Asperger syndrome (a specific autism spectrum disorder): Clinical features and diagnosis in children and adolescents

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INTRODUCTION — The American Psychiatric Association recognized Asperger disorder (also called Asperger syndrome) as a specific entity by publishing diagnostic criteria in the Diagnostic and Statistical Manual of Mental Disorders-IV (DSM-IV) in 1994 [1]. A modest literature on Asperger disorder has emerged since then, though there are no specific clinical features that establish Asperger disorder as distinct from the autism spectrum as a whole [2-4] and its status as a separate entity is uncertain. In the DSM-5, published in 2013, Asperger disorder is encompassed within the diagnosis of autism spectrum disorder [5]. In contrast, Asperger syndrome remains a distinct entity in the International Classification of Diseases, 10th Revision (ICD-10) [6].

This topic review summarizes the modest literature that describes Asperger syndrome as a separate entity. The terms “Asperger syndrome” and “autism spectrum disorder” (ASD) will be used interchangeably to acknowledge the two classification systems. Most of the information in this topic review will apply to children with ASD of Level 1 severity who do not have intellectual impairment, but may have language impairment. This approach is consistent with research that shows that subtypes of autism differ primarily on the basis of IQ [7,8]. (See 'Terminology' below and "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'.)

The clinical features and diagnosis of Asperger syndrome will be reviewed here. The management and prognosis are discussed separately. (See "Asperger syndrome (a specific autism spectrum disorder): Management and prognosis in children and adolescents".)

Overviews of the clinical features and diagnosis of ASD also are provided separately. (See "Clinical features of autism spectrum disorder" and "Diagnosis of autism spectrum disorder".)

TERMINOLOGY — Asperger syndrome is considered to be an "autism spectrum disorder" (ASD). ASD is a neurodevelopmental disorder that has diverse etiologies. It is characterized by a constellation of symptoms that includes deficits in reciprocal social interaction, social communication, and restricted and repetitive behavior (including stereotyped interests and activities) [5,6,9]. (See "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'.)

In the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5), a single diagnosis, “autism spectrum disorder” has replaced previous subtypes (eg, Asperger syndrome, “pervasive developmental disorder-not otherwise specified”) [5]. The clinical heterogeneity of
autism is indicated with specifiers for severity level and associated conditions (eg, intellectual impairment, language impairment). (See "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'.)

The DSM-5 indicates that individuals with a well-established DSM-IV diagnosis of Asperger syndrome (table 1), “should be given the diagnosis of autism spectrum disorder. Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder should be evaluated for social (pragmatic) communication disorder” [5]. Like ASD, social (pragmatic) communication disorder is characterized by persistent difficulties in the social use of verbal and nonverbal communication. It is distinguished from ASD by the absence of restricted repetitive patterns of behavior, interests, or activities [10]. (See "Terminology, epidemiology, and pathogenesis of autism spectrum disorder", section on 'DSM-5'.)

The International Classification of Diseases, 10th Revision (ICD-10) classifies ASD as “pervasive developmental disorders” and recognizes several subtypes, including childhood autism, atypical autism, and Asperger syndrome (table 2), among others [6]. (See "Terminology, epidemiology, and pathogenesis of autism spectrum disorder", section on 'International Classification of Diseases'.)

Characteristic features of Asperger syndrome (ICD-10) include [6,11]:

- Qualitative deficiencies in reciprocal social interaction
- Restricted, repetitive, stereotyped patterns of behavior, interests, and activities
- Absence of clinically significant delay in language or cognitive development

In this topic review, we will use the term “Asperger syndrome/ASD” to describe children who have autism spectrum disorder and who have generally normal intelligence and/or who have the clinical features compatible with an ICD-10 diagnosis of Asperger syndrome (table 2). In the United States, the DSM-5 terminology is “autism spectrum disorder” with particular specifiers for severity and associated conditions (eg, “without significant language or intellectual impairment”). Using DSM-5 terminology, this topic review describes the clinical features of individuals with ASD of Level 1 severity who do not have intellectual impairment, but who may have language impairment. (See "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'.)

EPIDEMIOLOGY — It is difficult to draw firm conclusions regarding the prevalence of Asperger syndrome [12]. Consistent with literature that fails to find criteria that clearly distinguish Asperger syndrome from autism spectrum disorder (ASD), there are few studies on Asperger syndrome as a distinct clinical entity [13-15]. Prevalence of Asperger syndrome is usually determined by subgrouping within the autism spectrum and varies widely depending upon the diagnostic criteria used to identify cases [16-20].

In systematic reviews, the prevalence of Asperger syndrome is estimated to be 6 cases per 10,000 based upon an autistic disorder-to-Asperger syndrome ratio of 3 or 4 to 1, though there are strong limitations of available data on Asperger syndrome [20,21].

