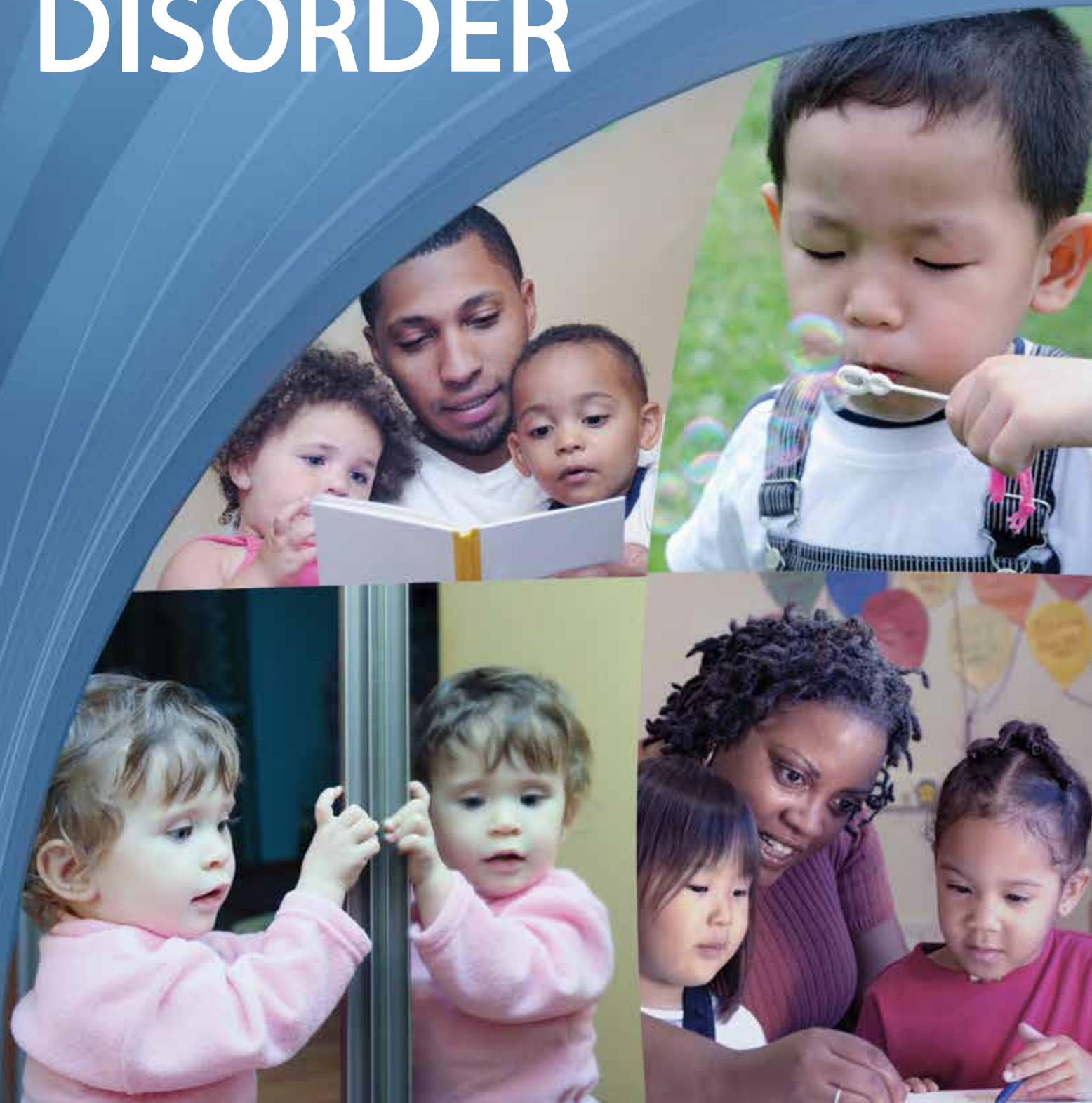


# CONNECTICUT GUIDELINES *For a Clinical Diagnosis of* AUTISM SPECTRUM DISORDER

Connecticut Guidelines for a Clinical Diagnosis of Autism Spectrum Disorder

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# CONNECTICUT GUIDELINES FOR A CLINICAL DIAGNOSIS OF AUTISM SPECTRUM DISORDER

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## Preface

Overall, the number of children who are diagnosed with autism spectrum disorders (ASD) has increased. The Centers for Disease Control (CDC; CDC, 2012a) now estimates that 1 in 88 children have ASD (1 in 54 boys and 1 in 252 girls). This represents a 23% increase from data collected two years previously (CDC, 2009). This increased prevalence suggests that there is a growing need for screening and further referral, when indicated, for a diagnostic evaluation for children suspected of having ASD. To receive appropriate diagnostic services, a child must be able to obtain a comprehensive evaluation conducted by competent and qualified personnel using a protocol of acceptable tools and procedures. This is especially critical since early diagnosis of ASD is needed to help children and their families to realize the positive outcomes that can be achieved by participating in appropriate intervention services at the earliest point (e.g., National Research Council, 2001; Volkmar, Reichow, & Doehring, 2011). It is essential then that parents, providers and educators remain vigilant in ensuring that all children, regardless of gender, race, ethnicity or socioeconomic status are appropriately diagnosed as early as possible, and provided with the individualized services that can lead to optimal outcomes. This document contains guidelines to meet the need for a common understanding across Connecticut regarding the elements essential in making an accurate diagnosis of ASD.

The *Connecticut Guidelines for a Clinical Diagnosis of Autism Spectrum Disorder* (hereafter referred to as *Guidelines*) are a result of collaborative efforts that were initiated under the Connecticut Act Early Project. This project began in 2007 as a partnership among the National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention (CDC), the Maternal and Child Health Bureau (MCHB) at the Health Resources and Services Administration (HRSA) and the Association of University Centers on Disabilities (AUCD). As part of the Act Early Campaign, regional summits of state teams were held during 2008-2010, with a Connecticut team participating in the New England Act Early Summit in Providence, Rhode Island in April 2010. The team consisted of representatives from the University of Connecticut Center for Excellence in Developmental Disabilities Education, Research and Service; the Connecticut Leadership Education in Neurodevelopmental and related Disabilities (both of the University of Connecticut Health Center); the Yale Child Study Center and the Yale Developmental-Behavioral Pediatrics Program (both of the Yale School of Medicine); Connecticut Children's Medical Center; Hospital for Special Care; the Connecticut State Departments of Children and Families, Developmental Services, Social Services; the Connecticut Office of Protection and Advocacy for Persons with Disabilities; the Connecticut chapter of the American Academy of Pediatrics; a local Head Start Agency; parent advocacy organizations. Parents of children and adults who have ASD were also on the team.

During the summit, the Connecticut Act Early Team developed plans to address the state need for improvement with the early identification, diagnosis and intervention of young children with ASD. To represent this mission, the team adopted the following 10 year vision for Connecticut:

*In order to assure valued life outcomes, all of Connecticut's diverse families and other stakeholders will be aware of the early signs of ASD and have knowledge about, and access to, evidenced-based, individualized, and timely screening, diagnostic evaluation and interventions implemented by a competent work force and a funded, coordinated system of care.*

In order to realize this vision, the team felt that a number of service components had to be defined and adopted throughout the state. In particular, the team decided to focus on the development of Connecticut diagnostic guidelines for the identification of young children with ASD.

To begin the process, the Act Early Team identified a number of principles to guide the development of the guidelines. These follow:

1. Early identification of children with ASD through accurate screening and diagnosis is essential to access individualized and effective interventions that result in optimal outcomes. While it is out of the purview of this document, the American Academy of Pediatrics recommends general developmental screening at the 9-, 18- and 30-month well child visits. Screening of all children for ASD using a standardized screening instrument is recommended at the 18 month visit and again at the 24 month visit, and whenever parents raise a concern about their child's development (see Johnson & Myers, 2007). See **Appendix A** for the American Academy of Pediatrics surveillance and screening algorithms.
2. Everyone in Connecticut, including diverse and underrepresented groups, should have easy and equitable access to diagnostic evaluations and intervention services. The *Guidelines* should not impede access to services for children and families, nor be interpreted as limiting a diagnostician's approach to assessing and evaluating children.
3. A family-centered approach is the foundation of all diagnostic services and interventions, and is represented throughout the *Guidelines*.
4. A medical home approach provides comprehensive primary care that is accessible, continuous, comprehensive, family-centered, coordinated, compassionate, and culturally effective. A medical home facilitates partnership between a child's family or caregiver, the child, and the primary health care provider (American Academy of Pediatrics, n.d.; <http://www.medicalhomeinfo.org/>), and the concept is supported through these *Guidelines*.
5. Information on existing state policies and programs for children with ASD (e.g., Birth to Three, special education, insurance coverage) should be made available and accessible to all.
6. While the focus of the *Guidelines* is on the early identification and diagnosis of young children with ASD, the principles included in the document apply to all children suspected of a disability or developmental delay.



7. Current research and scientific evidence should inform diagnostic evaluations to enable earlier and more accurate identification of children with ASD who live in Connecticut.

To accomplish this a multidisciplinary 12 member work group consisting of parents, autism researchers, educators, and practitioners from developmental behavioral pediatrics, early intervention, public schools/special education, developmental psychology, child psychiatry and law was enlisted to write the guidelines. The work group met monthly to draft the guidelines, using a facilitator to discuss the content and format of the guidelines. These discussions were recorded and written into a working document by one member of the group who was responsible for developing the written draft of the guidelines. Between meetings, the workgroup reviewed, edited and resolved differences on the written drafts.

The work of the work group was supported by a larger advisory group of 24 experts from Connecticut including parents of children with ASD, self-advocates, psychologists, professionals who inform intervention planning processes, educators, early intervention providers, and representatives of multiple state agencies serving children and families. This larger group brought together diverse perspectives to ensure that the guidelines were relevant to the evidence on best practice in diagnostic evaluation, as well as the Connecticut service delivery system. The larger advisory group was involved in three meetings during the process in order to review and approve decisions about key components of the guidelines. Most importantly, the group provided feedback on the social validity of the guidelines to diagnosticians, families, higher education faculty, public school administrators and personnel, advocates, and others. This collaborative process resulted in the Connecticut *Guidelines*.

These *Guidelines* provide recommendations and guidance for the clinical diagnostic evaluation of children who may have ASD in the State of Connecticut. The purpose of these *Guidelines* is to provide a consistent and comprehensive source of information for diagnosticians who conduct these evaluations.



# Chapter 1:

## Autism Spectrum Disorder



## History of Diagnostic Criteria

The earliest and most complete description of what is now called ASD was written by Leo Kanner in 1943. Kanner described 11 children who lacked the usual disposition to make social contact and had a strong resistance to change in their environment. Kanner called the condition “early infantile autism.” Beginning with the third edition of the American Psychiatric Association’s *Diagnostic and Statistical Manual (DSM-III, 1980)*, autism was included in the psychiatric manual for the first time as a pervasive developmental disorder (PDD). The DSM was revised in 1994 (APA, 1994) and expanded the number of PDDs to five (autistic disorder, Asperger’s disorder, Rett’s disorder, childhood disintegrative disorder, and pervasive developmental disorder, not otherwise specified), which were characterized by the triad of core deficits in socialization, communication, and restricted and repetitive behaviors. The *DSM-IV* and the international diagnostic system from the World Health Organization, the *International Classification of Disorders*, tenth edition (*ICD-10*; WHO, 1994) were aligned to ensure a universal definition for the PDDs was used. The *DSM-IV* was updated with a text revision in 2000 (*DSM-IV-TR*; APA, 2000).

The work on these guidelines spanned the publication of both the *Diagnostic and Statistical Manual of Mental Disorders*, fourth edition (*DSM-IV*, APA, 1994) and the *Diagnostic and Statistical Manual of Mental Disorders*, fifth edition (*DSM-5*, APA, 2013). In preparing the *Guidelines*, the work group determined it was important to present the most recent conceptualization and diagnostic criteria, which are now the *DSM-5*. While the focus of these guidelines is on the most recent diagnostic criteria (*DSM-5*), we are also providing brief descriptions of the *DSM-IV* Pervasive Developmental Disorders (PDDs) as well as comparisons between *DSM-IV* and *DSM-5* criteria. Since many readers might be familiar with the *DSM-IV* criteria, we have provided two illustrations of the changes that have been made. Table 1 provides a comparative summary of the characteristics of the diagnostic criteria for *DSM-IV* and *DSM-5*. **Appendix B, Figure B1** provides a cross-walk of the symptoms contained in the *DSM-IV* definitions to the corresponding symptoms in *DSM-5*. The *DSM-5* and *DSM-IV* criteria for Autism Spectrum Disorder and the Pervasive Developmental Disorders, respectively, are shown in **Appendices B and C**. The criteria in the World Health Organization’s *International Classification of Disease*, tenth edition (*ICD-10*, 1994) is also in **Appendix C**.

## Description of Current Diagnostic Criteria

The publication of the *DSM-5* in May 2013 (APA, 2013) presented major changes to the conceptualization of ASD. Most notably, the term Pervasive Developmental Disorders (PDDs) was replaced with Autism Spectrum Disorder (ASD) and four of the five categorical diagnoses of the *DSM-IV-TR* (autistic disorder, Asperger’s disorder, childhood disintegrative disorder, and pervasive developmental disorder, not otherwise specified [PDD-NOS]) were subsumed into one diagnostic category (Rett syndrome, which was found to have a unique genetic etiology was retained as a specifier, which is a new element of the *DSM-5* which is clarified later in this paragraph). Second, the triad of symptoms from the *DSM-IV-TR* (social interaction, communication, and restrictive and repetitive behaviors) was reduced to a dyad (social communication and social interaction

skills; and restricted, repetitive patterns of behavior, interests, or activities). Among restricted and repetitive behaviors, for the first time hyper- or hypo-reactivity to sensory input or unusual interests in sensory aspects of the environment was included. Third, the *DSM-5* states that criteria (e.g., symptoms) can be met currently or by history. The *DSM-5* also contains a note that individuals who have a well-established *DSM-IV* diagnosis of autistic disorder, Asperger's disorder, or PDD-NOS should continue to be given the diagnosis of autism spectrum disorder. Fourth, the *DSM-5* includes specifiers. The specifiers are intended to provide an "opportunity to define a more homogeneous subgrouping of individuals with the disorder who share certain features ... and to convey information that is relevant to the management of the individual's disorder" (APA, 2013, p. 21-22). The unique system of specifiers for ASD includes a functional severity level across a three-level scale (requiring support, requiring substantial support, and requiring very substantial support) for both the social communication and restricted, repetitive behavior domains. There are also specifiers for the presence of accompanying intellectual disability and/or language impairment and associations with other known medical or genetic conditions, environmental factors, other neurodevelopmental, mental, or behavioral disorders, and catatonia. The specifiers are not mutually exclusive or jointly exhaustive; thus more than one specifier can be given (e.g., ASD with intellectual impairment without language impairment). In *DSM-5*, Attention-Deficit/Hyperactivity Disorder (ADHD) is no longer excluded as a co-occurring condition, and catatonia is included as a specifier. Finally, a new diagnostic category, Social (Pragmatic) Communication Disorder (SCD), was added for individuals who present with social deficits in the absence of restricted, repetitive behaviors.

The *DSM-5* criteria are based on extensive research and provide a state of the art understanding of the spectrum of functional and pragmatic challenges associated with ASD. This understanding includes recognizing that the strengths and needs of those diagnosed with ASD represent a continuum. However, these diagnostic criteria were recently published, and have not been utilized extensively on a large scale. In order to provide clarity about prior conceptualizations of ASD diagnostic criteria, the next section offers this information to serve as a reference when reviewing client records.

## **Pervasive Developmental Disorders of *DSM-IV* and *ICD-10***

The following diagnostic criteria provide information for use in the context of *DSM-5*.

### **Autistic Disorder**

Autistic disorder (childhood autism in *ICD-10*) is characterized by impairment in each of the three core areas of social interaction, communication and restricted repetitive behaviors. The estimated prevalence of autistic disorder is 21/10,000 (Fombonne, 2009). The *DSM-IV-TR* criteria for autistic disorder includes two or more impairments in social interaction, one or more impairments in communication, and one or more restricted, repetitive and stereotyped behaviors, to total at least six. Prior to age three, delays or impaired skills and functioning should be found in at least one of the three areas: social interaction; language as used in social communication; or symbolic or imaginative play.



## Asperger's Disorder

Asperger's disorder (Asperger's syndrome in *ICD-10*) is characterized by typical language development through age two with deficits in social communication as well as restricted and repetitive behaviors. While estimates of the prevalence of Asperger's disorder are not as robust as for the other two disorders on the autism spectrum, due to its more recent inclusion in the *DSM*, Fombonne's (2009) analysis indicates a prevalence of approximately 6/10,000. The criteria for a diagnosis of Asperger's disorder include children who exhibit at least two behavioral impairment items from the social interaction list and at least one from the repetitive and stereotyped behaviors list. It is important to be aware that the diagnostic criteria for Asperger's disorder do not include significant delays in language. This is one of the primary differences between autistic disorder as compared to Asperger's disorder. Given that the criteria for Asperger's disorder include typical cognitive and language development in early childhood, the diagnosis is typically made later than autistic disorder. In fact, studies have found that children with Asperger's disorder are diagnosed on average between ages 7 and 9, which can be one to three years later than for children with autistic disorder and PDD-NOS (Wiggins, Baio, & Rice, 2006; Mandell, Novack, & Zubritsky, 2005; Noterdaeme, Springer, & Wriedt, 2008; Williams, Thomas, Sidebotham, & Emond, 2008).

## Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)

PDD-NOS (atypical autism in *ICD-10*) is a diagnosis used when children have significant and pervasive deficits in social interaction, and either communication deficits or repetitive and stereotyped behaviors but do not meet the threshold set for other PDDs (e.g., autistic disorder, Asperger's disorder). The prevalence of PDD-NOS is estimated to be 37/10,000 people, which is nearly twice the rate of autistic disorder (Fombonne, 2009). PDD-NOS is used clinically when a child has what a diagnostician considers to be a "mild" form of autism or when the onset of symptoms consistent with a pervasive developmental delay occur after age three. This diagnosis continues to be ambiguous as the criteria are less prescriptive than those for autistic disorder and Asperger's disorder. However, a recent study found that in a group of 66 young people with PDD-NOS, 97% had impairments in two of the three core domains of social interaction and communication, but no significant repetitive or restrictive behaviors (Mandy, Charman, Gilmour, & Skuse, 2011).

## Childhood Disintegrative Disorder (CDD)

CDD is characterized by regression in more than one area of functioning (e.g., motor, social, language) after at least two years of typical development. The loss of skills generally occurs over the course of several months with developmental regression occurring prior to age 10, but typically by the age of five. Children with this disorder display behaviors similar to children with autistic disorder, with impairment in social communication skills, and restricted, repetitive and stereotyped patterns of behavior. CDD is an extremely rare condition (approximately 2/100,000; Fombonne, 2009).

**Table 1. Comparison of key characteristics of diagnostic criteria for the Pervasive Developmental Disorders in *DSM-IV-TR* and Autism Spectrum Disorder in *DSM-5***

DSM-IV-TR		DSM-5	
Diagnostic Classification			
Pervasive Developmental Disorders		Autism Spectrum Disorder	
Number of Diagnostic Categories			
<div>5</div> <ul style="list-style-type: none"><li>Autistic Disorder</li><li>Asperger's Disorder</li><li>Rett's Disorder</li><li>Childhood Disintegrative Disorder</li><li>PDD-NOS</li></ul>		<div>1</div> <ul style="list-style-type: none"><li>Autism Spectrum Disorder</li></ul>	
Number of Domains			
<div>3<sup>a</sup></div> <ul style="list-style-type: none"><li>social interaction</li><li>communication</li><li>restricted, repetitive and stereotyped patterns of behavior</li></ul>		<div>2</div> <ul style="list-style-type: none"><li>social communication and social interaction</li><li>restricted, repetitive patterns of behavior</li></ul>	
Number of Criteria			
<div>12<sup>a</sup></div> <ul style="list-style-type: none"><li>4 social</li><li>4 communication</li><li>4 restricted, repetitive behavior</li></ul>		<div>7</div> <ul style="list-style-type: none"><li>3 social</li><li>4 restricted, repetitive behavior</li></ul>	
Number of Criteria Needed for Diagnosis			
<u>Autistic Disorder</u> – 6 criteria <ul style="list-style-type: none"><li>at least 2 social</li><li>1 communication</li><li>1 restricted repetitive behavior</li><li>2 additional from any category</li></ul> <u>Asperger's Disorder</u> - 3 criteria <ul style="list-style-type: none"><li>at least 2 social</li><li>1 restricted, repetitive behavior</li></ul> <u>PDD-NOS</u> – 2 criteria <ul style="list-style-type: none"><li>at least 1 social</li><li>1 from either communication or restricted, repetitive behavior</li></ul>		<u>Autism Spectrum Disorder</u> – 5 criteria <ul style="list-style-type: none"><li>3 of 3 social communication and social interaction</li><li>2 of 4 restricted, repetitive behavior</li></ul>	
Criteria (symptom) Presentation			
<div>Current</div>		<div>Current or by history</div>	
Age By Which Symptoms Must be Present			
Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.		Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life).	

