities.

Restricted Interests and Repetitive Behaviors

As with other domains, the assessment of restricted interests and repetitive behaviors requires knowledge of the typical course of development, as well as in-depth understanding of developmental abnormalities commonly associated with other conditions. Although motor mannerisms are often considered a hallmark sign of ASD, rhythmical and repetitive behaviors commonly occur in early infancy (Thelen 1979). Beyond infancy, notable motor movements can also occur. For example, toe-walking is often observed in infants who are learning to walk, and such behavior can persist into the toddler years. As a clinician, it is important to put these behaviors into context (Pernet et al. 2010). To continue with this example, if a child is toe-walking and has no other signs of developmental abnormality or delay, the behavior is probably idiopathic. However, if the child also has poor muscle coordination, one might consider a cerebral palsy diagnosis. Alternatively, if the behavior is unusually frequent or has a slightly different topography, and communication and social deficits are also noted, the behavior may indeed point to an ASD diagnosis. Thus the clinician's job is not to simply assess the presence of these behaviors, but rather, their frequency, the context in which they occur, and their functional significance (i.e., the degree to which such behaviors begin to pull a child's focus away from more functional play and social exchanges).

By the second birthday, children with ASD typically demonstrate unusual sensory interests, hand and finger mannerisms, as well as repetitive body movements (Brian et al. 2008; Chawarska et al. 2009; Werner et al. 2005). Sensory interests might include atypical exploration of objects (e.g., rubbing items on face, mouthing or sniffing objects), aversive response to some textures, and an interest in non-play items (e.g., bolts in chair, shoelaces, string, parts of toys). Motor mannerisms may manifest in repetitive hand flapping or jumping, finger posturing, and toe-walking. A child might insist on opening and closing

doors in a repetitive manner, following strict bedtime or bath time routines, or arranging objects in a specific manner (e.g., lining up blocks or cars). Stereotyped behaviors might also be expressed in speech, including echolalia and scripted speech (e.g., reciting parts of movies or books, preservative labeling of objects or repetition of questions; Kim and Lord 2010). While in some children repetitive behaviors and interests reach clinical levels in the second year of life, in others, these behaviors while relatively mildly expressed in the second year, become more pronounced thereafter (Chawarska et al. 2007; Richler et al. 2007).

As previously discussed, a "waiting room-to-feedback meeting" approach wherein the child's restricted interests and repetitive behaviors are observed from the moment the child is greeted in the waiting room until the last interactions before the feedback meeting occurs is ideal. Restricted interests and repetitive behaviors may occur during times of transition, free time in the waiting room, as well as during assessments designed to pull for such behaviors (e.g., ADOS-G).

Furthermore, given that restricted interests and repetitive behaviors may be more subtle in infants and toddlers than those observed in older children, it is important to couple direct observation with parent report to ensure that the child's behaviors are accurately assessed and included in diagnostic formulation. Considering that it might be difficult for parents to judge typicality of certain classes of repetitive and exploratory behaviors, it is often useful to employ an approach that allows parents to describe their child's preferred activities (e.g., 'Tell me how Johnny likes to play or what he finds particularly interesting," or "Are there any activities that Johnny likes so much that you dread having to interrupt him?"), without evaluative components (e.g., "Tell me what Johnny does that appears unusual or odd."). Assessing frequency, intensity, and the level of interference associated with repetitive behaviors in young children necessitates parent interviewing, as some of these behaviors might not become apparent during direct assessment and yet they might reach debilitating levels at home and need to be addressed directly through behavioral intervention.

Clinical judgment is the practitioner's best tool in completing a parent interview about the presence or absence of these behaviors, because there are not many formal assessment tools on which clinicians can rely. As previously noted, the ADI-R, which probes for these behaviors, is not ideal for very young children, and while some behaviors may be observed during the ADOS-G, there is a possibility that the atypical behaviors in the child's repertoire are so subtle or context-specific that they are not observed during the clinical assessment. Other measures that clinicians may find useful include the Infant/Toddler Sensory Profile (Dunn 2002) and the Repetitive Behavior Scale-Revised (Bodfish et al. 2000; Lam and Aman 2007), although the later is most commonly used in research.