Asperger syndrome is more commonly diagnosed in males. The male:female ratio is approximately to 5:1 [15-20].

ETIOLOGY — Most clinicians accept the hypothesis that Asperger syndrome is an autism spectrum disorder (ASD) and shares the etiology of ASD as a whole. Although the etiology of ASD has yet to be determined, genetic and neurobiologic factors appear to play a prominent role. (See
Case reports and family studies suggest that Asperger syndrome runs in families [22]. Forms of Asperger syndrome have been mapped to chromosome 3q (ASPG1), chromosome 17p (ASPG2), and chromosome 1q21-q22 (ASPG3) [23].

The precise neuroanatomical or neurophysiological differences that underlie Asperger syndrome are not clear [4,24].

CLINICAL FEATURES — The hallmark of Asperger syndrome/autism spectrum disorder (ASD) is atypical social development (eg, lack of social awareness, inability to socialize, lack of interest in socializing) accompanied by extreme behavioral rigidity. Atypical social development can be difficult to identify before approximately four years of age, although other symptoms may be present earlier. In a survey of parents of 156 children with Asperger syndrome, the average age at diagnosis was 11 years [25]. However, parental concerns were present at an average age of 30 months. The average age of diagnosis for Asperger syndrome may decline as the awareness of the autism spectrum grows.

Most of the information in this section will also apply to children with ASD of Level 1 severity who do not have intellectual impairment, but who may have language impairment. (See "Diagnosis of autism spectrum disorder", section on 'Severity level'.)

Early features — During the preschool years, children with Asperger syndrome/ASD may be asymptomatic, may have symptoms that are below the threshold of clinical concern, or may have symptoms related to an associated or comorbid condition (behavior disorder, attention deficit hyperactivity disorder, or anxiety) [26]. (See 'Associated conditions' below.)

Before four years of age, children with Asperger syndrome/ASD may have delayed use of the nonverbal components of communication (pointing to objects of interest, use of facial expressions or gestures). They may show limited, joint attention and may not always respond to social and communication demands successfully. However, because children with Asperger syndrome/usually have normal or near-normal language development and can communicate their wants and needs successfully, these atypical features may not appear salient to caregivers. This may partially account for delays in diagnosis. The presence of repetitive behaviors may result in an earlier diagnosis of autism, with a subsequent shift in diagnosis to Asperger syndrome as the child ages.

After four years of age, when social expectations increase, the atypical social development of children with Asperger syndrome/ASD becomes more noticeable [27]. Parents and teachers may begin to question social development, identifying a lack of social awareness or social ability, one of the two main diagnostic domains of Asperger syndrome/ASD.

Core features

Social skills — Normal social development and interaction encompass the following types of skills:

- Communication – Communicating thoughts, intentions, and emotions verbally and nonverbally.
- Social conventions – Understanding and using social conventions (eg, greetings, waiting turns, following rules, etc).
• Social reasoning ("theory of mind") – Being able to infer the thoughts, intentions, or emotions of others based upon verbal and nonverbal communication (eg, facial expression, gestures, body language).
• On-line modification of social behaviors – Modifying one's behavior during social interactions by considering information obtained through knowledge of the above three components.

The atypical social development of individuals with Asperger syndrome/ASD is usually obvious by the time the child reaches school age if it is not apparent earlier. Day care and preschool staff may notice the child's developmental differences before the parents do, having the benefit of seeing the child interact with same-age peers. The child may solicit the attention of other children or adults inappropriately (eg, by pushing, by approaching the peer while failing to respect personal boundaries, or by immediately initiating a discussion about their preferred topic of interest without checking to see if the peer or adult is listening or even interested). Alternatively, the child may play in the midst of other children without any awareness of what the other children are doing and may not show any ability or effort to join in their play.

During interactions with the clinician, the child may avoid eye gaze, may gaze too intently, or may gaze at the clinician's mouth or other parts of the face or body instead of directing his/her gaze at the eyes [28]. The child may have an invariant expression and may not use gestures as naturally or spontaneously as other individuals. Sometimes, children with Asperger syndrome/ASD make facial expressions or gestures that are exaggerated or "scripted," which also makes them appear awkward.

Language skills — Individuals with Asperger syndrome/ASD may perform normally on formal tests of language function (ie, tests that measure vocabulary, syntax, and semantics). However, they do not understand or use language normally [12,29]. Atypical features of language include atypical prosody (rate, rhythm, tone, and volume of speech) and atypical pragmatics (inappropriate use or absence of the nonverbal components of language, inability to produce responses in conversations that are appropriate to the social context). These atypical features are more likely to be apparent after the age of four years.

