Diagnostic Evaluation of Autism Spectrum Disorders

Marisela Huerta, PhD and Catherine Lord, PhD

Synopsis

Research on the identification and evaluation of autism spectrum disorders (ASD) is reviewed and best practices for clinical work are discussed. The latest research on diagnostic tools, and their recommended use, is also reviewed. Recommendations include the use of instruments designed to assess multiple domains of functioning and behavior, the inclusion of parents and caregivers as active partners, and the consideration of developmental factors throughout the diagnostic process.

Keywords

Autism spectrum disorder; diagnosis; assessment; diagnostic instruments

“Michael is a 9-year old male with a history of language delays. Currently, his expressive language seems to be at age level. However, he demonstrates difficulty following instructions at home and at school. Most notably, his peer interactions are poor. Though Michael is interested in his peers, he does not have friends. Recently, he has been struggling academically as well.”

“Olivia is a 5-year old female who is described as ‘precocious’. Her parents are most concerned about her increasing physical aggression. She hits and bites others. Olivia is also quite particular about how the activities in her day are conducted. Notably, Olivia does not play imaginatively with other children though she loves to watch and imitate her favorite videos.”

“Tony is a 2-year old boy who is often fearful. He demonstrates anxiety in new environments and often clings to his parents. Tony’s parents also expressed a concern about his lack of language. Tony uses a limited number of single words, but often, he does not speak at all. It is not clear how much speech Tony understands at this time.”

-adapted from clinical intake notes
Introduction

As illustrated in the examples above, referrals to autism specialty clinics frequently span the full spectrum of the disorder and often involve diverse symptom presentations. Such heterogeneity in autism spectrum disorders (ASD) makes identification of the disorder a complex process. How are physicians to contend with this challenge in diagnosing ASD? Given such variability in symptom expression, what makes for a reasonable ASD evaluation?

The past decade has seen many advances in the availability of standardized ASD testing tools as well as increased knowledge about the variability in symptom expression. In addition, researchers have learned much about caregivers’ varied experiences with the referral and diagnostic process that can inform clinical work. This chapter describes these insights with the goal of highlighting best practices for evaluating ASD.

Challenges in ASD Diagnostic Assessment

The American Academy of Pediatrics\(^1\) and the American Psychological Association\(^2\) have recommended an approach to the identification of ASD that involves step-wise, and at times, recursive surveillance. Starting at pediatric (e.g.; “well baby”) appointments, the approach calls for formal screening if behaviors of concern are noted during surveillance (see Patel’s review of screening recommendations in this issue). If screening, including any caregiver concern, indicates cause for attention, this is to be followed by formal diagnostic assessment. In the years following the publication of these guidelines, the identification of ASD appears to have improved somewhat.\(^3\) However, many children continue to be first identified by their educational programs\(^4,5\), and a significant minority of children with ASD are likely to be undiagnosed.\(^4,6\)

A contributing factor to the problem of under-identification likely results from the variability of symptom expression in ASD. As we have learned, the presentation of ASD can range from a child who is nonverbal and unlikely to make social initiations, to a child who is verbally fluent, but overly reliant on previously learned scripts of speech and social behavior (Ghaziuddin provides a detailed review of the clinical features of ASD in this issue). Because of this variability in symptom type and severity, diagnostic decision-making is a complex process; no singular algorithm can be applied to the diagnosis of ASD. In a clinical sense this has meant that no one behavior, such as responding to name or joint attention, excludes a diagnosis of ASD.\(^7\) For example, though responding to his or her name and responding to joint attention are important characteristics of ASD in toddlers, most children with ASD can carry out both of these actions by older preschool.\(^8\) Even as a toddler, a very intelligent child who understands his name and follows a point may still merit a diagnosis of ASD because he or she does not seek to share enjoyment with others, smile back to people except during intense physical activity, show any interest in his or her siblings or same-age peers and use language to answer questions or make socially-directed comments.