Play Skills

Impairments in play skills are well documented in children with ASD (Jarrold 2003). Toddlers with ASD show limited imagination and creativity in their play, as well as delays in motor imitation and functional use of toys (Brian et al. 2008; Charman et al. 1997; Chawarska et al. 2007). When given an opportunity to play with miniature toys (e.g., cars, dolls, animals, household items), toddlers with ASD often focus on their sensory qualities—how the toys feel, reflect light, or what kind of noise they make rather than on their function

(e.g., cup is for drinking, ball is for throwing). Their interest in toys is often fleeting and they might move from one object to another quickly. They often do not benefit from modeling and instruction due to deficits in imitation as well as symbolic representational deficits (Charman et al. 1997). When toddlers with ASD engage in play, their play is often repetitive and stereotyped (e.g., involves arranging toys in a certain way or replaying a play routine in the same way over and over again (e.g., feeding a doll). Moreover, while toddlers with ASD might be interested in toys and play with them for a long periods of time (e.g., pushing buttons to produce a sound), they direct their play acts less frequently towards others, making few attempts to bring others into their play (Landa et al. 2007). Play abnormalities can be observed during the administration of the ADOS-G, and given the myriad of manipulatives available during developmental, speech, and motor testing, play can also be observed in these contexts. Although there are a number of specific play assessments that are commercially available, if other measures noted throughout this guide are utilized, a separate evaluation of play skills should not be necessary in order to make a differential diagnosis of ASD.

Adaptive Behavior

When evaluating children for ASD, it is important to consider adaptive functioning, which reflects the child's capacity for personal and social self-sufficiency in real-life situations. For very young children, evaluation of adaptive behavior focuses on a myriad of skills related to the everyday use of appropriate expressive and receptive language, elementary social interaction and play, fundamental self-care such as dressing, bathing, and feeding, and participation in elementary chores (e.g., putting away toys). Studies of older children and adolescents with ASD have demonstrated a marked discrepancy between the level of cognitive functioning (IQ) and the actual level of adaptive functioning, with this discrepancy becoming more pronounced with age (Jacobson and Ackerman 1990; Lockyer and Rutter 1969; Lord and Schopler 1989; Perry et al. 2009; Saulnier and Klin 2007). Thus, older children with ASD have marked difficulties using their cognitive skills to function adaptively in their environment.

Empirical evidence suggests that the roots of this discrepancy can be found early in development and that the deficits in adaptive functioning in young children with ASD cannot be entirely explained by their developmental delays. Stone et al. (1999) reported that children with autism under the age of 3 years have weaker adaptive socialization and communication skills and show greater discrepancy between adaptive behavior and mental age than developmentally delayed controls matched for mental and chronological age. Similarly, Ventola et al. (2007) found that their ASD group (mean chronological age of 26.7 months) had significantly lower standard scores than the developmentally delayed and language disordered groups on the Vineland subtests of Communication, Daily Living, Socialization, and Motor Functioning. A recent study based on the assessment of 125 toddlers with ASD (mean chronological age of 31 months) suggests that delays in adaptive skills were evident by 2 years of age, and as a group, the toddlers with ASD had a profile of Vineland-II scores in which Motor Skills were higher than Daily Living Skills, which in turn, were higher than their Socialization scores, which exceeded scores on the Communication scale (Ray-Subramanian et al. 2011).

Although the empirical research evaluating adaptive behavior in infants and toddlers with ASD is still relatively limited, research to date has shown that, as a group, young children with ASD tend to demonstrate lower scores in the Socialization and Communication domains compared to chronological and mental-age-matched, non-spectrum children with intellectual disabilities. While an individual's adaptive behavior profile should not be used as the sole basis for diagnosis, the identification of a characteristic group profile may aid the process of differential diagnosis.

When assessing adaptive behavior in infants and toddlers, the Vineland Adaptive Behavior Scales-Second Edition (VABS-II; Sparrow et al. 2005), is often considered the gold standard, and it is the measure that is most commonly used in adaptive behavior research for young children with ASD. However, practitioners should be aware that there are other options available to them, including the Scales of Independent Behavior (SIB-R; Bruininks et al. 1996) and the Adaptive Behavior Assessment System-Second Edition (ABAS-II; Harrison and Oakland 2003). Additionally, for those clinicians who are practicing in offices where assessment time is limited, or these measures are simply not available, there are several assessments of overall development that include specific adaptive behavior subscales, including the Bayley (2005), the BDI-2 (Newborg 2005), and the DAYC (Voress and Maddox 1998). For additional information about these measures, see Table 1.

Recommendations and Early Intervention

Besides providing diagnostic clarification, one of the main goals of a comprehensive clinical evaluation is to provide detailed recommendations to both parents and clinicians regarding optimal strategies for facilitating the child's development and optimizing short- and long-term outcome. Numerous studies report on the effectiveness of early intervention in preschool children with ASD (e.g., Cohen et al. 2006; Dawson and Osterling 1997; Harris and Handleman 2000; Harris and Weiss 1998; Koegel et al. 2006; McEachin et al. 1995; Sheinkopf and Siegel 1998; Volkmar et al. 2004). Current best practices suggest that programs should begin early (i.e., as soon as the child is diagnosed), be individualized to the child's needs, be intensive (i.e., a minimum of 25 h per week), be comprehensive (i.e., addressing various areas of developmental need), include parent education and training, and be databased (i.e., program development and modifications being based on ongoing assessment and data collection) (NRC 2001; Iovannone et al. 2003; Woods and Wetherby 2003).

Researchers have suggested that intervention strategies designed for preschoolers may be effectively downward extended to infants and toddlers (Rogers 2009; Zwaigenbaum et al. 2009), although there are currently relatively few published studies of early intervention for very young children with ASD. Wetherby and Woods (2006) demonstrated significant improvements on standardized developmental measures of IQ and social communication for children approximately 24 months of age after 1 year participation in the Early Social Interaction (ESI) project, an intervention utilizing naturalistic teaching strategies in a parent mediated intervention format. The Walden Toddler Program (McGee et al. 1999) for toddlers 15–30 months of age also reported improvements in frequency of meaningful verbalizations after 1 year of an intensive, center-based intervention employing incidental

teaching. In the first randomized, controlled intervention trial for toddlers, Dawson et al. (2010) employed the Early Start Denver Model (ESDM), a program drawing on naturalistic behavioral and developmental strategies with children between 18 and 30 months of age. The study demonstrated significant improvement on standardized developmental measures after 1 and 2 years of intensive ESDM treatment in the treatment group compared to the treatment-as-usual (community-based) group. In addition to ASD-specific studies, evidence from intervention studies targeting communication and cognitive skills in infants with other, non-autistic disorders may also be highly informative in the development of interventions appropriate for infants and toddlers with ASD (Wallace and Rogers 2010), although such studies have not yet been completed. For the practitioner, it is encouraging to know that emerging research does support the use of early intervention procedures, and that by and large, procedures designed for preschools with ASD can be successfully downward extended to the infant and toddler population, thus appropriate intervention programs may be available with referral to local service providers.

Conclusions

Increasing prevalence of ASD, expanding awareness of early symptoms of developmental disabilities amongst parents and professionals, as well as close monitoring of infants at genetic risk for ASD, has led to a sharp increase in the number of children under the age of 3 years presenting for differential diagnosis. This situation calls for an immediate and comprehensive response from health care professionals and educators. Empirical evidence suggests that ASD can be diagnosed reliably, by well-trained clinicians, as early as the second year of life and that children benefit greatly from intensive and comprehensive early intervention services. Responding to the needs of children diagnosed with ASD, as well as the needs of their families, fulfills practitioners' fundamental role of improving the quality of life of those affected by this complex disorder. Research on phenotypic expression and etiology of autism have been advancing rapidly in the past years and, as a field, we are now very well positioned to translate this knowledge into diagnostic and treatment practices that may alleviate or perhaps someday eliminate the often devastating symptoms of ASD. As a practitioner, the early identification of infants and toddlers with ASD is crucial to this mission, and while certainly a complex process, there is currently substantial support in the literature to guide the diagnostic process and great momentum in the field to accomplish this task.

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Table 1

Measures for assessing autism spectrum disorders in infants and toddlers

	Measure	Acronym	Age range	Format/time	Brief description
ASD assessment	Autism Diagnostic Observation Schedule (Lord et al. 2002)	ADOS	Toddlers to adults	Play-based direct assessment; 30–45 min	Assessment divided into 4 modules based on language level. The assessment consists of standardized presses to elicit certain types of social, communicative, and play behaviors. A subset of items constitutes the diagnostic algorithm, for which cut-off scores determine “autism” or “ASD” classification. 5th module (Toddler Module) will be included in the ADOS II
	Autism Diagnostic Interview—Revised (Rutter et al. 2003)	ADI-R	Children and adults with mental age above 2 years	Parent interview; 1.5–2.5 h	Semi-structured parent interview designed to evaluate the child’s functioning in social, communication, and restricted and repetitive behavior domains. Useful for diagnosing ASD and distinguishing ASD from other developmental disorders
Adaptive behavior	Vineland Adaptive Behavior Scales II (Sparrow et al. 2005)	VABS II Vineland-II	Birth to 90 years	Interview or parent rating form; 20–90 min	Broad measure of adaptive functioning. Scales organized within a three domain structure: Communication, Daily Living, and Socialization, which corresponds to domains of functioning outlined by AAIDD. VABS II also assesses Motor Skills
	Scales of Independent Behavior—Revised (Bruininks et al. 1996)	SIB-R	Infancy to 80 + years	Interview or parent rating form; 15–60 min	Assessment of 14 areas of adaptive and 8 areas of maladaptive behaviors. Adaptive behavior areas are grouped into 4 clusters: motor skills, social interaction and communication skills, personal living skills, and community living skills
	Adaptive Behavior Assessment System—2nd edn. (Harrison and Oakland 2003)	ABAS-II	Birth to 89 years	Parent, teacher, and caregiver rating forms; 15–20 min	Assessment of three general areas of adaptive behavior (conceptual, social, practical), as well as 10 specific adaptive skill areas specified in the DSM-IV. Two of five rating forms are appropriate for early assessment: Parent/Primary Caregiver Form (0–5 years) and Teacher/Daycare Provider Form (2–5 years)
Motor	Peabody Developmental Motor Scales—2nd edn. (Folio and Fewell 2000)	PDMS-2	Birth to 5 years	Direct assessment; 45–60 min	Assesses both gross and fine motor development; recommends specific interventions
Overall development	Mullen Scales of Early Learning (Mullen 1995)	MSEL	Birth to 5:8	Direct assessment; 15–60 min	Developmental battery that measures expressive and receptive language, gross and fine motor, and visual reception skills
	Battelle Developmental Inventory, 2nd edn. (Newborg 2005)	BDI-2	Birth to 7:11	Structured, play-based activities, observation, and parent/caregiver interview; Full: 60–90 min, Screen: 10–30 min	Screens and evaluates early childhood developmental milestones in the domains of Personal-Social, Adaptive, Motor, Communication, and Cognitive
	Bayley Scales of Infant and Toddler Development, 3rd edn. (Bayley 2005)	Bayley-III	1–42 months	Direct assessment and parent questionnaires; 30–90 min	Evaluates cognitive, language, motor via direct assessment, and evaluates adaptive behavior and social-emotional functioning via parent questionnaires
	Developmental Assessment of Young Children (Voress and Maddox 1998)	DAYC	Birth to 5:11	Direct assessment, direct observation, and parent questionnaire; 60 min	Identifies delays in the domains of cognition, communication, social-emotional development, physical development, and adaptive behavior
Communication	Communication and Symbolic Behavior Scales Developmental	CSBS-DP	6 months to 6 years	Direct assessment and parent questionnaire; 60 min	Measures seven language predictors: emotion and eye gaze, communication, gestures, sounds, words, understanding, and object use. Can be used to screen children to determine delays in

	Measure	Acronym	Age range	Format/time	Brief description
	Profile (Wetherby and Prizant 2002)				social communication, expressive speech/language, and symbolic functioning, and to evaluate changes over time
	MacArthur-Bates Communicative Development Inventories, 2nd edn. (Fenson et al. 2007)	CDIs	8–37 months	Parent report inventory of words and gestures; 5–10 min	Parent report of child's understanding and use of early vocabulary; child's understanding of word forms and grammar use
	Preschool Language Scales-5 (Zimmerman et al. 2011)	PLS-5	Birth to 7:11	Direct assessment, direct observation, and parent questionnaire; 30–60 min	Measures receptive and expressive language, with supplemental screener for articulation, language sample, and home communication questionnaire
	Rossetti Infant-Toddler Language Scale (Rossetti 1990)	Rossetti	Birth to 3 years	Direct assessment, observation, and parent questionnaire; < 45 min	Assesses preverbal and verbal communication, including interaction-attachment, pragmatics, gestures, play, language comprehension, and language expression

Most questionnaires can be administered in interview format. Times vary depending on the form used and the age of the child