

# Diagnosis: screening, surveillance, assessment, and formulation

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### *Key Learning Objectives*

By the end of this chapter, readers will be able to:

- identify necessary team members and define team member roles for the diagnostic assessment of autism spectrum disorder (ASD);
- recognize how the caregiver history, history from other sources, physical examination, informal observation, and formal observation contribute to the diagnostic assessment;
- conceptualize and integrate the assessment findings into a formulation that addresses the diagnostic question in the context of the child's neurodevelopmental profile; and
- communicate the findings of the assessment effectively with families, providing an informative summary and interpretation.

## Assessment of autism spectrum disorder: surveillance and screening

Community health care providers and other professionals involved in supporting optimal child development (eg, early childhood educators) play a critical role in working with parents to identify early signs of ASD. Screening and surveillance are complementary processes aimed at identifying children who require further assessment, with an overall goal of reducing or preventing subsequent disability through earlier initiation of

intervention [1]. Screening refers to administering and scoring a specific instrument (eg, parent questionnaire) to identify at-risk individuals in need of further assessment. 'First stage' (or universal) screening targets all children regardless of level of concern or other risk factors; 'second stage' screening is limited to children who are flagged due to identified concerns, a positive family history, or other risk factors (eg, medical diagnosis with a known association with ASD).

In contrast, surveillance consists of an ongoing process that includes inquiry about parents' concerns and observations of the child, generally in the context of an ongoing clinical relationship (ie, by a community physician), to obtain an overall picture of the child's developmental health over time [2]. In that context, decisions about referral for further evaluation are made based on clinical judgment. Developmental surveillance can include the administration of standardized tools (including screening questionnaires), but with the aim of obtaining additional information to help inform clinical decision-making, rather than using scoring cut-points as a basis for referral.

There is growing evidence supporting the clinical utility of ASD screening by community health providers (ie, predictive value of a positive screen, potential to detect children earlier than by general monitoring of developmental concerns) [3–5]. However, most screening research focuses on accuracy (ie, 'correct' classification of children with ASD, but not children who do not have ASD, as being at-risk and in need of further assessment based on a positive screen) rather than meaningful end-points such as age of diagnosis, access to early intervention and long-term outcomes [6]. As a result, there is still considerable debate as to what should currently be considered 'best practice', with some authors advocating for broad-scale implementation of universal ASD screening as early as 18 months in accordance with current American Academy of Pediatrics recommendations [7,8], while others continue to argue that more evidence is needed [9,10].

There also remains uncertainty about the relative merits of a first- or second-stage screening strategy for ASD. First-stage screening has the potential advantage of higher sensitivity (and thus, more children with

ASD being correctly identified at an early age), but also tends to identify more children who do not have ASD (ie, false positives), with potential implications for parental stress and straining service capacity for appropriate follow-up assessments. Second-stage screening also presents challenges. To be effective, general developmental surveillance must correctly identify children with ASD for further assessment; however, there is evidence that current screening methods may miss some children who would otherwise be flagged by ASD-specific screens [5,11].

Recent research advances help reconcile the advantages and disadvantages of first- versus second-stage screening by offering a combined, integrated approach, while at the same time, further supporting the overall utility of ASD screening. For example, the Modified-Checklist for Autism in Toddlers (M-CHAT) is an ASD screening tool which has been evaluated in large community samples of 16- to 30-month-olds assessed during well child visits. By combining a 23-item parent questionnaire with a structured follow-up interview to clarify items endorsed by parents, the M-CHAT essentially functions as a combined level 1 and level 2 screening tool, with approximately 50–60% of screen positive children who are referred and assessed being subsequently diagnosed with ASD [12–14]. Recently, Robins et al [15] reported validation data for a new version of this instrument, the ‘Modified Checklist for Autism in Toddlers, Revised with Follow-up’ (M-CHAT-R/F). The questionnaire was reduced to 20 items and a scoring algorithm with three risk ranges was developed. Children in the ‘low-risk’ range (<3 items endorsed) did not require the follow-up interview or any other additional evaluation (93% of all cases). Children in the ‘medium-risk’ range (3–7 items endorsed; 6% of all cases) required the follow-up interview to clarify their risk for ASD; if at least 2 items remained positive, then referral for diagnostic evaluation was indicated. Children in the ‘high-risk’ range (8 or more items endorsed; 1% of all cases) were at sufficiently high risk to be referred directly for diagnostic assessment without the follow-up interview. This revised scoring and referral algorithm reduced the initial screen positive rate (from 9.2–7.2%) and increased the overall rate of ASD detection (67 vs. 45 per 10,000) [15].