Diagnosis in ASD can be difficult because behaviors seen in a child are often dependent on a number of non-autism-specific factors, including cognitive functioning and age.\(^9,10,11\) The diagnosis of ASD is further complicated because of the interactions that occur between development and ASD symptoms. At certain ages, a number of characteristics, especially when defined by informants, that are common to ASD are not actually specific to the

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\(^{a}\)Given that the current categorical classification system that makes distinctions between autism, Asperger Disorder, and Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS) is highly unreliable (Lord et al., in press) we use the term, autism spectrum disorders (ASD) to refer to these disorders collectively. This is the term currently used in the proposed Diagnostic and Statistical Manual of Mental Disorders (DSM-5; see www.dsm5.org).

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diagnosis and may occur in other disorders. In one study, as reported by parents and caregivers, stereotyped language was no more prevalent in children with ASD than in typically-developing children and children with other non-spectrum diagnoses who were under age 4 and in children who did not have complex language. These findings indicate that the types of behaviors to which a clinician must attend to for diagnosis are very much dependent on developmental factors such as age and language level, as well as the source of information (i.e., caregiver report through interview, questionnaire or clinician observation).

To address these challenges, the National Research Council Committee on Educational Interventions for Children with Autism advised that each child suspected of having ASD have an evaluation that incorporates the following standards: the assessment of multiple areas of functioning including adaptive skills, an appreciation that variability in performance and ability is common in autism, and the use of a developmental perspective when assessing behavior and synthesizing results. This is typically beyond the scope of usual pediatric practice and so, most cases will depend on appropriate referral. A wealth of recent studies, particularly those dedicated to the careful phenotyping of children who span the full spectrum of autism, have not only yielded data in support of these practices, they have also informed clinical practice with new methods and tools to implement these recommendations. These findings, and the practical implications of the guidelines, are discussed below.

**Best Practices in ASD Diagnostic Evaluations**

**The Diagnostic Process**

The inclusion of standardized cognitive and developmental testing, as well as an assessment of language, is particularly important to differentiate ASD from other developmental difficulties. These tests, which if carefully selected and carried out by professionals experienced in assessment of children with developmental disabilities, should provide information about the child’s overall level of ability and functioning in both verbal and nonverbal areas, thereby providing a crucial starting point for the clinician to make a best estimate of a clinical diagnosis. The separation of verbal and nonverbal estimates of functioning is particularly important because many children with ASD show much stronger nonverbal skills than one might expect from their language level or play. Standardized scores from a skilled examiner allow the clinician to consider such questions as: can intellectual disability and/or language delay explain the difficulties in social interaction? Given this level of ability, are observed difficulties in communication and social behavior above and beyond what would be expected? Does the level of social ability fall short of what is expected given cognitive or language ability?

The term “multidisciplinary” has often been used to describe best practice diagnostic evaluations. At some clinics this has meant that an autism evaluation is performed by multi-member diagnostic teams. However, this is not always feasible, and in some cases can be overwhelming for parents. Instead, “multidisciplinary” should be interpreted to mean that multiple areas of functioning should be considered during a diagnostic evaluation. Though a physician should always be available to provide a medical evaluation, it is most important that these multi-faceted evaluations be completed by clinicians with extensive experience in the standardized testing of children with particular expertise in assessment of ASD.

In addition to covering multiple domains, diagnostic evaluations should consider information from multiple sources. A comprehensive evaluation must, at minimum, include a parent interview and an observational assessment of the child’s current functioning by an experienced clinician in a context in which social-communicative behavior and play or peer interaction can be observed. For the diagnosis of ASD, diagnostic specificity is much worse
if only one or the other is performed. The clinician’s observation provides the opportunity to put the child’s behavior into the context of knowledge about other children, but information from caregivers provides a broader context needed in understanding the child’s day-to-day behavior in a wide range of situations, his or her history, as well as family expectations, resources and experiences and other important contextual factors. Thus, child testing and parent interviews should be viewed as complimentary and necessary components of the diagnostic evaluation. (Figure 1)