**Key:** <sup>a</sup> – for Autistic Disorder and PDD-NOS; Asperger's Disorder does not contain the communication domain and Rett's Disorder and CDD have unique domains and criteria



## Chapter 2

### The Diagnostic Evaluation



The essential elements of a clinical diagnostic evaluation include a detailed child and family/caregiver history, an assessment of the core features of ASD, and a comprehensive medical examination to exclude other diagnoses. See Table 7 for a summary of the essential components of a diagnostic evaluation. The diagnostician should also consider whether co-occurring disorders exist and if genetic testing or a referral for another type of diagnostic evaluation is indicated. It is recognized that conducting a high quality and comprehensive evaluation consumes a considerable amount of time which may not be commensurate with the level of public/private insurance reimbursement. While this document does not address the issue of financial reimbursement for diagnosticians, important work remains to be completed in this area.

Before describing the diagnostic process, we cannot overemphasize the importance of using a family-centered approach throughout. A significant portion of the process of diagnosing ASD involves listening to and talking with families about their child in order to understand the child's history, current behaviors, strengths, and weaknesses. Therefore it is critical that diagnosticians are trained to have a family-centered approach, such that family members/caregivers are viewed as essential and valuable partners in the process of diagnosing children (National Center for Family-Centered Care, 1989). One of the keys to providing family-centered care is to recognize that a family consists of those members the family chooses to call "family," thus diagnosticians must be inclusive when asking about and involving family members/caregivers in a diagnostic evaluation. This requires flexibility on the part of the diagnostician, in terms of scheduling, respecting social and cultural values, and building on the strengths of each child and family. Information gathered from the family when paired with observations and data gained from thorough assessments, contribute to an accurate diagnosis.

## **Child and Family/Caregiver History**

The initial parts of the clinical diagnostic evaluation involve obtaining a comprehensive child and family/caregiver history by reviewing medical and other available records, including prior evaluations, and interviewing the child's family/caregiver to understand their concerns and to learn about the child's past and present behavior and functioning.

### **Record Review**

The diagnostic clinician should request and obtain medical and other available records as part of the diagnostic evaluation process. This can ensure that duplicative tests and assessments will not be requested unnecessarily. A thorough record review provides information about the child's diagnostic history, previous diagnoses for the same or different presenting symptoms, assessments that may have been conducted and other medical assessments that have been completed. It is also important for the diagnostician to know whether the child is receiving early intervention or special education services under the Individuals with Disabilities Education Improvement Act category of autism (IDEA of 2004, 34 CFR §300.8 (1)(i)-(iii)) as eligibility for early intervention and special education is different



**Table 3. Examples of records to be reviewed as part of diagnostic evaluation**

<b>MEDICAL RECORDS</b>
Routine well-child care and concerns raised over time
Specialist evaluations by medical specialists and subspecialists (e.g., developmental behavioral pediatrician, neurologist, allergist, gastroenterologist, psychologist, psychiatrist, geneticist)
Birth records, including results of newborn screening conducted; currently includes testing for 41 disorders (Connecticut Department of Public Health, 2012)
Prior hospitalizations
Medical tests that have been completed and reasons the tests were conducted
Medication history (especially use of any psychotropic medications, complementary and alternative medications, dosage and child's response)
Prior medical treatments, including non-traditional therapies
Evidence of neonatal encephalopathy
<b>PRIOR ASSESSMENTS</b>
Developmental evaluations or assessments, including results of any ASD screenings and evaluations that have been completed
Other evaluations or assessments (e.g., neuropsychological evaluations, functional behavioral assessments, evaluations conducted by speech language pathologists, occupational therapists or physical therapists)
<b>EDUCATIONAL RECORDS (E.G., INDIVIDUALIZED FAMILY SERVICE PLANS [IFSPS], INDIVIDUALIZED EDUCATION PROGRAMS [IEPS])</b>
Birth to Three and educational evaluations, including those conducted by school personnel and related service providers (e.g., occupational therapists, physical therapists, speech therapists, school psychologists, social workers) to identify whether the child qualifies for early intervention or special education
Current school services and programs, including any specialty program provided by the school that the child may attend and the special education disability category identified by the child's Planning and Placement Team (PPT)
Any data related to the child's educational progress

than a clinical diagnostic assessment for ASD (see Chapter 4 for further reading on this topic). Ideally, records are reviewed and integrated prior to the child's evaluation. Table 3 shows examples of records that may be reviewed.

## **Family Interview**

Parents are a critical component of the diagnostic process and their perception and knowledge of their child's development should form the basis of the diagnostic process. The

**Table 4. Components of a family interview**

COMPONENTS OF A FAMILY INTERVIEW
Reason for the referral and the source of the referral (such as parent, school, or primary care physician)
Family concerns, including when the concerns were first raised by the family members/caregivers
Preliminary discussion of the family's/caregiver's goals for their child
Birth history, including pregnancy, labor, and delivery and neonatal course, if known
Developmental history including when major milestones were reached (e.g., motor, communication, social, cognitive, adaptive behavior)
Child's medical history, including history of any seizures, hearing or visual impairments, acquired brain injury, allergies, immunization history. Also includes a history of any symptoms that may be associated with ASD, including sleep difficulties, unusual diet, self-injurious behaviors, aggression, and anxiety.
Child's early intervention and educational history, if age appropriate, and the child's response.
Family history (medical, psychosocial), including any history of developmental disabilities, including autism, genetic conditions, learning problems, mental health and behavioral problems in family members. It is important to learn of family members with school problems, alcohol and substance abuse, incarceration and early deaths and those who may have had undiagnosed disorders/conditions as well as any diagnosed neurological and behavioral problems that family members may have experienced. This can be done during the family interview with focused questions about family members going back three generations. This discussion is preliminary to a clinical genetics testing for Fragile X and chromosome studies, which are indicated if the child is found to have ASD (Schaefer, Mendelsohn, & Professional Practice Guidelines Committee, 2008).
Discussion with the family to assess the impact a diagnosis of ASD may have on the family, including the family's ability to accept a diagnosis of ASD, family strengths, and the family's resources.

diagnostician should gather this information prior to, during, and after the formal assessment in order to validate the performance and behavior that is demonstrated by the child. The interview should be comprehensive and also conducted in the parent's primary language in a setting that is comfortable for the parents and other family informants. Sample elements of the interview are contained in Table 4.

## Assessment of Core Features

The central portion of the evaluation occurs during the visit with the child and family and involves the diagnostician observing the child's behavior in relation to the core features of ASD. For some children, especially those under age three who may be enrolled in a home visiting or early intervention program, this may occur in the home environment. For others it could be a school or clinic setting. No matter where, a comfortable "child friendly" environment with developmentally appropriate toys should be created to observe the child at play during the assessment. This type of environment allows the diagnostician to get a sense of how the child typically interacts with familiar and unfamiliar people. Input from those who know the child well may be critical when parents and other key informants find that the observational assessment in the diagnostician's office is not representative of the child's typical behavior.

### Assessment of Social Interaction and Communication Skills

The diagnostic clinician observes the child's behavior to assess whether the quality and characteristics of the child's social communication and interaction skills are consistent with a diagnosis of ASD. Social communication impairments are characterized by difficulty maintaining conversations, deficits in nonverbal communicative behaviors used for social

**Table 5. Signs and symptoms of social interaction and communication skills impairments**

SIGNS AND SYMPTOMS
Does not respond to name by 12 months of age
Avoids eye-contact
Primarily prefers to play alone
Does not share interests with others
Only interacts to achieve a desired goal
Has flat or inappropriate facial expressions
Does not adhere to culturally appropriate personal space boundaries
Avoids or resists physical contact
Is not comforted by others during distress
Uses few or no gestures (e.g., does not wave goodbye)
Does not point or respond to pointing
Appears not to listen to others' speech
Gives unrelated answers to questions
Uses words in idiosyncratic ways

interaction, pragmatic issues, such as over-literal understanding of language, and difficulty with make-believe play. Social interaction impairments are characterized by an inability or disinclination to share and direct attention with another person, called joint attention. Another feature of impaired social interaction is a lack of social referencing (e.g., taking cues from another's facial expression in a new situation). Atypical features may be displayed by the signs and symptoms shown in Table 5 as adapted from the CDC's *Autism spectrum disorders: Signs and symptoms* and the *DSM-5*.

## **Assessment of Restricted, Repetitive Patterns of Behavior**

Atypical behaviors in social interaction alone are not sufficient to make a diagnosis of ASD. For a child to receive a diagnosis of ASD, they must exhibit two or more restricted, repetitive behaviors, interests, or activities, which are the second core feature of ASD. Therefore, the diagnostician must observe the child for evidence that the child engages in such behaviors that are consistent with ASD. This set of symptoms is exhibited by an apparent adherence to routine and discomfort with change, preoccupying interests, and an apparent interest in the parts of objects rather than the whole or its functional use (e.g., spinning the tire of a toy car, rather than “zooming” the car). It is important to point out that restricted, repetitive patterns of behaviors do not emerge and become evident at the same time as the social communication and social interaction impairments associated with ASD. Rather, repetitive and restricted behaviors typically begin increasing around ages four to five years (Charman et al., 2005). Signs and symptoms to help providers and parents identify potential concerns are shown in Table 6, as adapted from the CDC *Autism spectrum disorders: Signs and symptoms* and the *DSM-5*.

## **Standardized Diagnostic Instruments for Autism**

For the majority of children who are evaluated, it may be unclear from the child and family history, interview with the family, record review, and observational assessment whether a definitive diagnosis of ASD can be made. When a clear clinical picture does not emerge from the evaluation, the diagnostic team or lead diagnostician should consider conducting a formal assessment using a standardized diagnostic instrument to assess autism symptoms or should refer the child and family to an appropriately trained and experienced clinician for a diagnostic evaluation. The purpose of the evaluation is to understand behaviors related to social interaction, to assess communication skills, and to ascertain whether restricted behaviors and repetitive interests are present. Importantly, studies have shown that using the Autism Diagnostic and Observation Schedule (ADOS) and the Autism Diagnostic Interview – Revised (ADI-R) in combination provided the greatest sensitivity and specificity for the *DSM-IV-TR* criteria (e.g., Risi et al., 2006), which has also been replicated in preliminary examinations of *DSM-5* criteria (Mazefsky, McPartland, Gastgeb, & Minshew; 2013).

When selecting an assessment instrument it is important to use those with good sensitivity (correctly identifying children who have ASD) and good specificity (correctly identifying children who do not have ASD). The selection of particular instruments is based on

**Table 6. Signs and symptoms of restricted, repetitive behaviors**

SIGNS AND SYMPTOMS
Lines up toys or other objects
Plays with toys the same way every time
Appears fascinated with parts of objects (e.g., wheels)
Gets upset by minor changes
Demonstrates preoccupying interests
Indicates a desire to follow certain routines
Flaps hands, rocks body, or spins self in circles
Repeats words or phrases over and over (echolalia)
Engages in self-directed speech by repeating learned 'scripts' without directing them to others or without apparent communicative intent
Demonstrates hyposensitivity to sensory input (e.g., apparent indifference to pain, heat, or cold)
Demonstrates hypersensitivity to sensory input (e.g., covers ears when on a bus, always must wear sunglasses, will not wear socks with a seam)

the specific questions about the child's behavior and development that need to be addressed, the age of the child, the child's presenting characteristics, the experience of the professional(s) conducting the testing and other factors related to the particular child. Importantly, the instruments discussed in the following section are to be used only with children who have a developmental age of at least 12 months. For those with lower developmental ages, conducting a developmental assessment with a focus on the core features of ASD, rather than an ASD specific assessment might provide better insight into the child's strengths and challenges. Descriptions of sample instruments that could be used to assist in the diagnosis of ASD follow.

**Autism Diagnostic Interview-Revised (ADI-R).** The ADI-R (Rutter, Le Couteur, & Lord, 2003) is a semi-structured diagnostic interview used to assess behaviors related to ASD. The ADI-R takes one and a half to two and a half hours to administer and contains questions about children's early development and developmental milestones and focuses on three functional domains: language/communication; reciprocal social interactions; and restricted, repetitive, and stereotyped behaviors. It is administered by a trained and experienced clinician through a formal interview with the child's caregiver(s). Training on administration of the ADI-R can be completed in 16 hours through a DVD series in addition to two hours of exercises with experienced clinicians. The ADI-R should be administered for children who have a mental age of at least 24 months. A study by Corsello et al. (2007) found that the ADI-R has excellent sensitivity (.90) in distinguishing children with ASD from those who do not have ASD, however the specificity is lower (.54) in distinguishing these two groups. The algorithm used to score the ADI-R distinguishes between children who have *DSM-IV-TR* autistic disorder



from those who do not; it does not diagnose children with PDD-NOS. Although it is likely that the ADI-R will continue to have strong reliability and validity when used for *DSM-5* criteria, revised algorithms are needed before it can be used in clinical practice (e.g., LeCouteur, James, Hammal, & McConachie, 2013).

**Autism Diagnostic Observation Schedule-2 (ADOS-2).** The ADOS-2 (Lord, Luyster, Gotham, & Guthrie, 2012) is a standardized observational assessment of ASD that consists of five modules, which are selected for administration based on the individual's age and use of speech. The ADOS can be administered to toddlers with a developmental age of 12 months through adulthood and to those with limited verbal speech. Through a series of play based tasks, the semi-structured instrument enables trained professionals to assess communication, social interaction, play and restricted and repetitive behaviors (Lord et al., 2012). The assessment takes 40 to 60 minutes to complete. The person administering the ADOS must receive 12 to 18 contact hours of training (in person or via video/DVD) or attend an approved ADOS-2 workshop. Across the five modules ADOS-2 has good sensitivity ( $>.80$ ) and specificity ( $\geq .80$ ) in discriminating autism and PDD from non-spectrum disorders. Scores from modules 1, 2 and 3 of the ADOS can be interpreted to indicate the range of concern raised from the assessment, across four categories from no evidence of autism spectrum disorder to a high level of autism spectrum disorder. The Toddler Module does not produce a score; only ranges of concern (little-or-no, mild-to-moderate, moderate-to-severe) result from administering the assessment (Luyster et al., 2009). Because the ADOS-2 was developed with the new diagnostic criteria in mind, it is not expected that changes to the diagnostic algorithms will be necessary.

**Childhood Autism Rating Scale, Second Edition (CARS2).** The CARS2 (Schopler, Van Bourgondien, Wellman, & Love, 2010) is a behavioral checklist designed to be completed by a trained interviewer/observer with children ages two and older. The instrument includes three forms: a) CARS2-ST (CARS Standard Version Rating Booklet) contains 15 items and is intended for use with individuals younger than 6 years of age and those with communication difficulties or those with below-average estimated IQs to assess functional areas; b) CARS2-HF (CARS High Functioning Rating Booklet) contains 15 items and is intended for use with verbally fluent individuals, 6 years of age and older, with IQ scores above 80 to assess functional areas; and c) CARS2-QPC (CARS Questionnaire for Parents and Caregivers) an unscored caregiver questionnaire. The CARS2 can be administered in 5 to 10 minutes, once the information needed to complete the assessment is collected. The sensitivity and specificity data as reported by the authors of the CARS2 are .81 and .87 respectively.

**Social Responsiveness Scale-2.** The SRS-2 is a measurement of the severity of autism spectrum symptoms as they occur in a child's natural social settings (Constantino & Gruber, 2012). The instrument aids in distinguishing autism spectrum conditions from other child psychiatric conditions by identifying the presence and extent of autistic social impairment. It is administered with children as young as 30 months through adulthood and takes 15 to 20 minutes to administer. There are multiple versions of the SRS-2 which are administered based on the person's age. There is a Pre School form for children ages 2.5 to 4.5 years and a School

Age Form for children four through 18 years. Both of these forms are completed by a parent or teacher. Each version contains a total of 65 items which sum to a total score and also include five subscales: social awareness; social cognition; social communication; social motivation; and restricted interests and repetitive behavior. Research has shown that the sensitivity and specificity are above .75 for this instrument (Constantino & Gruber, 2012). The author of the SRS-2 has developed two new *DSM-5* compatible subscales, based on proposed criteria: Social Communication and Interaction and restricted Interests and repetitive behavior.

**Gilliam Autism Rating Scale (GARS-2).** The GARS-2 (Gilliam, 2006) is a behavioral checklist which consists of 42 items across 3 subscales, a) stereotyped behaviors, b) communication, and c) social interaction. Items are posed in three ways, including a 4-point Likert scale, parent interview, and open-ended questions. It is designed to be completed by parents, teachers, or professionals who are assessing individuals 3 to 22 years of age. While there have been no independent studies published on the specificity and sensitivity of the GARS-2, the version that preceded the GARS-2 (the GARS), which is very similar in content, does not meet recommended levels of specificity and sensitivity. In their review of the GARS, Norris and Lecavalier (2010) found the sensitivity of the GARS to be in the range of .38 to .53 for four studies, with one study finding a sensitivity of .83 (Norris & Lecavalier, 2010). In terms of specificity, a study by Sikora, Hall, Hartley, Gerrard-Morris, & Cagle (2008) reported a specificity of .58. It is unclear how well the GARS-2 aligns with the *DSM-5* criteria.

### **Additional Standardized Measures When the Diagnosis of ASD is Undetermined**

After information is collected from the record reviews, family and child history, and the assessment of core features of ASD, it still may not be clear whether a child should be diagnosed with ASD. In such situations, further use of norm-referenced standardized measures that assess various aspects of development may provide additional diagnostic information. Although administration of standardized developmental and adaptive measures is not essential for a diagnosis of ASD in all cases, such test results may increase diagnostic accuracy and confidence and may be required in order to document eligibility for state- or school-based services. Therefore, when appropriate, it is recommended that standardized instruments be administered to assess the child's level of development or cognitive ability and current adaptive functioning as part of the diagnostic evaluation process. It is therefore important that familiarity with instruments that assess developmental and adaptive functioning occurs as it is often difficult to obtain a good estimate of cognitive functioning in those with ASD. If standardized instruments are not used to assess developmental level or cognitive ability and adaptive behavior prior to or during the initial diagnostic evaluation, the diagnostician should refer to a qualified professional who can complete cognitive and adaptive testing as part of the assessment for intervention planning or to contribute to intervention planning. Examples of assessments for developmental domains, adaptive functioning, communication and language development, social interaction, and behavior appear in **Appendix D**.

## Comprehensive Medical Examination

A comprehensive medical (physical) examination should be performed either during the initial evaluation for ASD or the diagnostician should refer the child for a comprehensive physical examination soon thereafter. The purpose of the medical examination is to identify co-occurring conditions that may require a thorough assessment and to ascertain whether there is an underlying etiology that explains the ASD symptoms. Accurately identifying co-occurring conditions assists in determining which interventions or treatments are appropriate for the child. The medical exam includes the components which follow. Special attention should address hearing and other sensory screens. Adaptations of traditional evaluation methods may be needed for individuals who lack verbal communication skills or use other alternative forms of communication.