Wetherby et al [16] have adopted a similar strategy with the Communication and Symbolic Behavioral Scales Infant Toddler Checklist (CSBS-ITC), recommending that children identified by this broadband screener, designed to detect communication delays, have an additional assessment for behavioral ‘red flags’ [17] to further determine which children require an ASD diagnostic assessment. The ITC has been demonstrated to have very high sensitivity in a community sample, detecting over 90% of children with ASD by age 24 months [16], although evaluation of the care pathway that includes the second level behavioral assessment has not yet been reported.

As ASD screening in community settings continues to be evaluated [6] and health policy regarding screening continues to evolve, developmental specialists will continue to have an important role in working with their community colleagues to encourage careful monitoring for early signs of ASD, understanding of referral and care pathways, and working collaboratively with families through the assessment process. This collaboration extends to helping parents understand the diagnosis and to navigate the service system so that the child and family access appropriate supports and services in a timely way. Establishing a positive working relationship with community health professionals also lays the foundation for ongoing co-management of the child in relation to their diagnosis, and other potential physical and emotional health issues that may arise [18].

## **Team structure and function**

The success of a diagnostic assessment hinges on effective teamwork. This is true whether the diagnosis is made with a multidisciplinary team of health care providers in a tertiary care setting or a single clinician in a community practice. Clinicians learn to appreciate that they work in concert with the child and his or her family to gather and synthesize information that may lead to a diagnosis but will, more importantly, foster a greater understanding of the child’s neurodevelopmental profile. This profile informs not only the family, but also service providers and members of the family’s community support system.

Most jurisdictions do not mandate which professionals need to be involved in the diagnostic assessment. Each child will present with unique

challenges, and different team members may need to be consulted with each new case. Table 2.1 lists potential contributions by family members and professionals.

Because of high demand for ASD assessments, constrained health care resources and limited geographic access to tertiary care centers, an in-person multidisciplinary team assessment may not be possible. In such cases, clinicians may be part of a 'virtual' team, in which assessments occurring across time and locations are incorporated into the diagnostic formulation. While virtual teams may allow for some efficiency, they create an additional challenge for the team leader to synthesize information from various contexts. Technological advances can now be used to

Team member	Role
Leader	Coordinates elements of the assessment
	Responsible for communication of assessment results to family
	Develops management and follow-up plan
Caregivers	Provide bulk of history
	Provide consent to contact other individuals with knowledge of child
Siblings	Source of valuable information
	Provide clinicians an opportunity to observe peer interaction
Physician	Medical and developmental history (+/- standardized interview tools)
	Physical examination
	Observation (formal and informal)
	Coordination of further testing (biochemical, genetic, imaging, etc)
Psychologist	Developmental history (+/- standardized interview)
	Observation (formal and informal)
	Intelligence or developmental assessment tools
	Other formal neuropsychological tests (adaptive function, executive function, achievement, attention/hyperactivity/impulsivity)
Speech-language pathologist	Formal assessment of language and communication skills
	Assessment of speech and language pragmatics
Occupational therapist	Sensory profile
	Motor coordination

Table 2.1 Team members and potential contributions to diagnostic assessment.

connect team members across settings to allow valuable team discussions to occur with all members present, which often provides richer insight into the child's profile.

## Components of diagnostic assessment

### Patient history

A thorough and accurate history provides the foundation for a diagnosis of ASD. Because it is often the first part of the assessment, it also allows the clinician to develop a therapeutic interaction with caregivers. This rapport greatly influences the tone of the assessment and can improve receptiveness to the results of the evaluation.

The clinician should begin by asking about the caregivers' understanding of the reason for the referral and their goals for the assessment. Caregivers may or may not have been told about the possibility of ASD. To provide informed consent for the assessment, caregivers must know that you will be evaluating for ASD. Some approaches to broach this topic include:

*"Sometimes families have specific things they want us to look for, such as autism spectrum disorder or attention deficit hyperactivity disorder. Is there anything you'd like us to look for in your child?"*

*"We will be looking at all areas of your child's development; however, your doctor wrote that (child's name) has challenges with his/her (language/social skills/play). In these cases, it is important that we look for any signs of an autism spectrum disorder."*

It is also important to start the interaction by explaining the various components of the assessment, including the amount of time and number of appointments you anticipate. By helping caregivers understand what to expect, clinicians can foster a trusting environment that will relieve some anxiety for the family.