Children with Asperger syndrome/ASD have difficulty understanding how the meaning of language changes depending upon the nonverbal components of the language and the context. They may not understand, for example, that a change in tone of voice can change the meaning of a word or sentence, or that the same word can have more than one meaning. This makes it difficult for them to understand metaphors, humor, sarcasm, teasing, or jokes/deception, skills that are present in typically developing children by six to seven years of age [29,30]. Although they may be able to produce grammatically correct sentences, children with Asperger syndrome/ASD may not know how to use changes in prosody to suit the context (eg, speaking more quietly versus speaking more loudly or speaking in a more theatrical tone of voice). When participating in a conversation, they may provide responses that are unrelated to the topic introduced by the social partner. They may not know how to choose the appropriate words or topics for the given social context. For example, they may speak too bluntly, not respecting differences between familiar and nonfamiliar listeners. When telling a story, they may not consider how much or how little the listener already knows. They may be disorganized/tangential or may focus only on a preferred topic.

Children with Asperger syndrome/ASD can be taught to notice ambiguous and social features of language such as those listed in the previous paragraph. However, they generally never fully master all of these subtleties of language. In addition, once they have learned to notice pragmatic functions (eg, sarcasm or humor), they may use them inappropriately (eg, by inappropriately making sarcastic
remarks to older peers or adults or by inappropriately attempting to make a joke), or they may misinterpret the comments of a peer as being sarcastic when they were not.

Behavioral features — Children with Asperger syndrome/ASD may present with behavioral disturbances before their atypical social or language skills are apparent. Behavior disturbances, such as an over-focus on preferred topics or difficulty with organization, can continue throughout life.

Repetitive and perseverative interests are defining features of Asperger syndrome/ASD and are especially obvious when they persist into school age. Although repetitive interests are common in young children, children with Asperger syndrome/ASD are different because their perseverative interests are more specific, unusual, and intense than is the case for their typically developing peers. Interest in mechanical topics, such as trains and cars, or topics from the natural sciences is common. The interest may be very narrow (eg, limited to ceiling fans or vacuum cleaners). They have marked difficulty shifting their attention away from their preferred topic, even when provided with multiple cues, prompts, or requests.

Inflexibility (cognitive rigidity) is another important behavioral feature in individuals with Asperger syndrome/ASD. This feature may be apparent in their choice of conversational topics, but may also be noticeable by their intolerance of changes to their routine or changes/violations of rules of conduct. They are quick to point out differences from what they perceive to be "normal" or "expected." Changes in routines or rules can provoke temper tantrums and/or anxiety.

Despite their adherence to routines, children with Asperger syndrome/ASD can be disorganized and have marked difficulty completing chores or daily tasks. Sometimes this is related to their overfixation on a preferred topic of interest, which diverts their attention away from the more routine tasks of the day. At other times, failure to complete daily tasks may be related to executive dysfunctions, such as remembering, planning, and completing tasks (both routine and novel), and their inability to solve novel problems in real-time situations.

Children with Asperger syndrome/ASD may be unusually sensitive to noises, touch, odors, tastes, or visual stimuli. These sensory sensitivities heighten their overall level of arousal and can contribute to inattention, anxiety, and/or anger management problems.

Other behavior disturbances in children with Asperger syndrome/ASD include anxiety, disruptive behavior disorders (eg, aggression, self-injury), and executive dysfunction, which also occurs in attention deficit hyperactivity disorder [31]. (See 'Associated conditions' below.)

Other features

Clumsiness — Clumsiness is commonly present [14,32], though it is not a defining feature in the ICD-10 criteria (table 2) and was not a defining feature in the DSM-IV criteria (table 1) [6,11,26].

School performance — Children with Asperger syndrome typically perform well with the rote learning that is the focus of schooling during the primary school years. However, school performance often declines in third or fourth grade, when reading comprehension and written expression become more important, a consequence of limited pragmatic language skills and social awareness. Children with Asperger syndrome perform especially poorly in topics not related to their preferred interests and/or when group learning or classroom discussion is required. Also, they can easily become overwhelmed when asked to manage their own schedule, to keep track of their own homework assignments, complete complex assignments such as book reports or essays, etc.
As children with Asperger syndrome enter high school, they may have difficulty understanding concepts that are taught in a typical high school curriculum (e.g., understanding character development or character motivation in works of fiction and historical and social concepts, such as "peace," "justice," "liberty," etc).

Special education supports often are needed by third or fourth grade, though not all school teams recognize the need for such supports because students with Asperger syndrome can perform successfully on standardized measures of educational achievement. Lack of academic supports can lead to frustration and behavior disturbances. (See "Asperger syndrome (a specific autism spectrum disorder): Management and prognosis in children and adolescents", section on 'Reading and writing'.)

Associated conditions — The comorbidity of ASD and other psychiatric conditions is high [33,34].