- An assessment of height and weight.
- An assessment of head circumference, as macrocephaly (defined as head size two standard deviations larger than average for age and gender) develops in approximately 20% of individuals with ASD (Fombonne, Roge, Claverie, Courty & Fremolle, 1999). A more recent study indicates that boys with regressive autism have typical head circumference at birth but an enlarged head by four to six months of age (Nordahl et al., 2011).
- A dysmorphology examination to look for developmental anomalies or unusual features (facial, limb, stature, etc.). The brief Autism Dysmorphology Measure (Miles et al., 2008) has been developed for use by non-dysmorphologists who evaluate children with autism.
- An examination of the skin, including Woods lamp examination, to determine whether there are hyperpigmented or hypopigmented lesions that may suggest a diagnosis of Tuberous sclerosis complex or neurofibromatosis type 1.
- An assessment of motor development and coordination to determine if there is evidence of subtle neurological findings or “soft signs,” although the literature regarding the validity of these findings is mixed. These soft signs may be exhibited by signs such as toe walking early in life, difficulty with rapidly alternating movements, or general clumsiness.
- A standardized hearing screening or diagnostic hearing test (e.g., brainstem auditory evoked otoacoustic emissions tests) and a full evaluation, if indicated, to determine if symptoms are consistent with a hearing loss rather than a social communication deficit, or if ASD and hearing loss are co-occurring (Filipek et al., 1999). The screening or diagnostic exam should occur even if the child’s newborn hearing screening results were normal since acquired hearing loss can occur after birth. This is consistent with the Connecticut Birth to Three Autism Guidelines (2011) and the Connecticut State Department of Education’s Guidelines for Identification and Education of Children and Youth with Autism (2005).
- Vision screening tests performed by the primary care provider to identify any impairments, in particular for amblyopia, also known as “lazy eye” (United States

Preventive Services Task Force, 2011). Examples of tests that might be performed include: visual acuity test; stereoacuity test; autorefraction; Hirschberg light reflex test; and cover-uncover test. Creedon (2006) suggests additional tests that specialists can perform for individuals with autism who need vision screening, including tests that do not require verbal communication on the part of the person having the test, in such cases, a referral to the appropriate physician should be made.

## Associated Medical Conditions

Part of the comprehensive medical exam should include an assessment as to whether the child has any associated medical conditions. The way the individual with ASD expresses symptoms can make it difficult for the diagnostician to identify a medical or health related problem accurately. For example, children with ASD may not indicate pain or discomfort from medical problems in the same way as typically developing children. Rather children with ASD may display outbursts or self-injurious behavior. Thus, the diagnostician must be thorough in reviewing possible co-occurring conditions that are commonly experienced by people with ASD. Where needed, the clinician should refer to a specialist for further testing for a more in-depth assessment of the cause of symptoms and behaviors.

**Seizure disorders.** Seizures are caused by abnormal cerebral electrical activity that varies in type and duration. Although they may occur at any age, seizures typically present in a bimodal fashion, either before 5 years of age or after age 10 (Bolton et al., 2011; Minshew, Sweeney, Bauman, & Webb, 2005). Further, seizures may be associated with significant language and cognitive impairments. Seizures should be evaluated and treated by neurologists. Epilepsy is a specific seizure disorder that is a brain disorder characterized by a history of at least one seizure and the potential for recurrence of seizures (Fisher et al., 2005). An electroencephalogram (EEG) can detect if a person is experiencing seizures and a head computed tomography (CT) or magnetic resonance imaging (MRI) may be useful for further evaluation. The co-occurrence of epilepsy and ASD is more likely in individuals who also have an intellectual disability, and is thought to occur in 20% to 33% of children with ASD (Shea & Mesibov, 2005). Landau-Kleffner Syndrome, also known as acquired epileptiform aphasia, is an epilepsy syndrome in which a person has typical language development which is followed by a progressive loss of receptive and expressive language concomitant with abnormal electroencephalography (EEG) findings (Khan & Al Baradie, 2012). Landau-Kleffner Syndrome is considered to be a very rare disorder, although an exact prevalence is unknown (Simpson, 2013; Stefanatos, Kinsbourne & Wasserstein, 2002).

**Sleep disturbances.** This may include insomnia, sleep disordered breathing, or parasomnias. Sleep disturbances occur in 50% to 80% of all children with ASD (Reynolds & Malow, 2011). Identification of a child's sleep patterns may warrant a sleep study which can be ordered by the pediatrician.

**Gastrointestinal disorders.** This may include constipation, diarrhea, gastroesophageal reflux disease (GERD), or irritable bowel syndrome (IBS). A multidisciplinary panel of experts

reviewing evidence gathered to date about ASD and gastrointestinal (GI) symptoms found the prevalence of GI symptoms in individuals with ASD unclear, with a range from 9 to 70% (Buie et al., 2010). Further, findings indicated that individuals with and without ASD experienced similar GI symptoms. The panel concluded that the same workup that is provided to individuals without ASD also be provided to individuals with ASD (Buie et al., 2010). Importantly, another study did not find statistically significant differences between the overall incidence of GI symptoms among individuals with ASD compared to a control group, although children with ASD did experience more constipation and more feeding issues/food selectivity (Ibrahim, Voigt, Katusic, Weaver, & Barbaresi, 2009).

**Feeding/Eating disorders.** Children with ASD often have restricted eating preferences which may cause nutritional deficiencies. Another feeding/eating problem that is found among some children with ASD is pica. Pica is the persistent mouthing of fingers or objects, which requires monitoring of blood lead levels, particularly in young children. Approximately 10% to 32% of typically developing children between one to six years of age experience some form of pica. The literature on the co-occurrence of ASD and pica is sparse, however, one study reported that among 70 children with autism, 60% had experienced pica behaviors, with the behaviors of nine children considered to be serious (Kinnell, 1985).

## Laboratory Tests

A synthesis of the information gathered from the child and family during the medical exam will inform which laboratory tests, if any, should be conducted. In some cases, it may be possible to determine if there is a known etiology to the presenting symptoms or if multiple conditions are evident. Several sets of medical guidelines have been issued by various groups including the American Academy of Pediatrics, the American Academy of Neurology, and the American College of Medical Genetics describing the laboratory testing that is indicated in the evaluation of the child with ASD. The technology to identify small abnormalities in the genome is rapidly changing and evolving and the expected yield from diagnostic studies is anticipated to increase over time. Following is an overview of current recommended laboratory, neuroimaging and other diagnostic tests.

- For children with signs of seizures and for those with language regression, an electroencephalography (EEG) is recommended (Miles, McCathren, Sticher, & Shinawi, 2010).
- For children with a head circumference 2.5 times greater than the age appropriate mean, phosphatase and tensin homolog (PTEN) gene testing is recommended (Schaefer, Mendelson, et al., 2008).
- For some children with specific clinical indicators such as macrocephaly, microcephaly, seizures, or an abnormal neurological exam, a brain magnetic resonance imaging (MRI) may be indicated (Schaefer, Mendelson, et al., 2008).
- Targeted studies such as metabolic testing should be considered when specific symptoms are reported such as cyclic vomiting, or lethargy with minor illness.



- A recent consensus statement issued by the American College of Medical Genetics have recommended that chromosomal microarray should be the first line evaluation for children with developmental disability, ASD, and congenital malformations due to a higher diagnostic yield excluding those children with recognizable chromosomal syndromes, such as Down syndrome (Miller et al., 2010). Chromosomal microarray is a genome-wide assay that examines the chromosomes for tiny, sub-microscopic deletions or duplications of DNA sequences. Balanced rearrangements may not be detected by this methodology (<1% of the time).

## Differential Diagnoses and Co-Occurring Conditions

In making a diagnosis of ASD, it is important to distinguish ASD from other disorders that may present similarly. The *DSM-5* (APA, 2013) provides specific guidance on such differential diagnosis between ASD and Rett syndrome, Selective Mutism, Language Disorder and Social (Pragmatic) Communication Disorder, Intellectual Disability, stereotypic movement disorder, Attention-Deficit/Hyperactivity Disorder, and Schizophrenia. In addition to the medical conditions that can co-occur with ASD, it is important to understand that having ASD does not preclude a child from having additional co-occurring conditions, including some conditions from which ASD must be differentiated. In fact, individuals with ASD often experience conditions that are associated with ASD and may have a higher rate than the typical population for some conditions (Bolton, 2009; Levy et al., 2010). It can also be useful to understand if an etiological factor is present. Information on differential disorders, co-occurring conditions, environmental factors, and etiological factors follow and these are listed on Table 8.

### Neurodevelopmental Disorders

**Attention-Deficit/Hyperactivity Disorder (ADHD).** Attention-Deficit/Hyperactivity Disorder (ADHD) is characterized by inattention, hyperactivity, and/or impulsivity that interfere with a person's functioning or development (APA, 2013). While social difficulties can be present in children with ADHD without ASD, the social impairment difficulties seen in ADHD are related primarily to impulsivity and executive functioning challenges of the disorder, not from the primary social and communication deficits seen in ASD. While the *DSM-IV* did not permit the diagnosis of ASD to be co-occurring with ADHD, this has been reversed in *DSM-5* and ADHD can now be used as a specifier for ASD. Because inattention, hyperactivity, and impulsivity can all be symptoms of ASD, ADHD should only be used as a specifier "when attentional difficulties or hyperactivity exceeds that typically seen in individuals of comparable mental age," (APA, 2013, p. 58). The prevalence of ADHD is estimated to be about 8% (American Academy of Pediatrics, 2011) with up to 40% of children with ASD also having symptoms of ADHD (Zeiner, Gievik, & Weidle, 2011).

**Intellectual Disability.** Intellectual disability (formerly referred to as mental retardation) is characterized by deficits in intellectual (cognitive) functioning as well as deficits in adaptive functioning, including conceptual, practical and social skills that are manifested during the developmental period. The common features of ASD and intellectual disability include lack of visual or auditory responses, lack of language and motor delays, behavior problems, and limitations in adaptive behaviors (APA, 2013). Although children with intellectual disability may show social-communicative deficits, these skills are commensurate with their cognitive level whereas in children with ASD there is a discrepancy between an individual's social competence and cognitive abilities. The prevalence of intellectual disability is estimated to be about 1% (Maulik, Mascarenhas, Mathers, Dua, & Saxena, 2011), with up to 50% of people with ASD also having an intellectual disability or global developmental delay (Johnson & Myers, 2007; CDC, 2012a).

**Language Disorder.** Language Disorder is characterized by difficulties in acquiring and using language because of impairments in comprehension or production of language (APA, 2013). Communication difficulties are also seen in children with ASD and Language Disorder, and as described in *DSM-5* (APA, 2013), can co-occur with ASD. Children who have Language Disorder without ASD may have social interaction problems because of their difficulties in understanding or using language, but do not show restricted interests and repetitive behaviors. The prevalence of language disorders in children without ASD has been estimated to be about 6% (Law, Boyle, Harris, Harkness, & Nye, 2000).

**Social (Pragmatic) Communication Disorder (SCD).** SCD is a new diagnostic classification in the *DSM-5* (APA, 2013) and describes a communication disorder in which individuals with social aspects of language such as initiating conversation (talking too much or to everyone), maintaining coherence, following rules of conversation and storytelling, understanding and matching context, and understanding communicative intent that is not explicitly stated. The onset of symptoms must occur in the early developmental period, although symptoms might not become fully manifested until social demands exceed limited capacities and cannot be attributed to another medical or neurological condition (including ASD, intellectual disability, or ADHD) or to low abilities in the domains of word structure and grammar. SCD cannot co-occur with ASD. These symptoms occur in the absence of restricted, repetitive patterns of behavior, which differentiates SCD from ASD. Because it is a new disorder, estimates of the prevalence cannot be made at this time.

**Stereotypic Movement Disorder.** Stereotypic Movement Disorder is characterized by repetitive movements with onset in the developmental period that appear to lack a purpose (e.g., arm flapping, head banging), might cause bodily harm, and interfere with everyday functioning (APA, 2013). Both individuals with Stereotypic Movement Disorder as well as those with ASD may have repetitive behaviors, such as rocking, hand shaking or waving, and head banging. However, those with Stereotypic Movement Disorder alone do not have the social interaction and communication deficits that are characteristic of ASD. Stereotypic Movement Disorder can co-occur with ASD but is only diagnosed in ASD when there is self-injurious behavior (APA, 2013). An estimated 7% of children have Stereotypic Movement Disorder (Zinner & Mink, 2010).

**Tourette's disorder.** Tourette's disorder is a tic disorder, that is characterized by motor and vocal tics that may occur simultaneously or at different times with onset prior to age 18 (APA, 2013). Repetitive, stereotypic movements or vocalizations are one of the core features of Tourette's disorder that may also be exhibited by individuals with ASD, however, onset of Tourette's disorder is typically after a child is six-years-old (Burnette & Singer, 2007) and the social interaction deficits characteristic of ASD are not found in individuals with Tourette's disorder. The prevalence of a lifetime diagnosis of Tourette's disorder has been estimated at less than 1% of children (Scahill, Bitsko, & Blumberg, 2009).

## **Mental/Behavioral Disorders**

**Anxiety disorders.** Anxiety disorders are characterized by persistent and “excessive fear and anxiety and related behavioral disturbances” (APA, 2013, p. 189). Children with anxiety disorders who do not have ASD are avoidant in particular situations, but capably display social interaction and communication skills among people with whom they are familiar. This is in contrast to children with ASD who, although they often have differential patterns of responses depending on the setting, will display atypical patterns in all social interactions. Data on the prevalence of anxiety disorders for adolescent-aged children is estimated to be about 13% (Costello et al., 1996) and the co-occurrence of anxiety and ASD has been shown to be about 40% (Siminoff et al., 2008).

**Depressive disorders.** Depressive disorders in children (ages 6 to 12 years) may be characterized by irritability, anxiety, sleeping and behavior problems, whereas adolescents (ages 13 to 18 years) may express feelings of hopelessness and guilt (Birmaher et al., 2007). Distinguishing between children who are depressed and children who have ASD requires an assessment as to whether symptoms such as social withdrawal are the result of general sadness that is experienced by the child (depression) or the result of a deficit in social communication skills (ASD). The prevalence of depression in children (ages 6 to 12 years) is estimated to be 2% and in adolescents (ages 13 to 18 years) between 4 to 8% (Birmaher et al., 2007). The rate of depression in individuals with ASD has been estimated to be between 2 to 38% (Magnuson & Constantino, 2011), with the highest co-occurrence seen in individuals who are considered higher-functioning.

**Obsessive Compulsive Disorders (OCD).** OCD are characterized by obsessions or compulsions that take up a considerable part of an individual's day or cause significant distress or impairment (APA, 2013). The diagnostician must determine whether the presenting symptoms are consistent with obsessions and compulsions and thus are indicative of OCD or whether they are restricted and repetitive interests, behaviors, or activities that are consistent with the core features of ASD. In children with OCD alone, impairments in social interactions and communication are usually not present, whereas these two areas are impaired in children with ASD. Estimates of prevalence of OCD are about 1% in children and up to 2% of adolescents (Flament et al., 1988). Studies have shown 2% to 81% of children with ASD have a co-existing diagnosis of OCD (Leyfer et al., 2006).

**Oppositional Defiant Disorder (ODD) and Conduct Disorders (CD).** ODD is characterized by “a pattern of angry/irritable mood, argumentative/defiant behavior, or vindictiveness” which may include losing one’s temper, arguing with adults, refusing to comply with requests or rules, often being angry, resentful or vindictive (APA, 2013, p. 462). CD is considered to be more severe in that the rights of others or major social norms are violated through aggression, property destruction, deceitfulness or theft, or serious rule violations (APA, 2013). Common overlapping behaviors among people with ODD, CD and ASD include difficulties with social interactions, but children with ODD and CD without ASD often do not present the restrictive and repetitive behaviors associated with ASD. The prevalence of an individual having ODD over a lifetime is estimated at 10%, based on retrospective self-report (Nock, Kazdin, Hiripi, & Kessler, 2007). The co-occurrence of ODD symptoms among children with ASD has been estimated to 13% to 26% (Gadow, DeVincent, & Drabick, 2008).

**Personality disorders.** Personality disorders are described as “an enduring pattern of inner experience and behavior that deviates markedly from the expectations of the individual’s culture” (APA, 2013, p. 646). Both people with personality disorders and those with ASD have behavior problems that interfere with their relationships. Individuals with personality disorders such as schizoid and schizotypal personality disorders have difficulties with social interactions but do not have communication impairments or repetitive behaviors, which are evident in individuals with ASD. The lifetime prevalence of personality disorders is estimated to be 9% (Lenzenweger, Lane, Loranger, & Kessler, 2007).

**Reactive Attachment Disorder (RAD).** Reactive Attachment Disorder is used to describe children who have not developed appropriate attachment behaviors related to comfort, support, protection, and nurturance, often due to neglect, abuse, or an otherwise unstable caregiving environment (APA, 2013). Children with Reactive Attachment Disorder may present with disinhibited social interactions and indiscriminate sociability that is disproportionate to the relationship, which is also seen in ASD. The differential features between Reactive Attachment Disorder and ASD include a history of neglect and the absence of restricted and repetitive behaviors. Reactive Attachment Disorder cannot co-occur with ASD. Prevalence of Reactive Attachment Disorder is not known but considered to be very rare (APA, 2013).

**Schizophrenia.** Schizophrenia is characterized by the presence of delusions, hallucinations, disorganized speech, disorganized or catatonic behavior, and negative symptoms (APA, 2013). Behaviors that are common between Schizophrenia and ASD include social isolation, idiosyncratic preoccupations, and flat affect. Schizophrenia can be distinguished from ASD in that individuals with Schizophrenia exhibit paranoid ideation (belief that one is being harassed or persecuted, having suspicions of others’ motives), which is not usually present in individuals with ASD, and onset of Schizophrenia typically does not occur until adolescence or early adulthood. The lifetime prevalence is estimated to be less than 1% (APA, 2013).

**Selective Mutism.** Selective Mutism describes a disorder in which a person does not communicate in particular settings, but exhibits intact communication skills in other settings (APA, 2013). Onset of Selective Mutism typically occurs before age five but may not be recognized until the child begins school. Selective Mutism is often accompanied by excessive shyness and fear of social embarrassment, which may present as atypical or withdrawn social interaction. In comparison, children with ASD may not communicate because of the core features of the disorder, which may pervade all interactions, not particular ones as in Selective Mutism. Another distinguishing factor is that children with Selective Mutism do not have restricted or repetitive behaviors. The prevalence of Selective Mutism is thought to be between 1- and 2% (Viana, Beidel, & Rabian, 2009).

## Genetic Etiologic Factors of ASD

**Angelman syndrome.** Communication deficits, hand-flapping, motor hyperactivity, sudden bursts of laughter, seizures, facial dysmorphism, and intellectual disability are core features of Angelman syndrome (Cohen et al., 2005). Children with Angelman syndrome enjoy being around other people and demonstrate a desire to communicate with others and display this in a variety of ways including the use of non-verbal gestures. Angelman syndrome is caused by the deletion of the maternally derived UBE3A, gene which is located on chromosome 15. Approximately 80 to 100% of people with Angelman syndrome also have ASD and 1% of people with ASD have Angelman syndrome (Cohen et al., 2005).

**Cornelia deLange syndrome.** Individuals with Cornelia deLange are typically of small stature, have limb abnormalities, and have facial dysmorphism. They may be hypersensitive, impulsive, exhibit self-injurious, aggressive and compulsive behavior, have expressive communication and cognitive deficits (Oliver, Arron, Sloneem, & Hall, 2008). Cornelia deLange syndrome is caused by a mutation in the Nipped-B-like gene (NIPBL) gene, which is linked to chromosome 3q26.3. The physical characteristics of Cornelia deLange syndrome are not present in individuals with ASD, which is how the two conditions are differentiated. The prevalence of Cornelia deLange syndrome is approximately 1 in 10,000 to 1/100,000 (Simpson, 2013). The co-occurrence of ASD among individuals with Cornelia deLange syndrome is estimated between 32% and 62% (Berney, Ireland & Burn, 1999; Oliver et al., 2008; Moss et al., 2008).

**Down syndrome.** Down syndrome, also known as trisomy 21, is a genetic disorder characterized by intellectual disability, language delays, characteristic facial features and weak muscle tone, with increased risk for a variety of medical conditions (Roizen, 2013). Chromosomal analysis can reveal whether a child has Down syndrome, which is caused by the presence of all or part of a third copy of chromosome 21. The prevalence of Down syndrome is about 12/10,000 (Roizen, 2013). Reports of the number of people who have Down syndrome and also have ASD range from 5 to 39% (Moss, Richards, Nelson & Oliver, 2012) and the number of people with ASD who also have Down syndrome ranges from 0% to 17% (Filipek, 2005).