The clinician should then gather more information about the caregivers' concerns and observations that have been communicated to

them by other people in the child's life. It is important to solicit information about how the child functions in multiple environments, such as daycare, school and recreational activities.

The history should explicitly cover all DSM diagnostic criteria with clear examples of each criterion when present. The Autism Diagnostic Interview – Revised (ADI-R) [19] is a standardized interview that can be completed by an experienced clinician in approximately 1.5 to 2 hours and is useful for children with a mental age of at least 2 years of age. The ADI-R consists of 93 items grouped into domains of Language/Communication; Reciprocal Social Interactions; and Restricted, Repetitive, and Stereotyped Behaviors and Interests.

The developmental history should cover all developmental domains regardless of the reason for referral. While the focus should be on current skills, the progression of communication and social skills is essential to elicit, as is any significant loss of previously acquired skills (regression). Many clinicians choose to incorporate a standardized adaptive skills interview, such as the Vineland Adaptive Behavior Scales II (VABS-II) [20], to measure current performance across domains of development. The VABS-II can be incorporated into a developmental history and provides a standardized measure of the child's current developmental function with standard scores, percentiles, and age equivalents.

The history should also include a thorough past medical history, including the pregnancy and neonatal period. The review of systems should pay particular attention to hearing, vision, sleep, nutrition, and gastrointestinal symptoms. A three-generation family history should be elicited including ASD, developmental delays, school failure, learning disabilities, language delays, attention deficit hyperactivity disorder, anxiety (particularly social anxiety) or other psychiatric conditions, hearing impairment, seizures, congenital anomalies, multiple spontaneous abortions, and consanguinity. A social history, including living situation, employment, and support is important and may influence the management plan.

## Questionnaires

In cases where a formal ASD interview tool such as the ADI-R is not feasible, clinicians may choose to provide caregivers with questionnaires,

which are less time-consuming and can be completed between appointments. These questionnaires can also be given to other individuals who have frequent contact with the child, such as child care staff and teachers.

The Social Communication Questionnaire (SCQ) [21] is a screening tool based on the ADI-R that can be used for children over 4 years with a mental age over 2 years of age. It consists of 40 'yes/no' items and can be quickly and easily completed by caregivers. Some clinicians prefer to use it as a guide in a semi-structured interview format to cover the diagnostic criteria systematically during the history taking.

The Social Responsiveness Scale-2 (SRS-2) [22] is designed to identify social impairment that is seen in ASD and to differentiate it from social difficulties that occur in other disorders. It can be completed for children as young as 30 months and takes approximately 15 minutes for caregivers to complete.

## **Informal observation**

The child may not need to be present for the entire developmental history; however, it is important to observe the child in the same room as the caregivers for part of the assessment. This time is filled with rich information about the interaction between the child and caregivers, how the child adapts to the new environment, the child's attempts to obtain, maintain, and direct the caregiver's attention, and how the caregiver deals with undesirable behavior.

## **Formal observation**

In most cases, clinicians will use a formal observational tool to help them systematically observe behaviors consistent with DSM criteria. The Autism Diagnostic Observation Schedule, 2nd edition (ADOS-2) [23] is a frequently utilized semi-structured interaction that provides structured opportunities to assess communication, social interaction, play, and restricted and repetitive behaviors. Clinicians must undergo training to administer and score the ADOS. Modules 1–4 provide cut-off scores for ASD and can be used in children as young as 31 months; the Toddler Module provides ranges of concern (instead of cut-off scores) and can be used in children between 12 and 30 months of age [23].



The Childhood Autism Rating Scale (CARS-2) is a brief observational scale with 15 items that can be used in children aged 2 and older.

## **Physical examination**

A physical examination by a physician is a necessary component of the diagnostic assessment because of the many medical comorbidities associated with ASD (see Chapter 3). If a physician is not part of the diagnostic team, the child's pediatrician can perform a thorough examination after the diagnosis is made. The physical examination should be comprehensive, with particular attention to growth parameters and dysmorphic features. A thorough neurologic examination is warranted in all children with suspected ASD.