The frequency of comorbid conditions for individuals with Asperger syndrome is uncertain, given limited research on Asperger syndrome as a distinct clinical entity and problems with case definition. However, several case series suggest that the majority of children and adolescents with Asperger syndrome have a comorbid psychiatric diagnosis or symptoms of a comorbid psychiatric diagnosis [35-38]. In one study of 50 9- to 16-year-old subjects with either Asperger syndrome or high-functioning autism, the rate of comorbid psychiatric conditions was estimated to be 74 percent [39]. Among the comorbid diagnoses, ADHD appears to be more common in preadolescents, and depression in adolescents and adults.

Symptoms of comorbid conditions (e.g., depression, anxiety) may be exacerbated by negative social experiences and increased awareness of differences and social difficulties (e.g., isolation, marginalization, and bullying) [3,40,41].

Psychiatric conditions that are associated with Asperger syndrome/ASD include [26,35-38,42,43]:

- Anxiety disorder; the rate of anxiety in ASD is significantly higher than in community samples, ranging from 8 to 13 percent [44]; there persists some debate about whether or not anxiety is a core feature of autism, a feature due to the impairments related to autism, or a separate condition associated with autism [45,46]
- Oppositional defiant disorder and other disruptive behavior disorders
- Attention deficit hyperactivity disorder (ADHD); the comorbidity of ADHD and autism is in the range of 30 percent [47,48] (see "Attention deficit hyperactivity disorder in children and adolescents: Clinical features and evaluation", section on 'Clinical features')
- Depression and other mood disorders
- Nonverbal learning disability (a condition not recognized by the DSM) (see "Specific learning disabilities in children: Clinical features", section on 'Nonverbal LD')
- Tic disorders (see "Hyperkinetic movement disorders in children", section on 'Tic disorders')
- Sleep disorders, including sleep-onset disturbances, maintenance of sleep, or abnormal sleep architecture [49]; the exact prevalence of sleep disturbance remains unclear, because of inconsistent case definition and case identification [50,51] (see "Assessment of sleep disorders in children")

DIAGNOSTIC APPROACH

Overview — Asperger syndrome/autism spectrum disorder (ASD) affects multiple areas of function. Clinical assessment of children with suspected Asperger syndrome/ASD is most effectively conducted by a multidisciplinary team [2,52]. The primary objective of the diagnostic
assessment is to establish a comprehensive profile of the child's strengths, weaknesses, and challenges to formulate an individualized management plan. (See "Asperger syndrome (a specific autism spectrum disorder): Management and prognosis in children and adolescents").

Role of the primary care provider — Asperger syndrome/ASD may not be suspected by the primary care provider unless the parents have specific concerns. Children with Asperger syndrome and ASD often are able to manage the social demands of a routine pediatric office visit successfully and would not necessarily appear to be "odd" or to lack social awareness, especially if they are intelligent and/or have been given some instruction about how to behave in the clinician’s office.

Questions that may be helpful in eliciting concerns related to the symptoms of Asperger syndrome/ASD include [53]:

- "Did you have concerns regarding your child's language development before the age of three?"
- "Does your child use correct grammar?"
- "Does your child have normal prosody? Prosody includes volume (the loudness or softness of your child’s voice); rate (how quickly or how slowly your child speaks); and rhythm and tone. (Language is a little bit like music. Does your child sound like other children his age?)"
- "When your child developed speech, did it appear to be overly formal or wordy? Does your child use ‘big’ words, or use words that are ‘too complicated’?"
- "Does your child give answers to your questions that are on-topic, or does s/he go ‘off topic’ or always talk about a preferred topic?"
- "Does your child have any unusual interests or preoccupations with certain topics? Does this make it difficult for them to have conversations or play with peers?"
- "Do you have any concerns regarding your child's socialization and play skills? What does s/he like to play with? Whom does s/he like to play with? What do they do together?"
- Some children do not tolerate changes in daily routines, how they play a game, or engage in preferred activities. They might complain, get anxious, or even get angry when changes occur. Does your child show behaviors such as this?

If parents express concerns suggestive of Asperger syndrome/ASD, the clinician can engage the child in an open-ended conversation and observe the child more closely for evidence of atypical social and language behaviors. The pediatric health care provider can ask the child about friendships, what s/he likes to do during play dates or play time, etc. Both the content of the child’s responses and the style of responding can reveal atypical language and social behaviors.

When Asperger syndrome/ASD is suspected, the primary care provider's main roles are to help the family access the resources needed to establish the diagnosis and to advocate for optimal service delivery at school and in the community. Although the primary care provider may be able to diagnose Asperger syndrome/ASD referral to a specialist is warranted to confirm the diagnosis and to assure that all of the child's needs are met. (See "Asperger syndrome (a specific autism spectrum disorder): Management and prognosis in children and adolescents").