**Fragile X syndrome.** Fragile X is an inherited disorder and the most commonly known inherited cause of intellectual disability characterized by poor eye contact, hand flapping and biting, attention deficits, anxiety, social avoidance, and language delays (Abrams et al., 2011). Mild facial dysmorphology (e.g., long face, large ears, and macroorchidism) is also present in Fragile X. Fragile X is caused by expansion of the CGG trinucleotide repeats and the effect of this is a reduction or elimination of the FMR1 (Fragile X gene) protein. It is estimated that 2.5/10,000 males and 1.25/10,000 females are born with the full mutation for Fragile X (Batshaw, Gropman, & Lanpher, 2013). Approximately 15 to 20% of people with ASD also have Fragile X (Schaefer, Mendelson, et al., 2008) and the co-occurrence of ASD with Fragile X in pre-mutation and full mutation males and females is approximately 35% (Bailey, Raspa, Olmsted, & Holiday, 2008).

**Prader-Willi syndrome.** Some of the features characteristic of Prader-Willi syndrome include speech delays, intellectual disability, repetitive and obsessive behaviors and self-injurious behaviors in addition to an insatiable appetite and obesity beginning in childhood when not controlled (Veltman, Craig, & Bolton, 2005). A deletion on part of chromosome 15 is the cause of Prader-Willi syndrome: either a deletion of 15q11-q13 on the maternally inherited chromosome, a paternal inheritance of both copies of chromosome 15, or a point mutation in the maternal copy of the UBE3A gene. Typically individuals with Prader-Willi syndrome have distinctive facial dysmorphology, which is not present in those with ASD, nor do individuals with ASD have the difficulties during infancy with growth and feeding followed by insatiable appetite as they develop through childhood. Prevalence is estimated at 1/10,000 to 1 in 15,000, of which 25% also have ASD (Veltman et al., 2005).

**Rett syndrome.** Rett syndrome is a condition found nearly exclusively among females and is characterized by typical development very early in life followed by a loss or slowing down of development (APA, 1994). Rett syndrome is caused by a genetic mutation to the methyl-CpG binding protein 2 (MeCP2) gene at Xq28. Stereotypic movements (e.g., hand wringing), intellectual impairment, and motor difficulties are some of the characteristics of Rett syndrome (Van Acker, Loncola & Van Acker, 2005). Early symptoms of Rett syndrome such as language loss and reduced social engagement may initially suggest a diagnosis of ASD, but Rett syndrome can be distinguished by decreasing head growth, hand wringing stereotypies and progressive gait disturbance. If an individual presents symptoms consistent with the *DSM-5* diagnostic criteria for ASD and Rett syndrome, the ASD diagnosis should be given, adding Rett syndrome as a specifier of a known genetic condition (e.g., autism spectrum disorder with Rett syndrome). It is estimated that 1/10,000-20,000 girls have Rett syndrome (Van Acker et al., 2005).

**Tuberous sclerosis complex (TSC).** TSC is a neurological condition characterized by lesions throughout several organs, such as the heart, liver, lungs and skin and ash leaf-shaped depigmented skin spots or macules and facial angiofibromas (Simpson, 2013; Sheehan, 2010). TSC is caused by a defect in the TSC1 or TSC2 gene that codes for a protein called tuberin. People with TSC may have stereotypies, as well as social and communication deficits common in individuals with ASD, and it is estimated that up to 60% of individuals with tuberous sclerosis have ASD (Curatolo, Porfirio, Manzi, & Seri, 2004). Estimates suggest that 1-4% of people with ASD also have Tuberous sclerosis (Hyman & Towbin, 2007).

**Williams syndrome.** Common traits of Williams syndrome are intellectual disability, communication delay in early childhood, extroverted personality, disinhibition in social interactions, and characteristic physical features (Simpson, 2013; Kaufmann, Capone, Carter & Lieberman, 2008). A combination of medical tests can be used to determine if a person has Williams syndrome (blood pressure, blood test, echocardiogram and Doppler ultrasound, and kidney ultrasound). Williams syndrome is the result of a deletion of the 7q11.23 region of chromosome 7 which contains the elastin gene. It is estimated that the prevalence of Williams syndrome is 1/7,500 to 1/20,000 (Simpson, 2013) and has been reported to co-occur with ASD in a small number of case studies (see Tordjman et al., 2012).

## Environmental Factors

**Fetal alcohol spectrum disorder.** Characteristics of fetal alcohol spectrum disorder are delayed development, deficits in executive functioning, impaired social skills and difficulties with adaptive skills (Pei, Job, Kully-Marten & Rasmussen, 2011). Children with fetal alcohol spectrum disorder often have difficulty reading social cues, have poor social judgment and can be indiscriminate in their social approach such that their social behaviors appear atypical (Wyper & Rasmussen, 2011). Children with the full manifestations of fetal alcohol syndrome will have, in addition to current or past growth abnormalities and functional impairment in neurodevelopment, the hallmark facial abnormalities, including small palpebral fissures (smaller eye openings), a smooth and often elongated philtrum (an underdeveloped groove between the nose and the upper lip) and a thin upper lip; children with ASD alone do not have these dysmorphic characteristics. Generally, it is reported that up to 30/10,000 infants are born with a pattern of physical, developmental, and functional problems indicative of fetal alcohol spectrum disorder (Chudley et al., 2005).

**Phenylketonuria (PKU).** Infants born with PKU appear typically developing at birth, but soon thereafter have difficulty feeding, and may experience vomiting and appear irritable (Clements, 2010). If the condition goes undetected and untreated, infants will not have an increase in head circumference and they will have spasms and abnormal EEG findings. Severe intellectual disability will result (Clements, 2010). PKU has been known both as a cause of ASD as well as a co-occurring condition. If identified at an early age, PKU can be treated and controlled with diet. The prevalence of PKU has been estimated to be 1/25,000 (American College of Medical Genetics, 2005), with early estimates of the co-occurrence of PKU and ASD at about 5% (Baieli, Pavone, Meli, Fiumara, & Coleman, 2003).

## Other Conditions

**Blind or visual impairment.** Blindness is “visual acuity of 20/200 or worse in the better eye with correction, or a visual field that subtends to an angle of not greater than 20 degrees instead of the usual 105 degrees” (Geddie, Bina, & Miller, 2013, p. 182). Features indicative of untreated visual impairment in young children include abnormal movement of the eyes, eyes that look in only one direction, eyes that do not react to stimuli or habitual pressing of the eyes (Geddie et al., 2013). ASD is the most common developmental disorder among

children who are blind with estimates of 31% of children with limited or no functional vision also having ASD (Parr, Dale, Shaffer, & Salt, 2010).

**Deaf or hard of hearing.** Those who are deaf or hard of hearing experience loss of hearing that may be mild, moderate, severe or profound and may be temporary or permanent (Bueth, Vohr, & Herer, 2013). Features of hearing loss in an infant include not awakening in reaction to loud noises and reduced, delayed or absent babbling by six months and later poor speech intelligibility (Bueth et al., 2013). Children with hearing loss may appear to be uninterested in social interactions or have limited social skills due to an inability to attend to what is being said. This is in contrast to children with ASD, who demonstrate impairments in social interaction and communication, often showing little or atypical interest in social interaction. Approximately 14/10,000 babies in the U. S. are born each year with hearing loss (CDC, 2012b), with profound deafness occurring in 4-11/10,000 school aged children. There is a higher than expected prevalence of ASD among children with hearing impairment (2%; Szymanski, Brice, Lam, & Hotto, 2012), and it is estimated that about 8% of children with ASD have mild to moderate hearing loss (Rosenhall, Nordin, Sandstrom, Ahlsen, & Gillberg, 1999).

**Mitochondrial disorders.** Mitochondrial disorders describe disorders that are caused by abnormal functioning of the mitochondria (energy producers of the cell) or mitochondrial metabolism. The diverse group of disorders often shares several features: drooping eyelid (ptosis), short stature, paralysis of external eye muscles and hypothyroidism (Simpson, 2013, p. 783). A review of studies looking at ASD and mitochondrial disease found that there are common characteristics among children with ASD that may indicate a need for further testing to determine if mitochondrial disease is co-occurring. Mitochondrial disorders are estimated to affect approximately 5/10,000 (Schaefer, McFarland, et al., 2008). Children with ASD with a history of regression and multi-organ system involvement have been shown to be at greatest risk for a co-occurring mitochondrial disorder (Haas, 2010).

**Regulation disorders of sensory processing.** Regulation disorders of sensory processing are exhibited by unusual reactions to sensory stimulation (e.g., an unusually high threshold for pain, high sensitivity to auditory and visual stimulation, odors, or textures; Zero to Three, 2005). Sensory features are commonly described as hyper-responsiveness, hypo-responsiveness, and sensory seeking (Baranek, 2002; Ben-Sasson et al., 2008; Liss, Saulnier, Fein, & Kisbourne, 2006). Brock et al. (2012) report a high prevalence of both hyper- and hypo-responsiveness in ASD, sometimes in the same child. Children who have regulation disorders of sensory processing without ASD will not exhibit the social communication difficulties that are a core feature of ASDs. Hyper- or hypo-responsiveness to sensory input or unusual interest in sensory aspects of the environment was added as a diagnostic symptom in *DSM-5*, but is not required for a diagnosis of ASD (2 of 4 restricted, repetitive behaviors are necessary, of which one can be the sensory symptom). Approximately 10 to 17% of children without ASD are over responsive to sensory input (Green, Ben-Sasson, Soto, & Carter, 2011), and estimates of sensory over responsivity in ASD have been estimated to be as high as 90% (Gomot & Wicker, 2011).

## Additional Considerations for the Diagnostic Evaluation

When conducting a diagnostic evaluation, there are additional considerations that should be taken into consideration, both in regard to the child and the diagnostician.

### Considerations for Children

**Very young children.** The evaluation of very young children, those less than 24 months old, presents particular challenges for diagnosticians and families, as these children display symptoms that may be more subtle and more difficult to distinguish from other developmental delays or even typical development (Zwaigenbaum et al., 2005; Rogers, 2009). When giving a diagnosis to a very young child, a follow-up evaluation may be needed, as the stability of early diagnoses have shown some individuals might not meet diagnostic criteria later in life (Zwaigenbaum, et al., 2009). Research conducted prior to the official release of the *DSM-5* indicates that the *DSM-5* criteria for ASD may be more difficult to meet than were the criteria under the *DSM-IV* for young children (Barton, Robins, Jashar, Brennan, & Fein, 2013; Worley & Matson, 2012). It will be important to track the implications of the new criteria on the diagnosis of young children.

**Older children, adolescents, and adults.** There are some school age children who perform well academically and therefore may not come to the attention of their teachers or parents. Some of these children may display social communication problems, experience social isolation, loneliness, be rejected socially by their peers, and/or have highly intense preoccupying interests. There are some older children, more specifically, adolescents, who may come to the attention of professionals when they experience significant anxiety or depression related to unsuccessful attempts at social engagement. It is recommended that these children have a clinical diagnostic evaluation. The signs and symptoms exhibited by children ages 11 and older can be found in **Appendix E**.

**Child's gender.** The prevalence of ASD in girls is one fourth the rate experienced by boys in the US (CDC, 2012a). In fact, research has found that even when symptoms are equally severe, boys are more likely to be identified with ASD than girls (Russell, Steer, & Golding, 2011). There is evidence to indicate that among children up to age eight, girls are diagnosed later than boys (6.1 years for girls and 5.6 years for boys; Shattuck et al., 2009). Therefore it is important that families and diagnosticians not rule out ASD as a possibility simply because the child is female.

**Child's race and ethnicity.** Reports of disparities in the rates of diagnosis of children by race and ethnicity in the United States show that Black and Hispanic children are diagnosed at a lower rate than White, non-Hispanic children (CDC, 2012a). Data from Connecticut's Birth to Three System and the Connecticut State Department of Education mirror the state's racial and ethnic population overall, indicating no disparities in the identification of children with ASD who receive early intervention services through Birth to Three and supports and

services through special education (Connecticut State Department of Education, personal communication, May 11, 2012). It is not possible from Connecticut's data to determine whether clinical diagnostic data are also proportional. Therefore, it is critical that parents, providers and educators remain vigilant in ensuring that all children, regardless of race, ethnicity or socioeconomic status are diagnosed early and provided with the individualized services that will result in optimal outcomes.

**Child's language.** Disparities in accessing medical care are generally found when families speak a language other than English. Providers can support families by ensuring access to professional interpreter services (e.g., Language Line Services). Further, careful attention should be paid to children who are English language learners as professionals may make assumptions about a child's communication abilities because English is the child's second language.

## Considerations for the Diagnostician

**Knowledge and skills.** Diagnosing ASD in children is challenging. Currently, there are no medical tests or procedures to definitively diagnose this complex neurodevelopmental disorder. As previously stated, ASD is characterized by a heterogeneous group of behaviors of varying severity, causing varying types and degrees of impairment. In addition, the research over the past several decades has evolved in its understanding of the core features of the disorder, how the disorder is expressed, and when the core features are first expressed (Karmel et al., 2010). These factors make it essential that clinicians acquire sufficient training and experience with children diagnosed with ASD, and the current diagnostic systems in use. Lastly, diagnosticians must also be knowledgeable of the systems of services and supports available to children with ASD and their families. Diagnosticians may need to refer a child to appropriate programs and services for interventions that are evidence-based, and to refer families to available family and medical support services.

A diagnostic evaluation may be conducted independently by a clinician or by a multidisciplinary team that could include to a developmental behavioral pediatrician, a neurologist, a psychiatrist, a psychologist, an advanced practice registered nurse, a clinical social worker, a speech-language pathologist, an occupational therapist, a physical therapist, a board certified behavior analyst, or an educator/special educator, or any combination. Importantly, if the diagnostic evaluation is conducted by one clinician, it must be within the scope of his or her practice. Regardless of the diagnostician's discipline, it is critical that the professional conducting the evaluation has extensive experience evaluating children with developmental disabilities and specifically children with ASD. That experience should include didactic learning regarding ASD, familiarity with methods and instruments to diagnose ASD, repeated opportunities to screen and evaluate a broad range of children with ASD, and training by professionals in the field as described below.

According to CT state statute (Conn. Gen. Stat. § 38a-514b, see Text Box 1, page 39), families with children who are diagnosed by a licensed physician, psychologist or and clinical social worker may seek to use their insurance to pay for autism services; other types



of professionals may not diagnose autism if the family wants to bill insurance for services. This requirement does not apply for families enrolled in employer funded self-insured plans, to families with children enrolled in Connecticut's Medicaid program called HUSKY A (Health insurance program for Uninsured Kids and Youth), nor to families who choose to pay for ASD services out of pocket. Notably, the three types professionals listed are not the only clinicians who are competent to diagnose a child accurately.

**Didactic and continuous learning.** Didactic learning is critical in understanding the core features of ASD, in assessing its manifestations and in differentiating it from other conditions. Diagnosticians must also remain abreast of the rapidly expanding literature on ASD. To remain proficient in the field, diagnosticians must be lifelong learners who view ongoing professional development as a critical component in the provision of evidence based care to children and families in a markedly changing field. Diagnosticians must also be familiar with recommendations provided by national groups related to ASD. For example, the AAP has published recommendations on identifying and evaluating children with ASD that provides a detailed surveillance and screening algorithm with which all diagnosticians should be familiar (Johnson & Myers, 2007 and reaffirmed 2010). The algorithm is included in **Appendix A**.

**Exposure to standardized instruments.** There are a number of standardized instruments that can be useful in determining whether a child has ASD and a list of some of these instruments is provided in the section on Standardized Diagnostic Instruments for Autism. Again, it is expected that a professional would become familiar with these instruments as they assist in collecting information about a child's symptoms and behaviors using a standardized methodology. As stated earlier, research has shown that using standardized behavioral observation instruments increases diagnostic accuracy in clinical settings (Risi et al., 2006; Mazefsky, et al., 2013). If the diagnostician finds that a standardized observation instrument is indicated, a diagnostic instrument with good sensitivity (appropriately identifies children who have ASD as having ASD) and good specificity (appropriately identifies children who do not have ASD as not having ASD) should be selected, appropriate to the child's age. Instruments should be administered in accordance with the publisher's instructions and the diagnostician must meet the training requirements set forth by the publisher of the specific instrument being used to ensure optimal validity.

**Repeated opportunities to evaluate children with ASD.** Recognizing and identifying the core features of ASD is achieved by assessing sufficient numbers of children with and without ASD to develop an understanding of the wide spectrum of the disorder. Research suggests that more experienced clinicians make more accurate diagnostic decisions than clinicians with less experience (Volkmar et al., 1994). Multiple opportunities to observe the signs and symptoms of the disorder are needed to diagnose ASD, and this should be done under the supervision of and mentoring by clinicians with significant experience diagnosing ASD. The supervisor/mentor should provide a trainee with a considerable number of opportunities to evaluate infants, children and adolescents who display the range of behaviors and symptoms indicative of ASD. Further, trainees should gain exposure to various types of interventions that

children with ASD receive and the settings in which the services occur (e.g. home, child care, public school, private practice). Finally, the training should also encompass the principles of family-centered care and medical home to ensure that recommended follow-up for the child is appropriately accessible by families and coordinated.

### **A Note about Use of Clinical Judgment**

In cases where a diagnosis of ASD is unambiguous, seasoned diagnosticians may rely upon their own clinical judgment based on current diagnostic criteria (Fein, 2010). While it may be appropriate to use clinical judgment solely to diagnose ASD, a professional's use of clinical judgment does not exempt the diagnostician from preparing a comprehensive report documenting how a child's presenting symptoms, behavior and history are consistent with a diagnosis of ASD. This includes a detailed discussion of how the child is displaying the core symptoms of ASD.

## Text Box 1: Connecticut Health Insurance Legislation

Connecticut's Health Insurance Coverage for Autism Spectrum Disorders Act of 2009 (Conn. Gen. Stat. § 38a-514b) states that insurance coverage for autism spectrum disorder services is provided when diagnosticians with state licensure as a physician, psychologist, or clinical social worker render a diagnosis of an ASD, based on the most recent edition of the American Psychological Association's DSM. This means that if a family is seeking to use their insurance to pay for autism services for children up to age 15, excluding services offered in public schools, the child must have a clinical diagnosis of autism (not just educational eligibility) and it must have been given by one of the types of diagnosticians listed above. The coverage for ASD does not apply to families enrolled in employer funded self-insured plans or to families with children enrolled in Connecticut's Medicaid program called HUSKY A (Health insurance program for UninSured Kids and Youth), nor does it apply to families who choose to pay for ASD services out of pocket. Connecticut's autism insurance statute speaks to a narrow pool of diagnosticians for the purposes of insurance reimbursement for services. For the purposes of these guidelines, we acknowledge that these are not the only clinicians who are competent to diagnose a child accurately.

The 2009 autism insurance legislation was amended in 2013 as Public Act 13-84. The amended legislation requires insurance companies to cover autism services for individuals who had previously been diagnosed with ASD under the *DSM-IV* criteria. In effect, those who met *DSM-IV* criteria for ASD but do not meet *DSM-5* criteria must be provided autism services.

It is important to note that this statute is currently in effect as of the date of the printing of these guidelines. Providers and families should be aware of future changes in insurance legislation.

**Table 7. Essential components of a diagnostic evaluation**

<b>HISTORY</b>	
<b>Obtain and review medical and other available records</b> <b>Conduct a family interview</b>	<b>MEDICAL RECORDS</b> <ul style="list-style-type: none"> <li>▪ Routine well-child care and concerns raised over time</li> <li>▪ Specialist evaluations by medical subspecialists</li> <li>▪ Birth records, including newborn screening results</li> <li>▪ Prior hospitalizations</li> <li>▪ Medical tests and indications for those tests</li> <li>▪ Medication history</li> <li>▪ Prior treatments</li> </ul> <b>PRIOR ASSESSMENTS</b> <ul style="list-style-type: none"> <li>▪ Developmental evaluations or assessments</li> <li>▪ Other evaluations or assessments</li> </ul> <b>EDUCATIONAL RECORDS</b> <ul style="list-style-type: none"> <li>▪ Birth to Three and educational evaluations</li> <li>▪ Current school services and programs</li> <li>▪ Data related to educational progress</li> </ul> <b>FAMILY RECORDS</b> <ul style="list-style-type: none"> <li>▪ Notes and videos</li> </ul>
<b>Conduct a family interview</b>	<b>ESSENTIAL ELEMENTS:</b> <ul style="list-style-type: none"> <li>▪ Reason for referral and referral source</li> <li>▪ Family concerns</li> <li>▪ Birth history, including pregnancy and neonatal course</li> <li>▪ Developmental history, including when milestones were reached</li> <li>▪ Child's medical history</li> <li>▪ Child's early intervention and educational history</li> <li>▪ Family history (medical, psychosocial) and three generation genetic history</li> <li>▪ Assessment of family's strengths and weaknesses</li> <li>▪ Discussion of family's goals</li> </ul>
<b>DIAGNOSTIC ASSESSMENT</b>	
<b>Assess core features of ASD</b>	<b>ASSESSMENT OF QUALITY AND CHARACTERISTICS OF SOCIAL INTERACTION AND COMMUNICATION</b> <ul style="list-style-type: none"> <li>▪ Deficits in social-emotional reciprocity</li> <li>▪ Deficits in nonverbal communicative behaviors used for social interaction</li> <li>▪ Deficits in developing, maintaining, and understanding relationships</li> </ul>

## DIAGNOSTIC ASSESSMENT (CONT.)

### Use autism-specific diagnostic instruments

#### **ASSESSMENT OF RESTRICTED, REPETITIVE AND STEREOTYPED PATTERNS OF BEHAVIOR SEEN IN CHILDREN WITH ASD**

- Stereotyped or repetitive motor movements, use of objects or speech
- Insistence on sameness, inflexible adherences to routines, or ritualized patterns of verbal or nonverbal behavior
- Highly restricted, fixated interests that are abnormal in intensity or focus
- Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment

#### **STANDARDIZED AUTISM-SPECIFIC DIAGNOSTIC INSTRUMENTS**

- Formal parent interview instruments
  - Autism Diagnostic Interview-Revised (ADI-R)
- Formal behavioral observation instruments
  - Autism Diagnostic Observation Schedule, 2<sup>nd</sup> Edition (ADOS-2)
  - Childhood Autism Rating Scale, 2<sup>nd</sup> Edition (CARS2)
  - Social Responsiveness Scale-2 (SRS)

### Additional standardized measures when the diagnosis of ASD is undetermined

#### **ADDITIONAL STANDARDIZED MEASURES PROVIDED IN APPENDIX D**

### Comprehensive medical exam

#### **COMPREHENSIVE MEDICAL EXAM**

- Height, weight and head circumference
- Dysmorphology examination
- Examination of the skin
- Assessment of motor development and coordination
- Hearing screening
- Vision screening or diagnostic

#### **ASSESS CHILD FOR ANY ASSOCIATED MEDICAL CONDITIONS**

- Seizure disorders
- Sleep disturbances
- Gastrointestinal disorders
- Feeding/eating disorders



## DIAGNOSTIC ASSESSMENT (CONT.)

### Differential diagnoses and co-occurring conditions

### Additional considerations for the diagnostic evaluation

#### LABORATORY TESTS

- Electroencephalography (EEG) when child has signs of seizures or language regression
- Phosphatase and tensin homolog (PTEN) gene testing when child's head circumference is 2.5 times greater than age mean
- A methyl CpG-binding protein 2 (MECP2) for girls with regressive features of ASD
- Brain magnetic resonance imaging (MRI) for children with macrocephaly, microcephaly, seizures, or abnormal neurological exam
- Metabolic testing when child has cyclic vomiting or lethargy with minor illness
- Chromosomal microarray

#### CONSIDER DIFFERENTIAL DIAGNOSES AND CO-OCCURRING CONDITIONS

- Neurodevelopmental disorders
- Mental/behavioral disorders
- Medical conditions
- Genetic/Etiological factors
- Environmental factors
- Other conditions

#### CONSIDERATIONS FOR CHILDREN

- Very young children
- Older children, adolescents, and adults
- Child's gender
- Child's race and ethnicity
- Child's language

#### CONSIDERATIONS FOR THE DIAGNOSTICIAN

- Didactic and continuous learning
- Exposure to standardized instruments
- Repeated opportunities to evaluate children with ASD
- Knowledge and skills

**Table 8. Differential disorders and co-occurring conditions**

CONDITION	DIFFERENTIAL DIAGNOSIS	CO-OCCURRING CONDITIONS
<b>Neurodevelopmental Disorders</b>		
Attention-Deficit/Hyperactivity Disorder	•	•
Intellectual Disability	•	•
Language Disorder	•	
Social (Pragmatic) Communication Disorder	•	
Stereotypic Movement Disorder	•	•
Tourette's disorder	•	•
<b>Mental/Behavioral Disorders</b>		
Anxiety disorders	•	•
Conduct Disorder	•	•
Depressive disorders	•	•
Obsessive Compulsive Disorder	•	•
Oppositional Defiant Disorder	•	•
Personality disorders	•	•
Reactive Attachment Disorder	•	
Schizophrenia	•	•
Selective Mutism	•	
<b>Medical Conditions</b>		
Epilepsy		•
Landau-Kleffner syndrome	•	
Neonatal encephalopathy		•
<b>Genetic Conditions</b>		
Angelman syndrome		•
Cornelia deLange syndrome		•
Down syndrome		•
Fragile X syndrome	•	•
Lesch-Nyhan syndrome		•
Prader-Willi syndrome		•
Rett syndrome	•	•
Smith-Lemli-Opitz syndrome		•
Tuberous sclerosis		•
Williams syndrome		•
<b>Environmental Factors</b>		
Fetal alcohol spectrum disorder	•	
Phenylketonuria (PKU)		•
<b>Other Conditions</b>		
Blind or vision impairment		•
Deaf or hard of hearing	•	•
Mitochondrial disorders	•	•
Regulation disorders of sensory processing	•	



# Chapter 3

## Evaluation Results



After completing a clinical diagnostic evaluation for ASD, the lead diagnostician and other members of the multidisciplinary team (as applicable), must review the information gathered and determine the appropriate diagnosis for the child. The results of the child's clinical diagnostic evaluation are then shared with the family/caregiver(s). The team may discuss the results with the family after the final assessment is completed or shortly thereafter, provided there is minimum delay between the final assessment and the visit in which results are communicated. The diagnostician should explain to the family the entire process prior to beginning the evaluation so that expectations about the timeline for providing results are clear from the start. A more thorough written report is provided to the family when the oral results are shared or at a later date.

## **Oral Feedback to the Family**

There are a number of factors professionals must consider when sharing a diagnosis of ASD with families, the most important of which is tailoring the message to the needs of the individual child and family in a sensitive and respectful manner. Communicating results to the family should be a thoughtful process that includes planning on how to deliver the diagnosis, the use of supportive and nonjudgmental verbal and nonverbal language during the delivery, the review of the assessment results in understandable language, the delivery of the diagnosis, and the discussion of next steps. Ample time must be allowed for the diagnostician to be sure the family understands what is being communicated and for the parents to ask questions.

## **Meeting with the Family**

The diagnostic process from the time the family seeks information about their child through the delivery of the diagnosis is never as fast as a family wishes it to be. Professionals should consider that the family may have waited a considerable length of time to have their child evaluated and then diagnosed. It is critical that professionals are mindful of and empathetic to parents as they plan for the meeting, paying particular attention to parents who may have waiting a long time. Research suggests that the less time families have waited for a diagnosis, and the fewer professionals families have seen during the process, the more satisfied they are with the diagnostic process (Goin-Kochel, Mackintosh, & Myers, 2006).

Professionals should communicate diagnostic results to families in an environment that is private, where families can discuss the information and ask questions without interruptions or distractions. For Birth to Three providers and others who conduct assessments in the home, attention should be paid to ensuring the family members are physically comfortable and free from interruptions. In a clinic or other setting outside of the home, it is important to reduce any additional stressors. For example, the arrangement of the room can impact the interaction and alleviate some of the tensions (Nissenbaum, Tollefson, & Reese, 2002). If in a traditional conference room, the diagnostician and diagnostic team (as applicable), should not sit at the head or other side of the table than the family. Rather,



everyone should be seated comfortably as a team with equal investment in the success of the meeting and the child's future.

It is generally recommended that young children are not in the room when the diagnostic results are discussed as they may understand some of what is communicated. However adolescents or adults may want to be involved in the discussion. If a family prefers their child to be present and the clinician prefers to meet with the parents privately, the diagnostician should express the importance of having an open dialogue without filters or constraints. For some families, feedback may need to be given over the course of two sessions, one with the parents and an additional session provided specifically for and with the child or teen.

Meetings with families must be conducted in the families' primary language and arrangements should be made in advance to have interpreters available, if needed. It is recommended that a neutral party and someone other than a family member serve as the interpreter. Under no circumstances should a sibling serve as an interpreter during a meeting with the family to discuss the results of a clinical diagnostic evaluation. Professional interpreter services are available by telephone and can be accessed for a fee.

It is critical for professionals to assess the family's level of awareness and knowledge of ASD and to determine whether the family has considered the possibility of an ASD diagnosis in advance of the meeting. This should have been assessed at the onset of the evaluation process. Not all families that bring their child for an evaluation suspect that their child might have ASD or another type of developmental delay. For some families, receiving a diagnosis of ASD can be overwhelming, especially if they have only been exposed to negative media portrayals. Other families may feel a sense of relief in obtaining the diagnosis because their concerns and questions have been confirmed and identified, and as such, conversations about the diagnosis may be easiest with them (Nissenbaum et al., 2002).

**Intentional use of verbal and non-verbal language.** Professionals should be thoughtful in their choice of words when communicating a diagnosis to parents. The emphasis should focus on the child and his or her individuality, not the diagnostic label of ASD as concerns have been expressed that labeling a child with ASD defines the entirety of the child (Hodge, 2005). For example, people first language should be used in all references to the child (e.g., "child with ASD", *not* "autistic child").

Certain words carry connotations that can mislead families and give the wrong impression. For example, it is preferable for professionals to use words such as "areas of challenge" and "difficulties" which will provide a more positive outlook of the child's prognosis than "deficits" or "impairments". When discussing observations of the child and/or the child's future (prognosis), words such as "concerns" are preferred over those such as "worries," because the term "worries" is perceived as ominous (Glascoe, 2000). Diagnosticians should always use concrete terms (e.g., autism affects social and communication skills and behavior) to describe ASD to parents and families. Thus,

diagnosticians need to be intentional in their use of language while discussing an ASD diagnosis with parents and should use common language that is culturally relevant to the family. Parents should be reassured that ASD is not their fault, and that autism is not a result of poor parenting or anything else they may have done.

The way words are delivered is as important as the words that are chosen; therefore non-verbal communication skills are critical. Reflective listening is especially helpful in discussing diagnostic information because non-verbal communication can intentionally or unintentionally send positive or negative cues. Positive cues show empathy and compassion which can help parents receive the diagnosis and include relaxed body language, leaning in toward the family, nodding appropriately and showing a genuine interest in the child and the family. Negative cues such as typing in an electronic medical record, responding to text messages, taking phone calls, looking frequently at a watch or clock, holding a rigid posture, eating and permitting numerous interruptions are all hindrances to the diagnostic conversation and should be avoided. Negative cues convey a lack of interest, even if it is not the intention. Families deserve the undivided attention of the diagnostician and other team members when discussing results.

## **Communicating the Diagnosis**

When delivering the results of the clinical diagnostic evaluation to families, diagnosticians should be clear and state a diagnosis of ASD using direct and understandable language. Even if it is hard to hear at the time, “sugarcoating” or glossing over a diagnosis may not be helpful to families. In doing this, diagnosticians should pace the meeting with the family, to be sure that the family is assimilating the information being discussed. They should offer to rephrase or repeat information to ensure the family understands the evaluations and conclusions. If a family decides that they need time before taking in more information, diagnosticians should accommodate the needs of the family by making arrangements to reconvene the meeting within a relatively short period of time. It is important that diagnosticians verbally acknowledge the family’s emotions and respond to the family’s verbal and non-verbal cues without judgment, showing both professionalism and empathy. It should never be assumed that families will be either overly emotional or completely unemotional when the diagnosis is given; different people handle important life-changing news in different ways (e.g., grief, anger, shock, frustration). Diagnosticians should not over personalize reactions families may have, but be reflective and work toward maintaining a therapeutic relationship with families.

Some parents may view diagnosticians as powerful people and believe that a diagnosis will change their child’s identity and determine what his/her future development will be (Hodge, 2005). Given that the prognosis for each child with ASD is dependent on a variety of factors, it is important for the diagnostician to be clear about what is known and what is not known about the child’s future.

Parents report that it is critical for diagnosticians to express hope when a diagnosis of ASD is given as it can activate parents to seek care for their children (Nissenbaum et al.,

2002). Hope can be provided to parents by discussing the child's strengths and a range of effective interventions that can help the child reach his/her optimum level of independence. At the same time, it is also important for diagnosticians to refrain from imposing feelings of any kind on families.

There is great variability in the information needs of the family after the diagnosis is given. It is important for diagnosticians to assess what families are ready to hear when communicating results (Osbourne & Reed, 2008). Some families may prefer to receive information in phases. Other families may want a detailed prognosis for the child. They also may want all of the information possible at the time of diagnosis, rather than processing pieces. This can lead to information overload which may overwhelm families, but some families express that they would rather have the information available as it may be needed in the future. It is recommended that when providing families with the written report, it is important to remind them that it includes the same information being shared (e.g., about the diagnosis, recommendations, local resources, family support groups, and how to contact Connecticut Child Development Infoline for further resources).

While some families think that diagnosticians are too negative in delivering and discussing a diagnosis, diagnosticians at times feel that families are too positive when discussing future outcomes for their child, so it is important to find an appropriate balance during the discussion (Nissenbaum et al., 2002). The long term future implications (e.g., about independent living, employment) are not appropriate unless the child is of transition age or older (e.g., age 13 and older). Key in the message to families should be an acknowledgement of the child's and family's strengths, while also recognizing the challenges of parenting a child with ASD. It is appropriate to ask a family at this point how they are coping with their challenges.

**Clear explanation of the diagnostic criteria.** The publication of the *DSM-5* (APA, 2013) might present challenges and confusion for families concerning their child's diagnosis and a potential change in diagnostic classification. It is essential that the diagnostician provide families with a clear explanation of what criteria they used to arrive at an ASD diagnosis. Families might have increased anxiety, especially families of children who currently have a *DSM-IV* (APA, 1994) diagnosis of autism who are worried their child might 'lose' the diagnosis. Diagnosticians should expect that families will have questions about what ASD is, how autism is different now than it was before, and if their child will be entitled to different services and supports. In these cases, the diagnostician should reassure families that their child will not lose their diagnosis and that the *DSM-5* contains specific language to protect this. Diagnosticians might also refer families to appropriate community supports, advocacy organizations, or online information that helps explain the changes that have been made.

## Discussing Next Steps

After talking with families about the diagnosis of ASD, diagnosticians should provide them with recommendations that can guide their next steps. For children under the age of

three, a referral to the early intervention system, Birth to Three, can be made by contacting Child Development Infoline (1-800-505-7000, also see **Appendix F**). For children who are age three or older, a referral for an evaluation by the local school district planning and placement team (PPT) to determine a child's need for special education services should be provided. If a school-age child is already receiving special education services, the parent may wish to request a PPT meeting to consider the information obtained from the clinical evaluation, and, if necessary, to request that the district conduct its own evaluation. Two important resources families may find helpful are the Connecticut State Department of Education's *Guidelines for Identification and Education of Children and Youth with Autism Spectrum Disorders* (2005) and the Connecticut Birth to Three System's *Autism Spectrum Disorders: Intervention Guidance for Parents* (2011). In addition, parents should be referred to the Connecticut Medical Home Initiative which provides care coordination for children and youth with special health care needs age newborn through 21 (see **Appendix F** for details). Finally, the diagnostician should discuss referrals and future visits to follow up on referrals (e.g., visits to specialists).

## The Written Evaluation Report

The report should be written in a manner that fosters collaboration among diagnosticians and parents and ensures optimal outcomes for the child. A thorough and clear written report is critical and should be provided to the child's family within a reasonable timeframe following the evaluation.

### Components of the Report

The report must contain an objective but sensitive discussion of how the core features of ASD are exhibited (or not) by the child. This is essential, even if the diagnosis is based solely on clinical judgment. The written report serves as a means of documenting clinical findings to the family and other diagnosticians (e.g., teachers, school administrators, special educators). The report may also be needed in order to access services for the child. The report's recommendations should serve to unify everyone involved with the child and family and should guide treatment as well as inform other providers such as early intervention and special education professionals.

The report should begin with a statement about the reason for the referral and any pertinent background information, including a developmental and family history. Next, a discussion of any testing performed previous to the evaluation, as well as the results of the review of the child's prior medical, educational, intervention and other records is described. The clinical portion of the report must include a description of the diagnostic process, including any instruments administered and the procedures and personnel involved in conducting the diagnostic evaluation with a clear description of which diagnostic criteria were used to arrive at the diagnosis. The number of times that the child was seen and the overall length of the evaluation should be included. Also vital to include in the clinical

section of the report are the data from the family interview and direct observations of the child to support the diagnosis of ASD. A description of how the child's presenting symptoms, behavior and history meet the current criteria for a diagnosis of ASD should be documented. The report should contain a section describing referrals for services, recommendations for interventions and for further assessment(s), if necessary, resources for parents, and a follow-up plan. The child's strengths should be prominently detailed throughout.

**Observations and concrete recommendations for resources that can lead a family to services.** A diagnostic evaluation is different from an assessment for intervention. The report resulting from the diagnostic evaluation should be individualized to the specific child and should refer to the assessments that were performed. For example, if a diagnostician finds that the child has a particular motivator (e.g., bubbles), that should be included in the report. It is helpful if the diagnostician provides a description of the challenges that need to be addressed (e.g., motor planning, following directions, initiating conversation) to assist those who will plan intervention services for the child and family. A report resulting from a diagnostic evaluation should not prescribe the service providers by name, or number of hours of services a child needs to meet educational and behavioral outcomes. Rather, the report should provide individualized recommendations that come from what the diagnostician has learned about the child from the evaluation.

The recommendations that diagnosticians give to parents are likely to be distributed in multiple venues (e.g. early intervention programs, special education programs, medical providers). It is then up to those who are responsible to plan and deliver intervention services to the child to apply the recommendations. The recommendations should be detailed, address all areas of a child's daily life and include all developmental domains (e.g., academic, social, behavioral). Recommendations may include for example, "the child will need to have a communication system as soon as possible to learn to express wants and needs to others," or "the child would benefit from programs that improve social skills and opportunities with non-disabled peers." The report should conclude with any referrals or consultations the child and family need. Sources of support and information for parents (e.g., parent advocacy groups as well as information and support groups) should be provided.

## **Sharing Diagnostic Information**

During the oral discussion with families, diagnosticians should emphasize the importance of communication and collaboration across those who are and will be helping the child and family. The diagnostician should explain to the family what a signed consent for the release of the written report means, and how they can choose to share the report with others. In particular, sharing the report with the child's pediatrician or medical home should be encouraged. It is also important to emphasize to families that sharing the report will assist with communication and collaboration among the different service agencies and providers as they help their child and them. Families must also know that the report is theirs to share or not share.





## Chapter 4

### The Use of the Evaluation for Early Intervention and Special Education



There are important distinctions between a *clinical diagnosis* of ASD and a determination of *eligibility for special education* because of ASD. Children with a clinical diagnosis of ASD do not automatically receive special education, nor do students who are eligible for special education under the Individuals with Disabilities Education Improvement Act of 2004 (IDEA) within the category of autism have to have a clinical diagnosis of ASD. The *clinical diagnosis* refers to the diagnostic process of identifying how specific symptoms and behaviors are exhibited by a person and the subsequent clinical diagnosis of ASD by a physician, psychologist, or other qualified health or mental health professional. An evaluation determining a student's eligibility for special education is conducted by a team of diagnosticians, including parents, and is conducted strictly for the purpose of identifying whether a child is eligible to receive special education and related services. An evaluation assessing eligibility for special education does not replace a clinical diagnosis of ASD, nor does a clinical diagnosis of ASD determine eligibility for special education. The IDEA and its related regulations are the legal and regulatory bases for early intervention, called Part C, as well as special education in public schools, called Part B.

## **Eligibility Determination for Children Ages Birth to Three**

Part C of IDEA is the federal program administered by states to provide early intervention services for eligible infants and toddlers. The Birth to Three System in Connecticut serves young children newborn up until age three and is currently administered by the CT State Department of Developmental Services (DDS). In Connecticut, all children referred to the Birth to Three System who are 16 months of age or older are screened for ASD. For those with a positive screening, an autism assessment is provided at no cost to the family either through the program that conducted the initial evaluation or by one of the autism-specific early intervention programs. The assessment will determine whether the child receives a diagnosis of ASD. In order for diagnostic assessments performed by diagnosticians outside of the Birth to Three programs to be accepted by Birth to Three, the diagnostician must be a licensed physician, clinical psychologist or clinical social worker and the assessment must meet the minimum standards of this guideline. Children under age three with a *DSM-5* diagnosis of ASD are automatically eligible for early intervention services and, at the discretion of the parent, may enroll in an autism-specific or general early intervention program. A child may receive the diagnosis either prior to or after the referral to Birth to Three.

Services for children with ASD in the Birth to Three System are delivered in accordance with *Service Guideline #1: Autism Spectrum Disorder, Intervention Guidance for Service Providers* (2011). A version especially for parents, is called *Service Guideline #1: Autism Spectrum Disorder, Intervention Guidance for Parents* (2011). Both publications are available on the Birth to Three website ([www.birth23.org](http://www.birth23.org)) or from any Birth to Three program.

## Eligibility Determination for Children Ages Three Through Twenty-One

Part B of IDEA defines children with disabilities as those children, ages 3–21, who have been evaluated by the school personnel and demonstrate developmental, functional, academic and/or behavioral needs that have an impact on the child’s ability to access and participate in their general education and requires special education. In CT, planning and placement teams (PPTs) determine eligibility for special education. PPTs consist of the child’s parents and professionals representing teaching, administrative and related service staff, as necessary. PPTs review referrals to special education, assess whether a child needs to be evaluated, determine what evaluations will be administered, decide whether a child is eligible for special education, and plan an appropriate individualized educational program (IEP) for the student (Connecticut State Department of Education, 2007).

During the course of the evaluation to determine eligibility for special education, educators and related service personnel draw upon information from a variety of sources, including parental report/answers to questionnaires, and ensure that information is documented and carefully considered. If the child is found eligible for special education, the evaluation information must be sufficient to guide the development of an IEP. The IEP addresses the unique needs of the child within the school environment, which may be similar to or different from the needs of the child in other environments. The IEP is developed, reviewed, and revised collaboratively by the PPT at least once annually.

### Autism Defined Under IDEA

A child who is found eligible for special education is classified into one of 13 disability categories, including autism. According to IDEA, autism is defined as:

*(1)(i) Autism means a developmental disability significantly affecting verbal and nonverbal communication and social interaction, generally evident before age three, that adversely affects a child’s educational performance. Other characteristics often associated with autism are engagement in repetitive activities and stereotyped movements, resistance to environmental change or change in daily routines, and unusual responses to sensory experiences. (ii) Autism does not apply if a child’s educational performance is adversely affected primarily because the child has an emotional disturbance, as defined in paragraph (c)(4) of this section. (iii) A child who manifests the characteristics of autism after age three could be identified as having autism if the criteria in paragraph (c)(1)(i) of this section are satisfied. (Individuals with Disabilities Act of 2004, 34 CFR §300.8 (1)(i)-(iii))*

The CT State Department of Education describes the possible impact of ASD on components of the educational process as follows: “Since the central deficits in ASD (i.e., social reciprocity and interaction, communication, and repetitive behaviors) affect components that are key to the educational process, ASD may adversely impact a child’s performance

in one, several, or all of the following areas: academics, social/emotional growth, life-skills acquisition, communication, and the ability to use and maintain skills across a range of applications and settings” (Connecticut State Department of Education, 2005, page 15).

IDEA mandates that consideration be given to the results of evaluations from outside providers, but PPTs are not required to adopt the recommendations from independent evaluators or use their evaluation information in determining a child’s eligibility for special education and/or in the development of a student’s educational program. Sometimes this statement is misinterpreted to mean that educators do not have to accept the diagnosis of ASD or other conditions. It is not the role of educators to challenge a clinical diagnosis. The decision about a child’s eligibility for special education is based upon a comprehensive evaluation of the child to determine if: 1) the child is a child with a disability; and 2) whether the child requires special education.

## **Developmental Delay**

In some instances, a young child will have a clinical diagnosis of ASD and subsequently will be evaluated by the school district PPT. The PPT may determine that the child is eligible for special education because of developmental delay rather than ASD. It is critical to understand that just because a child falls under developmental delay for special education eligibility does not negate the child’s clinical diagnosis of ASD. The disability category of developmental delay may apply to children from age three to six years. By their sixth birthday, children who continue to require special education services must be re-evaluated to determine if a disability that requires special education continues to exist and to identify a disability category other than developmental delay.

## **Summary**

This section distinguished between two separate processes: the process of obtaining a clinical diagnosis of ASD and the process of determining eligibility for early intervention services and for special education. The challenge is to achieve an optimal level of collaboration and communication between the family and the educational, medical and other diagnosticians and agencies involved in the clinical diagnosis and in the determination of eligibility for special education services. Parents are central in this process and are encouraged to collaborate with the medical and educational diagnosticians involved with their child by sharing results from clinical diagnosticians with schools and to share school evaluations with their child’s clinicians. Any parents with children eligible for special education because of autism are advised, if their children do not have a clinical diagnosis of ASD, to share the educational evaluation with the child’s pediatrician who should refer the child for a clinical evaluation which may be covered by insurance (see text box 1, page 39). School district personnel are separate from the clinical diagnostic process, they are not clinical personnel, and as members of the PPT are qualified to address educational interventions. Pediatricians and other clinical providers may have other recommendations that the school district may or may not address.



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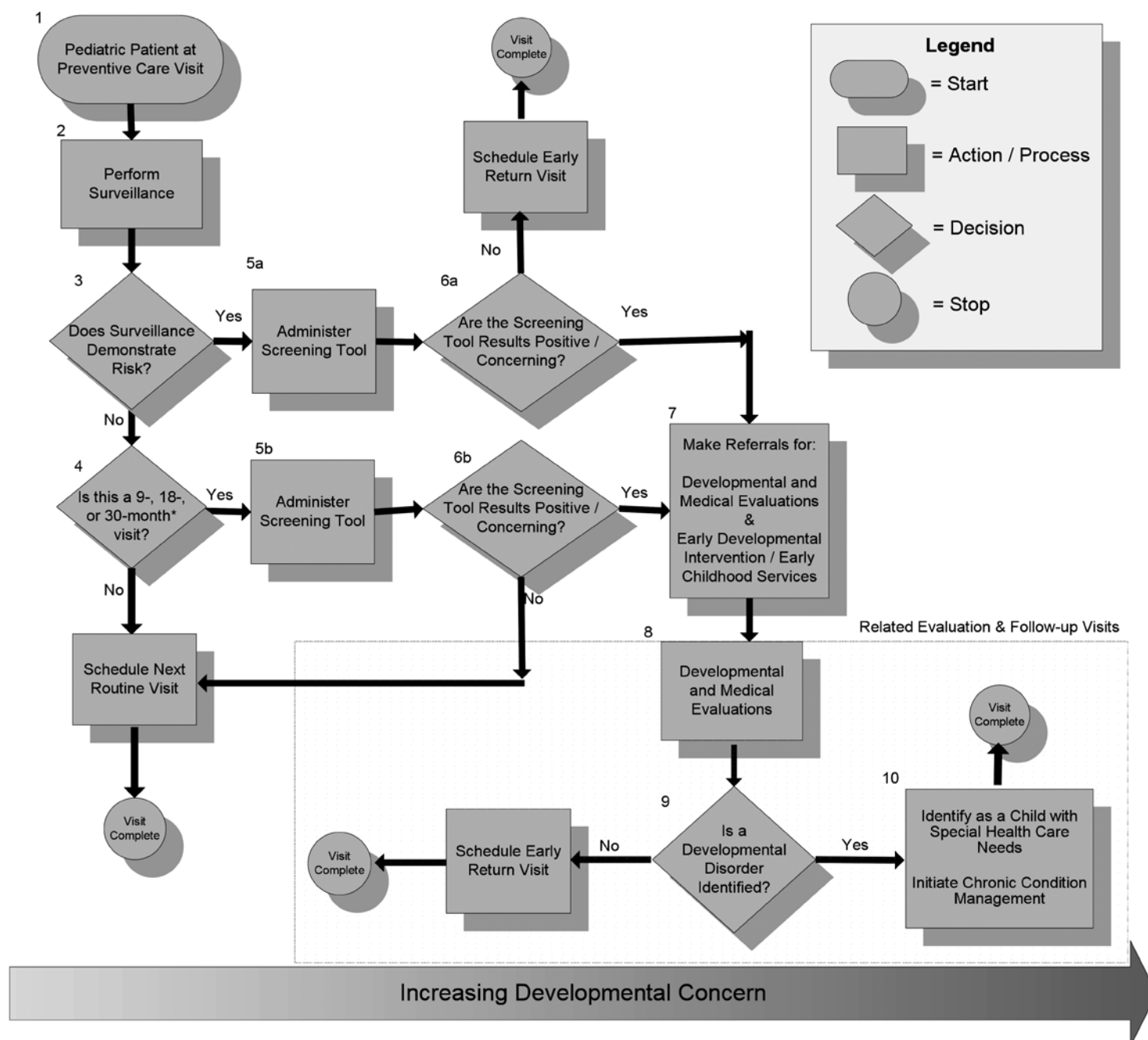




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# Developmental Surveillance and Screening Algorithm Within a Pediatric Preventive Care Visit



\*Because the 30-month visit is not yet a part of the preventive care system and is often not reimbursable by third-party payers at this time, developmental screening can be performed at 24 months of age.

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## Developmental and Screening Algorithm Within a Pediatric Preventive Care Visit

Pediatric Patient at Preventive Care Visit

1. Developmental concerns should be included as one of several health topics addressed at each pediatric preventive care visit throughout the first 5 years of life.<sup>5</sup>

2. **Developmental Surveillance** is a flexible, longitudinal, continuous, and cumulative process whereby knowledgeable health care professionals identify children who may have developmental problems. There are 5 components of development surveillance: eliciting and attending to the parents' concerns about their child's development, documenting and maintaining a developmental history, making accurate observations of the child, identifying the risk and protective factors, and maintaining an accurate record and documenting the process and findings.

Perform Surveillance

Does Surveillance Demonstrate Risk?

3. The concerns of both parents and child health professionals should be included in determining whether surveillance suggests the child may be at risk of developmental delay. If either parents or the child health professional express concern about the child's development, a developmental screening to address the concern specifically should be conducted.

4. All children should receive developmental screening using a standardized test. In the absence of established risk factors or parental or provider concerns, a general developmental screen is recommended at the 9-, 18-, and 30-month\* visits. Additionally, autism-specific screening is recommended for all children at the 18-month visit.

*\*Because the 30-month visit is not yet a part of the preventive care system and is often not reimbursable by third-party payers at this time, developmental screening can be performed at 24 months of age.*

Is this a 9-, 18-, or 30-month\* visit?

Administer Screening Tool

5a & 5b. **Developmental screening** is the administration of a brief standardized tool aiding the identification of children at risk of a developmental disorder. Developmental screening that targets the area of concern is indicated whenever a problem is identified during developmental surveillance.

6a & 6b. When the results of the periodic screening tool are normal, the child health professional can inform the parents and continue with other aspects of the preventive visit. When a screening tool is administered as a result of concerns about development, an early return visit to provide additional developmental surveillance should be scheduled, even if the screening tool results do not indicate a risk of delay.

Are the Screening Tool Results Positive / Concerning?

Make Referrals for:  
Developmental and Medical Evaluations & Early Developmental Intervention / Early Childhood Services

Developmental and Medical Evaluations

7-8. If screening results are concerning, the child should be scheduled for developmental and medical evaluations. **Developmental evaluation** is aimed at identifying the specific developmental disorder or disorders affecting the child. In addition to the developmental evaluation, a **medical diagnostic evaluation** to identify an underlying etiology should be undertaken. **Early Developmental Intervention/Early Childhood Services** can be particularly valuable when a child is first identified to be at high risk of delayed development, because these programs often provide evaluation services and can offer other services to the child and family even before an evaluation is complete.<sup>24</sup> Establishing an effective and efficient partnership with early childhood professionals is an important component of successful care coordination for children.<sup>39</sup>

9. If a developmental disorder is identified, the child should be identified as a child with special health care needs and chronic condition management should be initiated (see No. 10 below). If a developmental disorder is not identified through medical and developmental evaluation, the child should be scheduled for an early return visit for further surveillance. More frequent visits, with particular attention paid to areas of concern, will allow the child to be promptly referred for further evaluation if any further evidence of delayed development or a specific disorder emerges.

Is a Developmental Disorder Identified?

Identify as a Child with Special Health Care Needs  
Initiate Chronic Condition Management

10. When a child is discovered to have a significant developmental disorder, that child becomes a child with special health care needs, even if that child does not have a specific disease etiology identified. Such a child should be identified by the medical home for appropriate chronic condition management and regular monitoring and entered into the practice's children and youth with special health care needs registry.<sup>40</sup>

## Surveillance and Screening Algorithm: Autism Spectrum Disorders (ASDs)

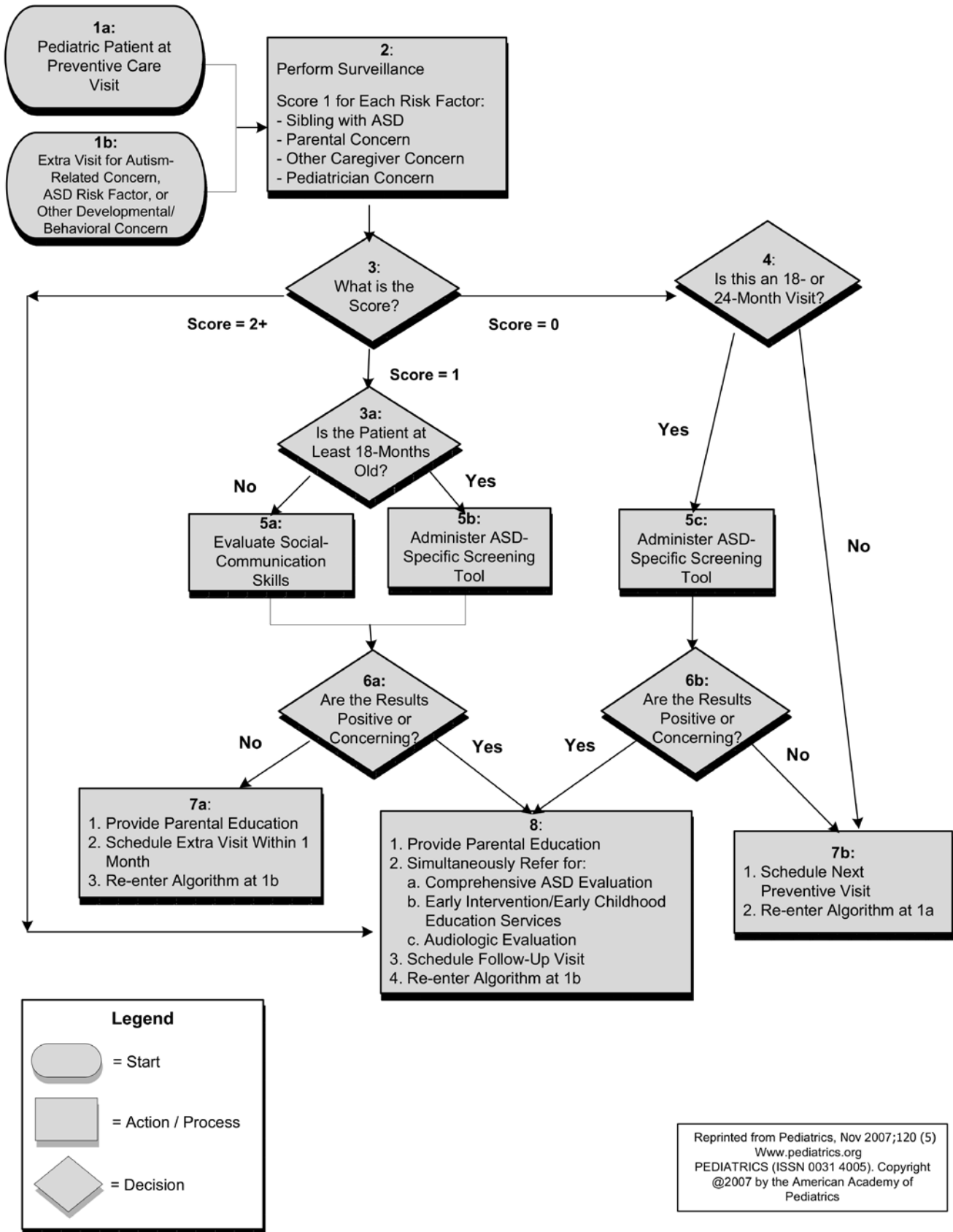
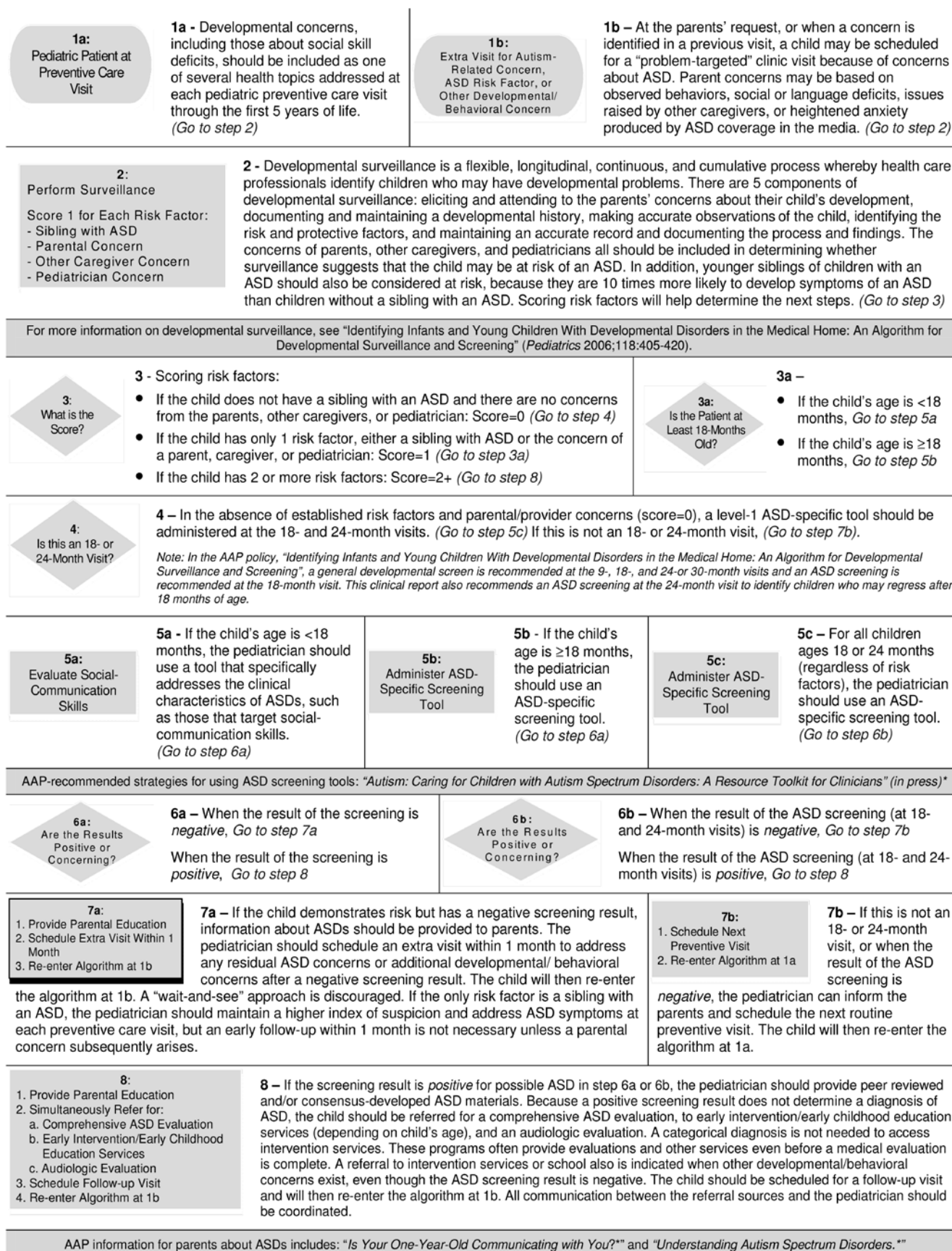


FIGURE 1  
 Surveillance and screening algorithm: ASDs. (Reproduced with permission from Pediatrics)



## Surveillance and Screening Algorithm: Autism Spectrum Disorders (ASDs)



\*Available at [www.aap.org](http://www.aap.org)



# Developmental Screening

## FACT SHEET

### What is child development?

A child's growth is more than just physical. Children grow, develop, and learn throughout their lives, starting at birth. A child's development can be followed by how they play, learn, speak, and behave.

### What is a developmental delay?

#### Will my child just grow out of it?

Skills such as taking a first step, smiling for the first time, and waving "bye bye" are called developmental milestones. Children reach milestones in playing, learning, speaking, behaving, and moving (crawling, walking, etc.). A developmental delay is when your child does not reach these milestones at the same time as other children the same age. If your child is not developing properly, there are things you can do that may help. Most of the time, a developmental problem is not something your child will "grow out of" on his or her own. But with help, your child could reach his or her full potential!

### What is developmental screening?

Doctors and nurses use developmental screening to tell if children are learning basic skills when they should, or if they might have problems. Your child's doctor may ask you questions or talk and play with your child during an exam to see how he or she learns, speaks, behaves, and moves. Since there is no lab or blood test to tell if your child may have a delay, the developmental screening will help tell if your child needs to see a specialist.

### Why is developmental screening important?

When a developmental delay is not recognized early, children must wait to get the help they need. This can make it hard for them to learn when they start school. In the United States, 17 percent of children have a developmental or behavioral disability such as autism, intellectual disability (also known as mental retardation), or Attention-Deficit/Hyperactivity Disorder (ADHD).

[www.cdc.gov/actearly](http://www.cdc.gov/actearly)

In addition, many children have delays in language or other areas. But, less than half of children with problems are identified before starting school. During this time, the child could have received help for these problems and may even have entered school more ready to learn.

### I have concerns that my child could have a developmental delay. Whom can I contact in my state to get a developmental assessment for my child?

Talk to your child's doctor or nurse if you have concerns about how your child is developing. If you or your doctor think there could be a problem, you can take your child to see a developmental pediatrician or other specialist, and you can contact your local early intervention agency (for children under 3) or public school (for children 3 and older) for help. To find out who to speak to in your area, you can contact the National Dissemination Center for Children with Disabilities by logging on to [www.nichcy.org/states.htm](http://www.nichcy.org/states.htm). In addition, the Centers for Disease Control and Prevention (CDC) has links to information for families at ([www.cdc.gov/actearly](http://www.cdc.gov/actearly)). If there is a problem, it is very important to get your child help as soon as possible.

### How can I help my child's development?

Proper nutrition, exercise, and rest are very important for children's health and development. Providing a safe and loving home and spending time with your child – playing, singing, reading, and even just talking – can also make a big difference in his or her development.

For other ideas of activities to do with your child, and for child safety information, go to [www.cdc.gov/ncbddd/child/](http://www.cdc.gov/ncbddd/child/) and look in the "developmental milestones" section.



**Learn the Signs. Act Early.**

# Hoja informativa sobre el análisis del desarrollo

## ¿Qué es el desarrollo infantil?

El crecimiento de un niño no es solo de tipo físico. Desde su nacimiento y durante toda su vida los niños crecen, se desarrollan y aprenden. El desarrollo de un niño se puede seguir por la manera en que juega, aprende, habla y se comporta.

## ¿Qué es un retraso en el desarrollo?

### ¿Podrá mi hijo superarlo por sí solo?

Ciertas destrezas como dar el primer paso, sonreír por primera vez y mover la mano para decir adiós se denominan indicadores importantes en el desarrollo. Cada niño alcanza estos indicadores importantes en áreas como el juego, el aprendizaje, el habla, la conducta y el movimiento (gatear, caminar etc.). Un retraso en el desarrollo ocurre cuando su hijo no alcanza estos indicadores importantes más o menos al mismo tiempo que otros niños de su misma edad. Si su hijo no se está desarrollando debidamente hay algunas cosas que puede hacer para ayudarlo. Generalmente los niños no superan los problemas de desarrollo por sí solos pero con su ayuda podrá alcanzar su máximo potencial.

## ¿Qué es el análisis del desarrollo?

Los doctores y enfermeras analizan el desarrollo para determinar si los niños están aprendiendo las destrezas básicas a su debido tiempo o si tienen problemas. Durante el examen, el doctor de su hijo o pediatra puede hacerle preguntas a usted o conversar y jugar con su hijo para observar su forma de aprender, de hablar, de comportarse y de moverse. Como no existe un análisis de sangre o de laboratorio que indique si su hijo tiene un retraso, el análisis del desarrollo determinará si su hijo necesita ver a un especialista.

## ¿Por qué es importante el análisis del desarrollo?

Cuando no se identifica en un comienzo el retraso en el desarrollo, los niños deben esperar más tiempo para recibir ayuda, lo cual puede dificultar su aprendizaje al ingresar a la escuela. En los Estados Unidos, el 17% de los niños presenta discapacidades en el desarrollo o la conducta tales como: autismo, discapacidad intelectual (también conocido como retraso mental) o trastorno de déficit de atención con hiperactividad (ADHD por sus siglas en inglés). Adicionalmente, muchos niños presentan retraso en el lenguaje y otras áreas.

Sin embargo, menos de la mitad de los niños con problemas son identificados antes de entrar a la escuela y por consiguiente no reciben la ayuda necesaria que les podría preparar mejor para el ingreso a la escuela.

## Me preocupa que mi hijo pueda tener un retraso en el desarrollo. ¿Con quién hablo en el estado en que vivo para que le hagan a mi hijo un análisis del desarrollo?

Hable con su doctor o enfermera si está preocupado por el desarrollo de su hijo. Si usted o su doctor piensan que existe algún problema, puede llevar a su hijo a un pediatra especializado en el desarrollo u otro especialista entrenado en este campo y puede llamar a su agencia local de intervención temprana (para niños menores de 3 años) o su escuela pública (para niños de 3 años o más) para que le presten ayuda. Para averiguar con quién puede hablar en su área puede comunicarse con el Centro Nacional de Información sobre Niños y Jóvenes con Discapacidades (NICHY por sus siglas en inglés) ya sea en la página de Internet [www.nichcy.org/states.htm](http://www.nichcy.org/states.htm). Los Centros para el Control y la Prevención de Enfermedades (CDC por sus siglas en inglés) también tienen enlaces con información para las familias en el sitio electrónico [www.cdc.gov/pronto](http://www.cdc.gov/pronto). Si existe algún problema es de suma importancia buscar ayuda para su hijo lo más pronto posible.

## ¿Cómo puedo ayudar al desarrollo de mi hijo?

La nutrición, el ejercicio y el descanso apropiados son partes muy importantes en la salud y el desarrollo de los niños. Usted también puede tener una gran influencia en el desarrollo de su hijo si le brinda cariño, un hogar seguro y le dedica tiempo ya sea jugando, cantando, leyendo o simplemente platicando.

Para obtener otras ideas sobre actividades que puede realizar con su hijo, así como información sobre la seguridad infantil, vaya a [www.cdc.gov/ncbddd/child/](http://www.cdc.gov/ncbddd/child/) y busque la sección de indicadores importantes ("developmental milestones").

[www.cdc.gov/pronto](http://www.cdc.gov/pronto)



Aprenda los signos. Reaccione pronto.

## *DSM-5 Definition of Autism Spectrum Disorder*

### **299.00 Autism Spectrum Disorder**

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history (examples are illustrative, not exhaustive; see text):

1. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back-and-forth conversation; to reduced sharing of interest, emotions, or affect; to failure to initiate or respond to social interactions.
2. Deficits in nonverbal communicative behaviors used to social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
3. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts, to difficulties in sharing imaginative play or in making friends; to absence of interest in peers.

*Specify* current severity:

**Severity is based on social communication impairments and restricted, repetitive patterns of behavior** (see Table A1)

B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history (examples are illustrative, not exhaustive; see text):

1. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases)
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food every day).
3. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g., strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).
4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment (e.g., apparent indifference to pain/temperature, adverse response

to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement).

*Specify* current severity:

**Severity is based on social communication impairments and restricted, repetitive patterns of behavior** (see Table A1)

- C. Symptoms must be present in the early developmental period (but may not become fully manifested until social demands exceed limited capacities, or may be masked by learned strategies in later life).
- D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.
- E. These disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay. Intellectual disability and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected of general developmental level.

**Note:** Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger's disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder. Individuals who have marked deficits in social communication, but whose symptoms do not otherwise need criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.

*Specify* if:

**With or without accompanying intellectual impairment**

**With or without accompanying language impairment**

**Associated with a known medical or genetic condition or environmental factor** (**Coding note:** Use additional code to identify the associated medical or genetic condition.)

**Associated with another neurological, mental, or behavioral disorder** (**Coding note:** Use additional code[s] to identify the associated neurodevelopmental, mental, or behavioral disorder[s].)

**With catatonia** (refer to the criteria for catatonia associated with another mental disorder, pp. 119-120, for definition) (**Coding note:** Use additional code 293.89 [F06.1] catatonia associated with autism spectrum disorder to indicate the presence of the comorbid catatonia).

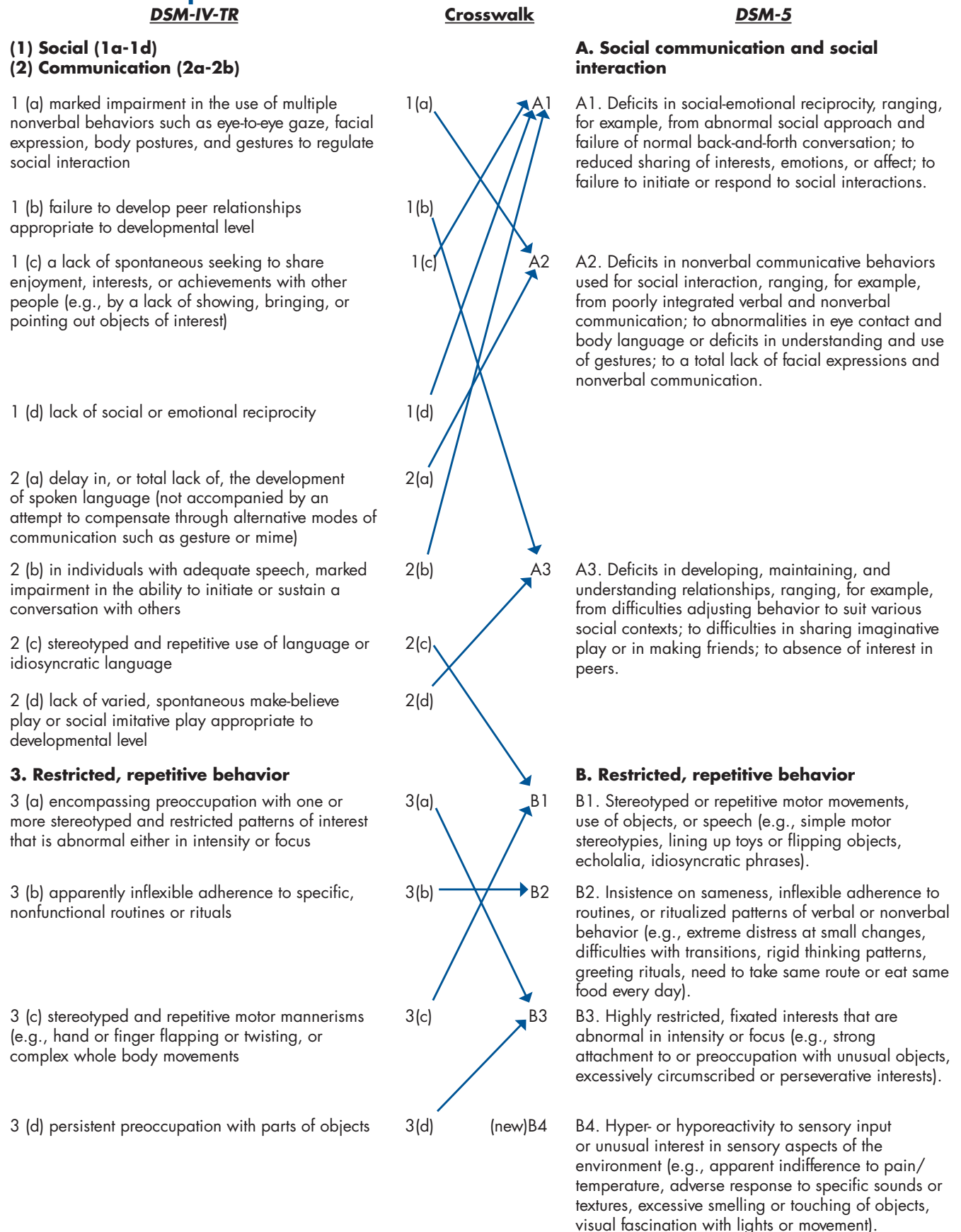


Table B1. *Severity levels for DSM-5 definition of autism spectrum disorder*

<b>Severity Level</b>	<b>Social communication</b>	<b>Restricted, repetitive behaviors</b>
<p>Level 3</p> <p>“Requiring very substantial support”</p>	<p>Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning, very limited initiation of social interactions, and minimal response to social overtures from others. For example, a person with few words of intelligible speech who rarely initiates interaction and, when he or she does, makes unusual approaches to meet needs only and responds to only very direct social approaches.</p>	<p>Inflexibility of behavior, extreme difficulty coping with change, or other restricted/</p> <p>repetitive behaviors markedly interfere with functioning in all spheres. Great distress/</p> <p>difficulty changing focus or action.</p>
<p>Level 2</p> <p>“Requiring substantial support”</p>	<p>Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place; limited initiation of social interactions; and reduced or abnormal responses to social overtures from others. For example, a person who speaks simple sentences, whose interaction is limited to narrow special interests, and who has markedly odd nonverbal communication.</p>	<p>Inflexibility of behavior, difficulty coping with change, or other restricted/ repetitive behaviors appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts. Distress and/or difficulty changing focus or action.</p>
<p>Level 1</p> <p>“Requiring support”</p>	<p>Without supports in place, deficits in social communication cause noticeable impairments. Difficulty initiating social interactions, and clear examples of atypical or unsuccessful responses to social overtures of others. May appear to have decreased interest in social interactions. For example, a person who is able to speak in full sentences and engages in communication but whose to-and-fro conversation with others fails, and whose attempts to make friends are odd and typically unsuccessful.</p>	<p>Inflexibility of behavior causes significant interference with functioning in one or more contexts. Difficulty switching between activities. Problems of organization and planning hamper independence.</p>

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## Figure B1. Crosswalk of diagnostic criteria for *DSM-IV-TR* Autistic Disorder and *DSM-5* Autism Spectrum Disorder





***DSM-IV-TR* Definition of Autistic Disorder, Asperger syndrome and Pervasive  
Developmental Delay-Not Otherwise Specified**

**299.00 Autistic Disorder**

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):

- (1) qualitative impairment in social interaction, as manifested by at least two of the following:
  - (a) marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
  - (b) failure to develop peer relationships appropriate to developmental level
  - (c) a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
  - (d) lack of social or emotional reciprocity
- (2) qualitative impairments in communication as manifested by at least one of the following:
  - (a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
  - (b) in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
  - (c) stereotyped and repetitive use of language or idiosyncratic language
  - (d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
- (3) restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
  - (a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
  - (b) apparently inflexible adherence to specific, nonfunctional routines or rituals
  - (c) stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
  - (d) persistent preoccupation with parts of objects.

- B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.
- C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

### **299.80 Asperger's Disorder**

- A. Qualitative impairment in social interaction, as manifested by at least two of the following:
  - (1) marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
  - (2) failure to develop peer relationships appropriate to developmental level
  - (3) a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest to other people)
  - (4) lack of social or emotional reciprocity.
- B. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
  - (1) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
  - (2) apparently inflexible adherence to specific, nonfunctional routines or rituals
  - (3) stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
  - (4) persistent preoccupation with parts of objects.
- C. The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.
- D. There is no clinically significant general delay in language (e.g., single words used by age 2 years, communicative phrases used by age 3 years).
- E. There is no clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood.
- F. Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia.

### **299.80 Pervasive Developmental Disorder Not Otherwise Specified (Including Atypical Autism)**

This category should be used when there is a severe and pervasive impairment in the development of reciprocal social interaction associated with impairment in either verbal or nonverbal communication skills or with the presence of stereotyped behavior, interests, and activities, but the criteria are not met for a specific Pervasive Developmental Disorder, Schizophrenia, Schizotypal Personality Disorder, or Avoidant Personality Disorder. For example, this category includes “atypical autism”—presentations that do not meet the criteria for Autistic Disorder because of late age at onset, atypical symptomatology, or subthreshold symptomatology, or all of these.

Reprinted from: American Psychiatric Association, *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision (DMS-IV-TR)*. Washington, DC: American Psychiatric Publishing, 2000.

## ***ICD-10 Criteria for Childhood Autism (F84.0)***

A. Abnormal or impaired development is evident before the age of 3 years in at least one out of the following areas:

- (1) receptive or expressive language as used in social communication;
- (2) the development of selective social attachments or of reciprocal social interaction;
- (3) functional or symbolic play.

B. A total of at least six symptoms from (1), (2), and (3) must be present, with at least two from (1) and at least one from each of (2) and (3):

(1) Qualitative abnormalities in reciprocal social interaction are manifest in at least two of the following areas:

- (a) failure adequately to use eye-to-eye gaze, facial expression, body posture and gesture to regulate social interaction;
- (b) failure to develop (in a manner appropriate to mental age, and despite ample opportunities) peer relationships that involve a mutual sharing of interests, activities and emotions;
- (c) A lack of socio-emotional reciprocity as shown by an impaired or deviant response to other people's emotions; or lack of modulation of behaviour according to social context, or a weak integration of social, emotional and communicative behaviours;
- (d) lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., a lack of showing, bringing, or pointing out to other people objects of interest to the individual).

(2) Qualitative abnormalities in communication are manifest in at least one of the following areas:

- (a) a delay in, or total lack of, development of spoken language that is *not* accompanied by an attempt to compensate through the use of gesture or mime as alternative modes of communication (often preceded by a lack of communicative babbling);
- (b) relative failure to initiate or sustain conversational interchange (at whatever level of language skills are present) in which there is reciprocal to and from responsiveness to the communications of the other person;
- (c) stereotyped and repetitive use of language or idiosyncratic use of words or phrases;
- (d) lack of varied spontaneous make-believe or (when young) social imitative play.

(3) Restricted, repetitive, and stereotyped patterns of behaviour, interests and activities are manifest in at least one of the following areas:

- (a) an encompassing preoccupation with one or more stereotyped and restricted patterns of interest that are abnormal in content or focus; or one or more interests that are abnormal in their intensity and circumscribed nature although not abnormal in their content or focus;
- (b) apparently compulsive adherence to specific, non-functional, routines or rituals;
- (c) stereotyped and repetitive motor mannerisms that involve either hand or finger flapping or twisting, or complex whole body movements;
- (d) preoccupations with part-objects or non-functional elements of play materials (such as their odour, the feel of their surface, or the noise or vibration that they generate);

C. The clinical picture is not attributable to the other varieties of pervasive developmental disorder; specific developmental disorder of receptive language (F82.0) with secondary socio-emotional problems; reactive attachment disorder (F94.1) or disinhibited attachment disorder (F94.2); mental retardation (F70- F72) with some associated emotional or behavioural disorder; schizophrenia (F20.-) of unusually early onset; and Rett's syndrome (F82.4).

Source: *International Classification of Diseases: Diagnostic Criteria for Research*, tenth edition, by the World Health Organization, 1992, Geneva Switzerland: Author. Reprinted with permission



Table D1. *Additional standardized measures*

Instrument (Reference)	Description	Method of Administration	Age Range	Administered by	Administration Time
<b>Developmental assessments</b>					
Battelle De- velopmental Inventory, 2 <sup>nd</sup> Edition  (Newborg, 2004)	Measures person- al-social, adap- tive, motor, com- munication, and cognitive ability	Formal child interaction	Birth to 7 years 11 months	Teachers, in- cluding infant, preschool, primary, and special educa- tion teachers	Assessment: 60-90 minutes Screening test: 10-30 minutes
Bayley Scales of Infant and Toddler Devel- opment–III  (Bayley, 2006)	Measures develop- ment across five scales: cognitive, motor, language, social-emotional, adaptive behav- ior.	Formal child interaction and parent report via question- naire	One to 42 months	Highly trained person. Trained staff without gradu- ate or profes- sional training in assessment may adminis- ter and score the assessment under supervi- sion.	30 to 90 minutes
Brigance Inventory of Early Devel- opment–II  (Brigance, 2004)	Measures physi- cal and language development, aca- demic/cognitive functioning, daily living skills, fine and gross motor skills, receptive and expressive communication, social and emo- tional skills and adaptive behav- ior.	Child interaction/ observation and parent interview	Birth through seven years	Teacher, school psy- chologist or developmental expert, or other early education professional.	20 to 55 minutes

<b>Instrument (Reference)</b>	<b>Description</b>	<b>Method of Administration</b>	<b>Age Range</b>	<b>Administered by</b>	<b>Administration Time</b>
Leiter International Performance Scale, Revised  (Roid & Miller, 1998)	Measures intelligence among children who are non-verbal, non English speaking, cognitively impaired, or hearing or speech impaired. Measures include reasoning, visualization, memory, and attention.	Individually administered game-like tasks	Ages two to 21 years	Professional who has received supervised training and practice. It should be interpreted by someone with graduate training in psychological assessment.	40 to 90 minutes
Mullen Scales of Early Learning  (Mullen, 1995)	Measures motor and cognitive abilities across five scales: gross motor, visual reception, fine motor, expressive language, and receptive language.	Formal child interaction	Birth to 68 months	A professional with training or practical experience in the clinical assessment of infants and young children.	15 to 60 minutes, depending on the age of the child.
Stanford-Binet Intelligence Scales, 5 <sup>th</sup> edition  (Roid, 2003)	Measures four areas of cognitive ability: verbal reasoning; quantitative reasoning; abstract/visual reasoning; and short-term memory.	Formal child interaction	2 to 23 years	The test is administered by trained diagnosticians	30 to 90 minutes
Wechsler Preschool and Primary Scales of Intelligence, III  (Wechsler, 2002)	Measures full scale IQ, verbal IQ, performance IQ, and processing speed, with an optional general language composite.	Formal child interaction	Two subsets: ages 2.5 years -4 years and ages 4 to 7 years	Trained professional	Takes 30 to 90 minutes to complete, depending on the age of the child

Instrument (Reference)	Description	Method of Administration	Age Range	Administered by	Administration Time
<b>Adaptive functioning</b>					
Adaptive Behavior Assessment System-Second Edition  (Harrison & Oakland, 2003)	Measures adaptive behavior skills in three broad domains: conceptual, social and practical.	Behavior rating format completed by parents, teachers, or other caregivers	Birth to adult	Psychologists, social workers, neurologists and others	Takes 15 to 20 minutes to administer
Scales of Independent Behavior-Revised  (Bruininks, Woodcock, Weatherman, & Hill, 1996)	Measures 14 areas of adaptive behavior and 8 areas of problem behavior. Four domains include: motor skills, social interaction and communication skills, personal living skills, and community living skills.	Paper and pencil reported by caregiver/parent	Birth to adult	Trained person	Depending on which scale is used 15-20 or 45-60 minutes
Vineland Adaptive Behavior Scales, Second Edition  (Sparrow, Cicchetti, & Balla, 2005)	Measures personal and social skills for individuals. Includes measures across the following domains: communication, daily living skills, socialization, motor skills, and maladaptive behavior index (optional). Versions available to be completed by parents or teachers.	Paper and pencil reported by caregiver/parent	Birth through 21 years	Interviewers should have graduate-level education in psychology or social work as well as in individual assessment and test interpretation	20 to 60 minutes

<b>Instrument (Reference)</b>	<b>Description</b>	<b>Method of Administration</b>	<b>Age Range</b>	<b>Administered by</b>	<b>Administration Time</b>
Clinical Evaluation of Language Fundamentals – Preschool, Second Edition  (Wiig, Secord, & Semel, 2004)	Measures expressive and receptive language, language content and structure.	Formal child interaction and observation	Three to six years	Trained person	30 to 60 minutes
Communication and Symbolic Behavior Scales Developmental Profile  (Wetherby, & Prizant, 2002)	A screening and evaluation tool that helps determine the communicative competence (use of eye gaze, gestures, sounds, words, understanding, and play).	Paper and pencil parent form and behavioral observation	Functional communication age 6 to 24 months (chronological age 6 months to 6 years)	Certified SLP, early interventionist, psychologist, pediatrician, or other professional trained to assess developmentally young children	5 to 10 minutes
Comprehensive Assessment of Spoken Language (CASL)  (Carrow-Woolfolk, 1999)	Measures oral skills and the literal, figurative, and social aspects of language. Consists of 15 individually administered tests with multiple choice responses.	Test books are self-standing with images shown to the child to respond to	Ages 3 to 21 years	A professional in psychology, education, occupational therapy, SLP, or social work	30-35 minutes
MacArthur-Bates Communicative Development Inventories, Third Edition  (Fenson, Marchman, Thal, Dale, Reznick, & Bates, 2007)	Measures a child's understanding of words, gestures, sentences.	Parent report forms	Eight to 37 months	Clerical program staff	20 to 40 minutes

<b>Instrument (Reference)</b>	<b>Description</b>	<b>Method of Administration</b>	<b>Age Range</b>	<b>Administered by</b>	<b>Administration Time</b>
New Reynell Developmental Language Scales (RDLS), 4 <sup>th</sup> Edition (Edwards, Letts, & Sinka, 2011)	Measures a child's understanding of selected vocabulary items and grammatical features a child's production of the same features of language.	Picture books and stimulus materials for child to respond to	3 years to 7 years 6 months	Therapists, clinicians, educators	Varies based on the individual child
Peabody Picture Vocabulary Test-Fourth Edition (Dunn & Dunn, 2007)	Assessment of one word receptive vocabulary for children.	Paper and pencil	30 months to adult	Trained person with a bachelor's degree with coursework in measurement	10 to 15 minutes
Preschool Language Scale, Fifth Edition (Zimmerman, Steiner, & Pond, 2011)	Measures total language, auditory comprehension and expressive communication for children.	Child interaction and parent/caregiver questionnaire	Birth through 7.11 years	Consultant or expert with clinical training  Paraprofessional staff with training	45 to 60 minutes
Test of Language Competence (TLC) (Wiig & Secord, 1989)	Measures the ability to perceive, interpret, and respond to the contextual and situational demands of conversation as well as semantic and syntactic abilities.	Paper and pencil	Level 1 ages 5-9  Level 2 ages 10-18	A trained professional	Takes less than 60 minutes to administer
Test of Pragmatic Language, Second Edition (TOPL-2) (Phelps-Terasaki & Phelps-Gunn, 2007)	Measures the effectiveness and appropriateness of seven core subcomponents: physical context, audience, topic, purpose, visual-gestural cues, abstractions; and pragmatic evaluation.	Formal child interaction	Six to 18 year olds	Speech-language pathologists	45-60 minutes



<b>Instrument (Reference)</b>	<b>Description</b>	<b>Method of Administration</b>	<b>Age Range</b>	<b>Administered by</b>	<b>Administration Time</b>
Test of Problem Solving (TOPS) (Bowers, Huis- ingh & LoGu- idice, 2005)	Measures language- based thinking abili- ties and strategies using logic experi- ence.	The Picture Stimuli Book presents situa- tions in full-color photographs. The student refers to these photo- graphs when answering the examiner's questions	Available in two ver- sions:  -TOPS 2: Adoles- cent ages 12-17  -TOPS 3: Elemen- tary ages 6-12	Trained profes- sional familiar with language disorders (e.g., speech- language pathologist, psychologists)	35-40 minutes
<b>Social interaction</b>					
Social Skills improvement System (SSiS) (Greshman & Elliott, 2008)	Assesses social skills (e.g., commu- nication, engage- ment), behaviors (e.g., bullying, autism spectrum), and academic competence (e.g., math, reading). Re- places the Social Skills Rating Scales (Gresham & Elliott, 1990).	Paper and pencil rating	3 to 18 years	Teacher, parent and student forms	10 to 25 minutes
<b>Behavior assessment</b>					
Aberrant Be- havior Check- list, Commu- nity Version (Aman & Singh, 1986)	A behavior rating scale that includes 58 items across 5 domains of behavior: irrita- bility/agitation/ crying; lethargy/ social withdrawal; stereotypic behav- ior; hyperactivity/ noncompliance; and inappropriate speech.	Paper and pencil rating	6 to 51+ years	Parent, teacher, caretaker	5 minutes

<b>Instrument (Reference)</b>	<b>Description</b>	<b>Method of Administration</b>	<b>Age Range</b>	<b>Administered by</b>	<b>Administration Time</b>
Behavior Assessment System for Children, Second Edition (BASC-2)  (Reynolds & Kamphaus, 2004)	Measures anxiety, aggression, attention, atypical behaviors, social skills, and adaptive behaviors.	Paper and pencil rating forms completed and observation form	2 to 21 years and 11 months	A clinician administers the Student Observation System; teacher and parent administer other modules	10 to 20 minutes
Child Behavior Checklist  CBCL for preschoolers: (Achenbach & Rescorla, 2000)  CBCL for school aged children: (Achenbach, 2001)	The preschool version includes 99 items concerning behavioral, emotional, and social problems. The school aged version includes 113 behavioral problems the parent rates in addition to several questions about the child's social and academic development.	Paper and pencil or on line report	Preschool version:  18 months to 5 years  School aged version: 6 to 18 year olds	Parent	20 minutes

### **Signs and Symptoms of Possible Autism in Secondary School Children (older than 11 years or equivalent “mental” age)**

#### **Social interaction and reciprocal communication behaviours**

##### *Spoken language*

- Spoken language may be unusual in several ways:
  - o very limited use
  - o monotonous tone
  - o repetitive speech, frequent use of stereotyped (learnt) phrases, content dominated by excessive information on topics of own interest
  - o talking “at” others rather than sharing a two-way conversation
  - o responses to others can seem rude or inappropriate

##### *Interacting with others*

- Reduced or absent awareness of personal space, or unusually intolerant of people entering their personal space
- Long-standing difficulties in reciprocal social communication and interaction: few close friends or reciprocal relationships
- Reduced or absent understanding of friendship; often an unsuccessful desire to have friends (although may find it easier with adults or younger children)
- Social isolation and apparent preference for aloneness
- Reduced or absent greeting and farewell behaviors
- Lack of awareness and understanding of socially expected behavior
- Problems losing at games, turn-taking and understanding „changing the rules“
- May appear unaware or uninterested in what other young people his or her age are interested in
- Unable to adapt style of communication to social situations, for example may be overly formal or inappropriately familiar
- Subtle difficulties in understanding other’s intentions; may take things literally and misunderstand sarcasm or metaphor
- Makes comments without awareness of social niceties or hierarchies

- Unusually negative response to the requests of others (demand avoidant behavior)

*Eye contact, pointing and other gestures*

- Poorly integrated gestures, facial expressions, body orientation, eye contact (looking at people's eyes when speaking) assuming adequate vision, and spoken language used in social communication

*Ideas and imagination*

- History of a lack of flexible social imaginative play and creativity, although scenes seen on visual media (for example, television) may be re-enacted

**Unusual or restricted interests and/or rigid and repetitive behaviors**

- Repetitive “stereotypical” movements such as hand flapping, body rocking while standing, spinning, finger flicking
- Preference for highly specific interests or hobbies
- A strong adherence to rules or fairness that leads to argument
- Highly repetitive behaviors or rituals that negatively affect the young person's daily activities
- Excessive emotional distress at what seems trivial to others, for example change in routine
- Dislike of change, which often leads to anxiety or other forms of distress including aggression
- Over or under reaction to sensory stimuli, for example textures, sounds, smells
- Excessive reaction to taste, smell, texture or appearance of food and/or extreme food fads

**Other factors that may support a concern about autism**

- Unusual profile of skills and deficits (for example, social or motor coordination skills poorly developed, while particular areas of knowledge, reading or vocabulary skills are advanced for chronological or mental age)
- Social and emotional development more immature than other areas of development, excessive trusting (naivety), lack of common sense, less independent than peers

Reproduced from: NICE. *Autism: Recognition, referral and diagnosis of children and young people on the autism spectrum*. NICE Clinical Guideline 128, September 2011, with the permission of the Royal College of the Obstetricians and Gynaecologists on behalf of the National Collaborating Centre for Women's and Children's Health.



# Connecticut Medical Home Initiative (CMHI) for Children & Youth with Special Health Care Needs

## Who is eligible?

Children & youth age 0 to 21 who have, or are at increased risk for, a chronic physical, developmental, behavioral or emotional condition and who also require health and related services of a type or amount beyond that required by children generally.

## Services available?

All families of eligible children and youth with special health care needs (CYSHCN), regardless of income, will receive a respectful working partnership with you and your child’s medical home; care coordination services and family support referrals.

Uninsured or underinsured families, who fall within income guidelines, can also benefit from payment for limited services (i.e. durable medical equipment, prescriptions, and special nutritional formulas).

**Contact the Connecticut Medical Home Initiative at FAVOR, Inc. at 1-855-436-6544 (toll free).**

SOUTHWEST	SOUTH CENTRAL	EASTERN	NORTH CENTRAL	NORTHWEST
Stamford Hospital Stamford	Family-centered Services of CT, Inc. New Haven	United Community and Family Services, Inc. Norwich	Connecticut Children’s Medical Center Hartford	St. Mary’s Hospital Waterbury
1-866-239-3907 (toll free)	1-877-624-2601 (toll free)	1-866-923-8237 (toll free)	1-877-835-5768 (toll free)	1-866-517-4388 (toll free)

## United Way of Connecticut’s Child Development Infoline

The central access point for Connecticut’s Medical Home Initiative for CYSHCN.  
Provides information about medical, educational and recreational resources  
1-800-505-7000

## Connecticut Family Support Network

Contact for family support, information and advocacy at 1-877- FSN-2DAY



# CMHI Regional Town Listings

SOUTHWEST REGION	SOUTH CENTRAL REGION	EASTERN REGION	NORTH CENTRAL REGION	NORTHWEST REGION
<b>Stamford Health Systems</b> <b>Stamford</b> <b>Toll Free 866-239-3907</b>	<b>Family-centered Services of CT</b> <b>New Haven</b> <b>Toll Free 877-624-2601</b>	<b>United Community and Family Services</b> <b>Norwich</b> <b>Toll Free 866-923-8237</b>	<b>Connecticut Children's Medical Center</b> <b>Hartford</b> <b>Toll Free 877-835-5768</b>	<b>St. Mary's Hospital</b> <b>Waterbury</b> <b>Toll Free 866-517-4388</b>
BRIDGEPORT DARIEN EASTON FAIRFIELD GREENWICH MONROE NEW CANAAN NORWALK STAMFORD STRATFORD TRUMBULL WESTON WESTPORT WILTON	ANSONIA BETHANY BRANFORD CHESTER CLINTON CROMWELL DEEP RIVER DERBY DURHAM EAST HADDAM EAST HAMPTON EAST HAVEN ESSEX GUILFORD HADDAM HAMDEN KILLINGWORTH LYME MADISON MERIDEN MIDDLEFIELD MIDDLETOWN MILFORD NEW HAVEN NORTH BRANFORD NORTH HAVEN OLD LYME OLD SAYBROOK ORANGE PORTLAND SEYMOUR SHELTON WALLINGFORD WEST HAVEN WESTBROOK WOODBIDGE	ASHFORD BOZRAH BROOKLYN CANTERBURY CHAPLIN COLCHESTER COLUMBIA COVENTRY DANIELSON EAST LYME EASTFORD FRANKLIN GRISWOLD GROTON HAMPTON KILLINGLY LEBANON LEDYARD LISBON MANSFIELD MONTVILLE MOOSUP NEW LONDON NIANTIC NORTH STONINGTON NORWICH PLAINFIELD POMFRET PRESTON PUTNAM SALEM SCOTLAND SPRAGUE STERLING STONINGTON THOMPSON UNCASVILLE UNION VOLUNTOWN WATERFORD WILLINGTON WILLIMANTIC WINDHAM WOODSTOCK	ANDOVER AVON BERLIN BLOOMFIELD BOLTON BRISTOL BURLINGTON CANTON EAST GRANBY EAST HARTFORD EAST WINDSOR ELLINGTON ENFIELD FARMINGTON GEORGETOWN GLASTONBURY GRANBY HARTFORD HEBRON MANCHESTER MARLBOROUGH NEW BRITAIN NEWINGTON PLAINVILLE PLYMOUTH ROCKY HILL SIMSBURY SOMERS SOUTH WINDSOR SOUTHINGTON STAFFORD SUFFIELD TOLLAND VERNON WEST HARTFORD WETHERSFIELD WINDSOR WINDSOR LOCKS	BARKHAMSTED BEACON FALLS BETHEL BETHLEHEM BRIDGEWATER BROOKFIELD CANAAN CHESHIRE COLEBROOK CORNWALL DANBURY GOSHEN HARTLAND HARWINTON KENT LITCHFIELD MIDDLEBURY MORRIS NAUGATUCK NEW FAIRFIELD NEW HARTFORD NEW MILFORD NEWTOWN NORFOLK NORTH CANAAN OXFORD PROSPECT REDDING RIDGEFIELD ROXBURY SALISBURY SHARON SHERMAN SOUTHBURY THOMASTON TORRINGTON WARREN WASHINGTON WATERBURY WATERTOWN WINCHESTER WOLCOTT WOODBURY

# Connecticut's Child Development Infoline

The Gateway to Help and Referrals for  
Parents ● Providers ● Pediatric Professionals

**1-800-505-7000**

## Connecticut Birth to Three System

*Birth to 36 months of age*

*For children birth-36 months  
of age with developmental  
delays or disabilities.*

- Free Developmental Evaluation
- Service Coordination
- Individualized Family Service Plan (IFSP)
- Services from Early Childhood Therapists and Teachers as identified in the IFSP
- Focus on assisting families through natural routines and activities

## Help Me Grow

*Birth through Age 8*

*For children birth through age 8  
considered 'at-risk' for  
developmental or behavioral  
problems*

- Connects families to community based resources
- Provides Ages and Stages child monitoring program
- Trains child health providers in developmental screening
- Facilitates regional community networking

## Early Childhood Special Education

*Ages 3 through 5*

*For children ages 3 through 5  
who are found eligible for  
special education services*

- Evaluation
- Services to eligible children:
  - o Individualized Education Program (IEP)
  - o Special education and related services

## Children and Youth with Special Health Care Needs

*Birth to Age 21*

*For children and youth birth to  
age 21 with chronic physical,  
developmental, behavioral, or  
emotional conditions who require  
more health and related services  
than other children the same age.*

- Service Needs Assessment
- Care Coordination
- Benefits Coordination
- Family/Caregiver Support
- Respite Planning
- Links to medical home initiative
- Referrals to community based resources
- Transition Planning

## Participating Agencies

Children's Trust Fund ■ Department of Developmental Services ■ Department of Public Health ■ Department of Education ■ United Way of Connecticut



July 2008

## NOTES

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