Because of sensory processing difficulties, children with ASD often have difficulty with the physical examination. It is best to attempt the exam when the child is comfortable with the environment and in a good mood. Because the diagnosis itself does not rely on the results of the physical examination, it may be appropriate to complete it at a later date.

## **Additional assessment components**

Other types of assessments, as listed in Table 2.1, may be useful depending on the individual needs of the child. At a minimum, hearing and vision development must be documented prior to a diagnosis of ASD. A psycho-educational assessment can be performed at the time of diagnosis or when the child reaches school age. Many children will also benefit from speech and occupational therapy assessments, which may help to inform the diagnosis and formulation or can be part of the management plan.

## **Formulation and feedback**

The formulation synthesizes the results of the assessment and feedback involves presenting diagnostic impressions. The formulation is typically a written summary of findings from the patient assessment(s) and is what will be consulted by family and service providers going forward after a diagnosis. Feedback involves presenting the formulation and sharing a diagnosis; parents have described the impact of the feedback session as “the hour changed our lives forever.” Because of the importance of the

feedback session, it is useful for the clinician to finalize a formulation before giving a diagnosis.

## **Formulation**

Introducing the formulation section with a brief summary of patient information and observations collected from parents, the referring clinician, and others in the community sets the stage for making a diagnosis. It can include relevant developmental and medical history, symptoms that address the diagnostic criteria, and how initial concerns about ASD developed. After gathering a patient history, results of assessments can be briefly summarized, along with reports from teachers, childcare professionals, and other relevant individuals who have come into contact with the patient. This information can consist of both formal and informal observations relating to the criteria that led to a diagnostic impression.

One method of doing this is via a 'diagnostic paragraph' which may help a parent or caregiver to better understand the diagnostic criteria that appear to have been met. An explanation of the current severity, prognosis, and ASD subtype (even if an unofficial one, such as an Asperger's disorder) may be included in the diagnostic paragraph or may warrant a separate discussion. Similarly, the clinician should explain how ASD may affect dimensions of development, such as language, cognition, developmental status, and functional or adaptive skills. Other coexisting diagnoses such as intellectual disability, language impairment, motor speech disorders, attention deficit hyperactivity disorder, and social or other forms of anxiety disorders need to be discussed.

An itemized management plan and list of recommendations should accompany a diagnosis. This can include a medical follow-up and investigation, referrals to community services, access to helpful resources for families, and strategies tailored to the child's various environments to promote engagement and learning. This list should be as comprehensive as possible, as it is often referenced by families and health care professionals throughout the patient's life.

## Feedback

A feedback session follows a similar structure to the formulation. At each stage, the clinician should ensure the parents/caregiver understands and agrees with clinical observations. This will help the family to appreciate the evidence upon which a diagnosis is based and help to prevent later disagreement or confusion.

Some clinicians find it hard to use the term ‘autism’ with parents and caregivers; this could be made less difficult if the medical terms have been discussed throughout the assessment process and are reiterated during the feedback session. After presenting the evidence, clinicians must communicate the diagnosis in a clear and unambiguous statement. An example of a way to communicate the diagnosis is as follows:

*“[Child’s name] has difficulty sharing his/her feelings with another person. He/she has very focused interests and insists that the world ‘stays the same.’ He/she also has intense sensory interests and repetitive movements. When we see this combination, the name we give it is autism spectrum disorder. Based on my assessment, he/she has autism spectrum disorder.”*

Families will have varying reactions in the moments after they receive the diagnosis. Though the ensuing silence can be uncomfortable, it is important to let families reflect on the impact of the diagnosis. This is the time when the family may wish to guide the remainder of the feedback session with the many questions they may have about their child.

Prognosis will often be the first question from parents. Explaining the predictors of outcome, such as early identification and intervention, intellectual level, age when language emerges, and coexistence of other medical issues, can be helpful for parents. Unfortunately, there is still a great deal of uncertainty in predicting the outcome of individuals with ASD and this must also be communicated to the family. Discussion of a management plan is vital but may be difficult during the first session

when the diagnosis is shared. The family may require a number of sessions to discuss the modality and intensity of interventions, medical issues, diet and nutrition, alternative and complementary therapies, and management of difficult behaviors and emotion dysregulation.

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