If the clinician suspects Asperger syndrome/ASD, the child should be referred for evaluation by a developmental-behavioral pediatrician, a child psychiatrist, a child neurologist, or a neuropsychologist with expertise in autism spectrum disorder for a multidisciplinary evaluation [29]. (See 'Multidisciplinary evaluation' below.)
Referral for a multidisciplinary evaluation often involves a long waiting period. The primary care provider can facilitate the process by helping the parents to gather information in advance of consultation with the specialist. As an example, the parents could be provided with rating scales for ASD, Asperger syndrome, child psychiatric disorders, and/or behavioral disorders. Several scales have been designed specifically for Asperger syndrome (eg, Asperger Syndrome Diagnostic Scale, Gilliam Asperger Disorder Scale). However, their psychometric properties are not always reliable \cite{9,54}, and their utility is debatable given changes in diagnostic classification. A general behavioral screening tool is worthwhile to capture some of the behavioral or atypical features (eg, the Behavior Assessment System for Children [BASC] or the Child Behavior Checklist [CBCL]) and facilitates the assessment of comorbid conditions and other conditions in the differential diagnosis. (See 'Diagnosis' below and "Screening tools for autism spectrum disorder" and "Developmental and behavioral screening tests in primary care").

The primary care provider also can request or recommend psychoeducational and speech/language testing by the child's school district. The main purposes of school testing are to assess the child's cognitive and educational functioning and language skills. It is useful to ask the school team to look at pragmatic functions specifically, and to assess social skills and behavioral regulation (capacity for self-organization, keeping track of homework and of personal belongings, following routine classroom rules and other rules of conduct). Additional areas may warrant evaluation if the child has motor coordination problems or there are other concerns (eg, behavioral or emotional problems).

Finally, the primary care provider can refer parents and patients to support organizations and/or Web sites. (See 'Resources' below.)

Multidisciplinary evaluation — Asperger syndrome/ASD affects multiple areas of function, and clinical assessment of children with suspected Asperger syndrome/ASD is most effectively conducted by a multidisciplinary team \cite{2,29}. The primary objective of the assessment is to establish a comprehensive, detailed profile of the child's strengths, weaknesses, and challenges.

The evaluation should include a history of the child's early development, current symptoms, speech and language assessment, and neuropsychologic testing to differentiate Asperger syndrome/ASD from other conditions in the differential diagnosis (table 3) \cite{2,29}. (See 'Differential diagnosis' below and 'Associated conditions' above.)

- The developmental and health history may help to distinguish Asperger syndrome/ASD from other biological and psychiatric disorders. It should include observations of the child during more and less structured periods. Specific areas for observation and inquiry include \cite{2}:
  - Development of peer relationships and friendships
  - Patterns of special interest and leisure time
  - Presence of obsessions or compulsions
  - Ritualized behaviors
  - Depression and panic attacks or other symptoms of anxiety
  - Capacity for self-awareness and perspective-taking
  - Level of insight into social and behavioral problems
  - Typical reactions in novel situations
  - Social and affective presentation
  - Quality of attachment to family members
  - Ability to infer another person's feelings, intentions, beliefs

- The primary care provider also can request or recommend psychoeducational and speech/language testing by the child's school district. The main purposes of school testing are to assess the child's cognitive and educational functioning and language skills. It is useful to ask the school team to look at pragmatic functions specifically, and to assess social skills and behavioral regulation (capacity for self-organization, keeping track of homework and of personal belongings, following routine classroom rules and other rules of conduct). Additional areas may warrant evaluation if the child has motor coordination problems or there are other concerns (eg, behavioral or emotional problems).

- Finally, the primary care provider can refer parents and patients to support organizations and/or Web sites. (See 'Resources' below.)

- Multidisciplinary evaluation — Asperger syndrome/ASD affects multiple areas of function, and clinical assessment of children with suspected Asperger syndrome/ASD is most effectively conducted by a multidisciplinary team. The primary objective of the assessment is to establish a comprehensive, detailed profile of the child's strengths, weaknesses, and challenges.

The evaluation should include a history of the child's early development, current symptoms, speech and language assessment, and neuropsychologic testing to differentiate Asperger syndrome/ASD from other conditions in the differential diagnosis (table 3). (See 'Differential diagnosis' below and 'Associated conditions' above.)
• Ability to understand ambiguous nonliteral communication (e.g., the ability to understand an expression like "it's raining cats and dogs"; metaphors; sarcasm; etc)

• The physical examination of children with Asperger syndrome/ASD usually is normal. Important aspects of the examination include assessment of dysmorphic features, neurologic examination, and evaluation of the skin for pigmentary changes [9]. (See "Diagnosis of autism spectrum disorder", section on 'Examination'.)