Ultimately, it is the job of the clinician to integrate information across the various sources of information. No single result (e.g., a borderline score on one instrument; a high score on another) is sufficient. A clinician can take into account that a caregiver was very uncomfortable saying anything negative about his or her child or, as a new parent, unfamiliar with typical development, or that a child was so hyperactive or tired or shy that his or her behavior in the office was not likely representative of other times. In some cases, diagnostic clarity is not easily obtained despite access to formal testing results. Research on the early diagnosis of ASD suggests that for diagnostically complex cases, best practices are to adopt a surveillance approach that is recursive, meaning repeated screening and assessment. Such a strategy may be particularly appropriate for very young children when formal estimates of functioning may not always remain stable, for children with very difficult behaviors (e.g., aggression, self-injury) where there are very pressing day-to-day concerns and for children with the very mildest difficulties where diagnosis may be less accurate.

When best-practice evaluation results in a diagnosis of ASD, the referring physician should expect to receive specific details about the child’s functioning that supports the diagnosis from the evaluator. The evaluation report should explain why this child does or does not have autism. In addition, the report should include recommendations that are specific to the child’s particular constellation of difficulties and the contexts in which they occur. Other important questions that should be answered by a diagnostic evaluation are found in Box 1.

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**Box 1**

**Key questions for ASD Diagnostic Evaluations**

1. What diagnostic classification best describes this child’s pattern of behaviors and difficulties? If no diagnosis is confirmed, how often should this child continue to be monitored?

2. What is this child’s overall level of functioning? Is cognitive functioning, language ability, or learning impaired? Or are results suspect?

3. Are there additional behaviors and/or diagnoses to consider for this child when planning treatment?

4. What behaviors/symptoms are most concerning to the family/caregivers? What role would they like to play in the child’s treatment?

5. Given the current constellation of symptoms, what are the most important targets for intervention at this time?

6. Given family circumstances, community resources, and other contextual factors, what recommendations are most appropriate?

7. What is the prognosis for this child in the short-term? Is there enough information to make predictions about prognosis over the long-term?
Referrals for formal testing should not be limited to diagnostic evaluations. Children who have been diagnosed with ASD will require re-evaluations periodically, particularly at times of transition (e.g., prior to the start of primary school). A child who receives a diagnosis of ASD at age 2 or 3 should receive testing the following year, sooner if any test results were suspect. Reevaluations need not be as comprehensive as the initial diagnostic testing, but they are important; over time, children with ASD change, not so much in diagnosis, but in their needs, strengths, and difficulties.  

**Diagnostic Instruments for ASD**

A quick review of the current literature on ASD reveals an abundance of available diagnostic instruments. It may be difficult for clinicians to identify gold-standard instruments, but this becomes easier if one remembers the key elements set forth by the National Research Council Committee on Educational Interventions for Children with Autism (2001): best practice diagnostic tools should assess social functioning in a developmental context and they should take into account the variability of behavior across settings. Following these criteria, a useful child testing tool in autism evaluations continues to be the Autism Diagnostic Observational Schedule. Recent revisions to the ADOS have improved its accuracy and expanded its clinical utility to include an indicator of severity. Another development has been the toddler version of the ADOS, which employs a surveillance model of classification for very young children. The Screening Tool for Autism in Toddlers and Young Children (STAT) and the Communication and Symbolic Behavior Scales (CSBS) are also recommended for use in young children. A particular strength of all of these instruments lies in the fact that they are standardized in their administration and coding, ensuring careful assessment of something as complex as social behavior.

For the parent interview, the ADI-R has been established as a useful diagnostic tool in the assessment of ASD. For use with very young children, toddler versions of the ADI-R diagnostic algorithm were recently created. Importantly, though the ADI-R is quite useful for diagnosis, it is a lengthy instrument that requires significant training. Alternative methods of collecting parent report are needed. Currently, the use of the Social Communication Questionnaire (SCQ) in conjunction with the ADOS appears to be a reasonable replacement when it is not possible to do an ADI.