• The speech and language assessment provides a profile of language skills and may differentiate Asperger syndrome/ASD from language disorder or speech sound disorder. Important aspects of the assessment include an assessment of pragmatic language functions, such as nonverbal communication, nonliteral language (e.g., metaphor, humor), content of conversations, and capacity to respond to conversational demands with contingent responses (staying on topic). Prosodic features such as rate, rhythm, and volume of speech are often abnormal [2]. These language features are not always captured in a speech/language assessment that focuses on formal language functions only (vocabulary, grammar and syntax). The tester has to understand pragmatic language functions and probe for them specifically. Pragmatic language tests are subject to significant observer interpretation; and individuals with Asperger syndrome/ASD may perform successfully in the 1:1 testing situation, even though they do not show normal pragmatic language skills when asked to participate in real-time situations, such as classroom discussions or peer interactions.

Children who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder (DSM-5 terminology) may have social (pragmatic) communication disorder [5]. Social (pragmatic) communication disorder is characterized by persistent difficulties in the social use of verbal and nonverbal communication in the absence of restricted repetitive patterns of behavior, interests, or activities [10].

• Neuropsychological assessment is performed to establish the overall level of intellectual functioning, profiles of psychomotor functioning, verbal and nonverbal cognitive strengths and weaknesses, style of learning, and independent living skills [2].

DIAGNOSIS — The standard for making a diagnosis of Asperger syndrome/autism spectrum disorder (ASD) combines clinical judgment (i.e., identification of the clinical features (table 2) using history and clinical observations) and a formal diagnostic interview (i.e., using a diagnostic tool for ASD) [12]. (See "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'.)

In countries that use the DSM-5 system, children who have clinical features of Asperger syndrome generally will be diagnosed with ASD of Level 1 severity without intellectual impairment or without intellectual or language impairment (see "Diagnosis of autism spectrum disorder", section on 'Diagnostic criteria'). In countries that use the ICD-10, we suggest that the ICD-10 diagnostic guidelines be used to make the diagnosis of Asperger syndrome (table 2). Other sets of diagnostic criteria include the Gillberg criteria [14] and the Szatmari criteria [55]. Each of these sets of criteria requires a certain number of items to be fulfilled, but none measures symptom severity [19].

Rating scales for the diagnosis of ASD and additional evaluation of children diagnosed with ASD are discussed separately. (See "Diagnosis of autism spectrum disorder.")

DIFFERENTIAL DIAGNOSIS — The differential diagnosis of Asperger syndrome includes other neurodevelopmental disorders (table 3). Clinical features that help to distinguish Asperger syndrome/autism spectrum disorder (ASD) from comorbid psychiatric conditions include the atypical social and communication skills, and normal cognitive skills (table 3) [56]. (See 'Social
Diagnostic overlap and comorbidity occur in most mental health conditions, and complicate the differential diagnosis; ASD is no exception [57]. Not only are comorbid conditions present in individuals with ASD, symptoms of ASD (eg, weak social skills and others) are present in other mental health conditions (eg, obsessive-compulsive disorder, attention deficit hyperactivity disorder) [58,59].

Superior intelligence can mimic Asperger syndrome/ASD. This is especially true if the child with superior intelligence has comorbid ADHD, learning disability, or anxiety. (See "The gifted child: Characteristics and identification").

RESOURCES — Resources on Asperger syndrome/autism spectrum disorder for providers, patients, and families are provided in the Table (table 4).

INFORMATION FOR PATIENTS — UpToDate offers two types of patient education materials, “The Basics” and “Beyond the Basics.” The Basics patient education pieces are written in plain language, at the 5th to 6th grade reading level, and they answer the four or five key questions a patient might have about a given condition. These articles are best for patients who want a general overview and who prefer short, easy-to-read materials. Beyond the Basics patient education pieces are longer, more sophisticated, and more detailed. These articles are written at the 10th to 12th grade reading level and are best for patients who want in-depth information and are comfortable with some medical jargon.

Here are the patient education articles that are relevant to this topic. We encourage you to print or e-mail these topics to your patients. (You can also locate patient education articles on a variety of subjects by searching on “patient info” and the keyword(s) of interest.)

- Basics topic (see "Patient information: Asperger syndrome (The Basics)")

SUMMARY AND RECOMMENDATIONS

- The hallmark of Asperger syndrome/autism spectrum disorder (ASD) is atypical social development accompanied by extreme behavioral rigidity, which is usually apparent by school age (table 2). (See Clinical features' above.)
- Early features may include delayed use of gestures, joint attention, and pointing and referring to an object of interest, but these may escape notice because of normal language and cognitive development. (See Early features' above.)
- Manifestations of atypical social development include failure to respect personal boundaries, one-sided conversations about a preferred topic of interest, disregarding the level of interest or knowledge of the listener, lack of interest in or effort to join in activities of other children, abnormal gaze (avoidant or overly intense), and abnormal facial expressions (invariant or exaggerated). (See Social skills' above.)
- Children with Asperger syndrome/ASD may have normal vocabulary and grammar skills, but abnormal prosody and pragmatics (functional and social use of language). They understand language literally and have difficulty understanding that meaning may change with context. (See Language skills' above.)
- Behavioral features of Asperger syndrome/ASD include repetitive and perseverative interests, inflexibility, disorganization, difficulty completing chores or daily tasks, anxiety,
disruptive behaviors, executive dysfunction, and sensory sensitivity. (See 'Behavioral features' above.)

- Psychiatric problems that are associated with Asperger syndrome/ASD include anxiety disorder, oppositional defiant disorder, attention deficit hyperactivity disorder, developmental coordination disorder, depression, and other mood disorders. (See 'Associated conditions' above.)

- Assessment of children with suspected autism, including Asperger syndrome/ASD is most effectively conducted by a multidisciplinary team. The central objective is to establish a comprehensive, detailed profile of the child's strengths, weaknesses, and challenges. (See 'Overview' above.)

- The multidisciplinary evaluation should include a history of the child's early development, current symptoms, speech and language evaluation, and neuropsychologic testing to differentiate Asperger syndrome/ASD from other conditions in the differential diagnosis (table 3). (See 'Multidisciplinary evaluation' above.)

- The standard for making a diagnosis of Asperger syndrome/ASD combines clinical judgment (ie, identification of the clinical features using history and clinical observations) and a formal diagnostic interview (ie, using a diagnostic tool for autism or Asperger syndrome). (See 'Diagnosis' above.)

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REFERENCES


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GRAPHICS

Diagnostic criteria for Asperger disorder
A. Qualitative impairment in social interaction, as manifested by at least two of the following:
   1. Marked impairments 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 (eg, 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 are abnormal either in intensity or focus
   2. Apparently inflexible adherence to specific, nonfunctional routines or rituals
   3. Stereotyped and repetitive motor mannerisms (eg, 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 (eg, single words used by age two years, communicative phrases used by age three 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 social interaction), and curiosity about the environment in childhood
F. Criteria are not met for another specific pervasive developmental disorder or schizophrenia


ICD-10 diagnostic criteria for Asperger syndrome
Clinical description
A disorder of uncertain nosological validity, characterized by the same kind of qualitative abnormalities of reciprocal social interaction that typify autism, together with a restricted, stereotyped, repetitive repertoire of interests and activities. The disorder differs from autism primarily in that there is no general delay or retardation in language or in cognitive development. Most individuals are of normal general intelligence but it is common for them to be markedly
clumsy; the condition occurs predominantly in boys (in a ratio of about eight boys to one girl). It seems highly likely that at least some cases represent mild varieties of autism, but it is uncertain whether or not that is so for all. There is a strong tendency for the abnormalities to persist into adolescence and adult life and it seems that they represent individual characteristics that are not greatly affected by environmental influences. Psychotic episodes occasionally occur in early adult life.

Diagnostic guidelines
Diagnosis is based on the combination of a lack of any clinically significant general delay in language or cognitive development plus, as with autism, the presence of qualitative deficiencies in reciprocal social interaction and restricted, repetitive, stereotyped patterns of behavior, interests, and activities. There may or may not be problems in communication similar to those associated with autism, but significant language retardation would rule out the diagnosis.

Diagnostic criteria (for research)
A. A lack of any clinically significant general delay in spoken or receptive language or cognitive development. Diagnosis requires that single words should have developed by two years of age or earlier and that communicative phrases be used by three years of age or earlier. Self-help skills, adaptive behaviour and curiosity about the environment during the first three years should be at a level consistent with normal intellectual development. However, motor milestones may be somewhat delayed and motor clumsiness is usual (although not a necessary diagnostic feature). Isolated special skills, often related to abnormal preoccupations, are common, but are not required for diagnosis.
B. Qualitative abnormalities in reciprocal social interaction (criteria as for autism).
C. An unusually intense circumscribed interest or restricted, repetitive, and stereotyped patterns of behavior, interests and activities (criteria as for autism; however it would be less usual for these to include either motor mannerisms or preoccupations with part-objects or non-functional elements of play materials).
D. The disorder is not attributable to the other varieties of pervasive developmental disorder; schizotypal disorder; simple schizophrenia; reactive and disinhibited attachment disorder of childhood; obsessional personality disorder; obsessive-compulsive disorder.