An emerging area of research involves validating ASD diagnostic instruments that have been translated into a number of different languages. However, the cultural equivalence of these translated instruments has not yet been established. In fact, for some instruments, there is emerging evidence that certain items function differently for non-English speaking parents. For example, the restricted and repetitive behaviors on the ADI-R tend to be underendorsed by Spanish-speaking caregivers compared to their English-speaking counterparts.

**The Importance of Diagnostic Evaluations**

Though time-intensive, and often not adequately funded, physicians must continue to refer children for formal diagnostic evaluations when ASD is suspected. Some school systems provide detailed assessments but many do not. In addition, school assessments, while including parents and caregivers as informants, are not typically focused on helping families understand their children’s difficulties and strengths and what families can do, but are necessarily oriented to educational services. Formal diagnostic testing is important for a number of reasons. First, although ASD very likely has genetic and neurobiological underpinnings, no accurate biologically-based test exists. As a result, the diagnosis of ASD is one based on behavior, or more accurately, based on information about patterns of behavior.
behavior and symptoms.\textsuperscript{28} Second, estimates of functioning in key domains can also provide important prognostic information about the course of autism symptoms. For example, nonverbal cognitive skills at age 2 predict verbal ability at age 5.\textsuperscript{29} Poor adaptive skills coupled with repetitive behaviors at 2 also predict more behavioral difficulties in older children with ASD; using any words at 2 and having more expressive language at 3 are predictors of more positive social outcomes.\textsuperscript{30}

Third, even when diagnostic certainty is high, formal diagnostic assessment can contribute to treatment planning. The results obtained from a diagnostic evaluation can shed light on the function of behaviors and identify both “near” and “eventual” treatment targets, thereby increasing the utility of interventions.\textsuperscript{31} This does not always happen as the result of diagnostic assessments, but if not, the pediatrician should consider alternative referrals.

A number of qualitative studies report that diagnostic evaluations also provide intangible benefits to the family of the child with ASD. Diagnostic confirmation, particularly for those with complex or atypical symptom presentations, often provides relief to the family.\textsuperscript{32} Further, formal testing is designed to provide additional information beyond diagnostic classification; a profile of a child’s impairments and strengths satisfies many parents and caregivers who are trying to make sense of their child’s behavior and ASD.\textsuperscript{5} Moreover, when asked about their experiences with clinical providers, parents and caregivers frequently express a desire to be active participants in the process. Their feedback suggests that clinical best practices should include open communication with families about the suspected diagnoses and the procedures involved in evaluations.\textsuperscript{33}

**Conclusion**

In summary, good diagnostic evaluations of ASD include the use of instruments designed to assess multiple domains of functioning and behavior, the inclusion of parents and caregivers as active partners, and the consideration of developmental factors throughout the diagnostic process. Physicians play a crucial role in ensuring best practices in autism evaluations, through initial identification, selection of a referral, and discussions with parents and caregivers about what to expect and how to get the most from a diagnostic evaluation. Pediatricians in particular, given their more frequent contact with children compared to most specialists, are in the best position to monitor the needs of children with ASD over the course of development and within the context of their overall physical health. Pediatricians can also serve as very important advocates within healthcare systems, with insurance companies and government agencies to ensure all families have access to appropriate diagnostic and treatment services.

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**References**


Figure 1.
Components of an ASD Evaluation

- **Parent/Caregiver Interview:**
  Medical/developmental history and report of current behavior and abilities (teacher report is also ideal, when possible)

- **Observational Assessment:**
  Medical evaluation, cognitive or developmental testing, language testing, and structured assessment of social interaction (for some children this may also include classroom observations)

- **Integration of information**

- **Diagnosis:**
  Diagnostic summary should include domain-specific information regarding level of functioning and severity