References:

Differential diagnosis of Asperger syndrome

<table>
<thead>
<tr>
<th>Condition</th>
<th>Features that may help to distinguish the condition from Asperger syndrome (ICD-10 terminology) or ASD without intellectual or language impairment (DSM-5 terminology)</th>
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</thead>
<tbody>
<tr>
<td>Pervasive developmental disorders (this section applies only if the ICD-10 is used for diagnosis)*</td>
<td>Significant abnormalities in language and cognitive skills</td>
</tr>
<tr>
<td>Childhood autism and atypical autism</td>
<td>Some, but not all, of the criteria for childhood autism are present (may have late age of presentation, or atypical or subthreshold symptoms; or may meet only some of the criteria for childhood autism)</td>
</tr>
<tr>
<td>Rett disorder</td>
<td>Female predominance; head growth deceleration; loss of skills; impaired</td>
</tr>
</tbody>
</table>
language skills

Childhood disintegrative disorder
Marked intellectual disability and language impairment; loss of skills

Other disorders
Less impairment in social skills and pragmatic language than in individuals on the autism spectrum; absence of restricted, repetitive patterns of behavior, interests, and activities; normal pragmatic language skills

Attention deficit hyperactivity disorder
Hallucinations; delusions; disorganized speech

Schizophrenia
Lack of pervasive impairments in social development; lack of circumscribed interests; subjective distress about social interactions and about specific situations

Social phobia/anxiety
Lack of impairment in social interaction; normal ability for communication in some settings; absence of restricted, repetitive patterns of behavior, interests, or activities

Obsessive compulsive disorder
Normal social skills; normal pragmatic language; restricted/repetitive behaviors are a source of anxiety rather than a pleasure (as they are in autistic disorder or Asperger syndrome)

Developmental coordination disorder
Absence of restricted, repetitive patterns of behavior, interests, or activities; normal pragmatic language

Oppositional defiant disorder
Absence of restricted, repetitive patterns of behavior, interests, or activities; normal pragmatic language

Language disorder
Normal social skills; absence of restricted, repetitive patterns of behavior, interests, or activities; lack of stereotypies

Social (pragmatic) communication disorder
Absence of restricted, repetitive patterns of behavior, interests, or activities

Nonverbal learning disorder
Less impairment in social skills and pragmatic language than for individuals on the autism spectrum; verbal skills typically stronger than nonverbal skills; lack of restricted, repetitive patterns of behavior, interests, or activities

Social awkwardness related to intellectual giftedness
Normal pragmatic language skills; intense interests are functional, varied, and can be explained by the child; generally enjoy social interaction

Normal social awkwardness
Normal age-appropriate interests and hobbies; normal pragmatic language skills

ICD-10: International Classification of Diseases, 10th Revision; DSM-5: Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition; ASD: autism spectrum disorder.

* The DSM-5 does not recognize diagnostic subtypes of autism spectrum disorder; Asperger syndrome and pervasive developmental disorder—not otherwise specified have been encompassed within the single diagnostic category of autism spectrum disorder.

Δ May occur comorbidly with Asperger disorder.
◊ This is a new disorder, recognized for the first time in DSM-5; it is not included in ICD-10.

Data from


Resources on Asperger syndrome for healthcare providers, educators, patients, and families

Web sites

Asperger's Association of New England (AANE)
www.aane.org
To foster awareness, respect, acceptance, and support for individuals with Asperger syndrome and related conditions and their families

Asperger Syndrome Education Network (ASPEN)
www.aspennj.org
Devoted to education, support, and advocacy

Autism Society
www.autism-society.org
Provides written materials, including newsletters, book lists, and research updates

The Gray Center for Social Learning and Understanding
www.thegraycenter.org
Dedicated to individuals with autism spectrum disorders (ASD) and those who work alongside them to improve mutual understanding

Online Asperger Syndrome Information and Support at MAAP
www.aspergersyndrome.org
Provides information and advice to families of individuals with autism, Asperger syndrome, and pervasive developmental disorders

University Students with Autism and Asperger's Syndrome
www.users.dircon.co.uk/~cns/
Focuses on concerns of young adults in higher education

Books for providers, educators, and parents

- A Parent's Guide to Asperger Syndrome & High-Functioning Autism: How to Meet the Challenges and Help Your Child Thrive by Sally Ozonoff, Geraldine Dawson, and James McPartland

- Asperger Syndrome: A Guide for Educators and Parents by Brenda Smith Myles and Richard Simpson

- Asperger Syndrome: A Guide for Parents and Professionals by Tony Attwood

- Asperger Syndrome: A Practical Guide for Teachers by Val Cumine, Julia Dunlop, and Gill Stevenson

- Asperger Syndrome and Difficult Moments: Practical Solutions for Tantrums, Rage, and Meltdowns by Brenda Smith Myles and Jack Southwick

- Asperger Syndrome and Sensory Issues: Practical Solutions for Making Sense of the World by Brenda Smith Myles, Katherine Tapscott Cook, Nancy E Miller, Louann Rinner,
and Lisa A Robbins

- **Comic Strip Conversations** by Carol Gray
- **The New Social Story Book** by Carol Gray

Books for children
- **Asperger's Huh?** by John Strachan and Rosina G Schnurr
- **Asperger's: What Does it Mean to Me?** by Catherine Faherty and Gary B Mesibov
- **Finding out About Asperger Syndrome, High Functioning Autism and PDD** by Gunilla Gerland
- **This Is Asperger Syndrome** by Elisa Ganon and Brenda Smith Myles

Data from: