DIAGNOSTIC AND STATISTICAL MANUAL OF MENTAL DISORDERS

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TEXT REVISION

DSM-5-TR™

separate diagnosis is not given unless the social communication deficits are clearly in excess of the intellectual limitations.

Unspecified Communication Disorder

F80.9

This category applies to presentations in which symptoms characteristic of communication disorder that cause clinically significant distress or impairment in social, occupational, or other important areas of functioning predominate but do not meet the full criteria for communication disorder or for any of the disorders in the neurodevelopmental disorders diagnostic class. The unspecified communication disorder category is used in situations in which the clinician chooses *not* to specify the reason that the criteria are not met for communication disorder or for a specific neurodevelopmental disorder, and includes presentations in which there is insufficient information to make a more specific diagnosis.

Autism Spectrum Disorder

Autism Spectrum Disorder

Diagnostic Criteria **F84.0**

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by all of 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 interests, emotions, or affect; to failure to initiate or respond to social interactions.
2. Deficits in nonverbal communicative behaviors used for 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.

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).

C. 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).

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 developmental disorder (intellectual disability) or global developmental delay. Intellectual developmental disorder and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual developmental disorder, social communication should be below that expected for 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 meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.

Specify current severity based on social communication impairments and restricted, repetitive patterns of behavior (see Table 2):

Requiring very substantial support

Requiring substantial support

Requiring support

Specify if:

With or without accompanying intellectual impairment

With or without accompanying language impairment

Specify if:

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

Associated with a neurodevelopmental, mental, or behavioral problem

Specify if:

With catatonia (refer to the criteria for catatonia associated with another mental disorder, p. 135, for definition) (**Coding note:** Use additional code F06.1 catatonia associated with autism spectrum disorder to indicate the presence of the comorbid catatonia.)

TABLE 2 Severity levels for autism spectrum disorder (examples of level of support needs)

Severity level	Social communication	Restricted, repetitive behaviors
Level 3 "Requiring very substantial support"	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.	Inflexibility of behavior, extreme difficulty coping with change, or other restricted/repetitive behaviors markedly interfere with functioning in all spheres. Great distress/difficulty changing focus or action.

Severity level	Social communication	Restricted, repetitive behaviors
Level 2 "Requiring substantial support"	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.	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.
Level 1 "Requiring support"	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.	Inflexibility of behavior causes significant interference with functioning in one or more contexts. Difficulty switching between activities. Problems of organization and planning hamper independence.

Recording Procedures

It may be helpful to note level of support needed for each of the two core psychopathological domains in Table 2 (e.g., "requiring very substantial support for deficits in social communication and requiring substantial support for restricted, repetitive behaviors"). Specification of "with accompanying intellectual impairment" or "without accompanying

intellectual impairment" should be recorded next. Language impairment specification should be recorded thereafter. If there is accompanying language impairment, the current level of verbal functioning should be

recorded (e.g., “with accompanying language impairment—no intelligible speech” or “with accompanying language impairment—phrase speech”).

For autism spectrum disorder for which the specifiers “associated with a known genetic or other medical condition or environmental factor” or “associated with a neurodevelopmental, mental, or behavioral problem” are appropriate, record autism spectrum disorder associated with (name of condition, disorder, or factor) (e.g., autism spectrum disorder associated with tuberous sclerosis complex). These specifiers apply to presentations in which the listed condition or problem is potentially relevant to the clinical care of the individual and do not necessarily indicate that the condition or problem is causally related to the autism spectrum disorder. If the associated neurodevelopmental, mental, or behavioral problem meets criteria for a neurodevelopmental or other mental disorder, both autism spectrum disorder and the other disorder should be diagnosed.

If catatonia is present, record separately “catatonia associated with autism spectrum disorder.” For more information, see criteria for catatonia associated with another mental disorder in the chapter “Schizophrenia Spectrum and Other Psychotic Disorders.”

Specifiers

The severity specifiers (see Table 2) may be used to describe succinctly the current symptomatology (which might fall below level 1), with the recognition that severity may vary by context and fluctuate over time. Severity of social communication difficulties and restricted, repetitive behaviors should be separately rated. The descriptive severity categories should not be used to determine eligibility for and provision of services. Indeed, individuals with relatively better skills overall may experience different or even greater psychosocial challenges. Thus, service needs can only be developed at an individual level and through discussion of personal priorities and targets.

Regarding the specifier “with or without accompanying intellectual impairment,” understanding the (often uneven) intellectual profile of a child or adult with autism spectrum disorder is necessary for interpreting diagnostic features. Separate estimates of verbal and nonverbal skill are

necessary (e.g., using untimed nonverbal tests to assess potential strengths in individuals with limited language).

To use the specifier "with or without accompanying language impairment," the current level of verbal functioning should be assessed and described. Examples of the specific descriptions for "with accompanying language impairment" might include no intelligible speech (nonverbal), single words only, or phrase speech. Language level in individuals "without accompanying language impairment" might be further described as speaks in full sentences or has fluent speech. Since receptive language may lag behind expressive language development in autism spectrum disorder, receptive and expressive language skills should be considered separately.

The specifier "associated with a known genetic or other medical condition or environmental factor" can be applied when an individual has a known genetic condition (e.g., Rett syndrome, fragile X syndrome, Down syndrome), a known medical condition (e.g., epilepsy), or a history of environmental exposure in utero to a known teratogen or infection (e.g., fetal valproate syndrome, fetal alcohol syndrome, fetal rubella). This specifier should not be viewed as synonymous with causation of autism spectrum disorder. A condition may be listed as being associated with autism spectrum disorder when it is thought to be potentially clinically relevant or inform care and not because the clinician is asserting a cause. Examples include autism spectrum disorder associated with a unique genomic copy number variant that could be clinically relevant even if the specific abnormality may not have directly caused nor have previously been linked to autism spectrum disorder, or Crohn's disease, which could exacerbate behavioral symptoms.

The specifier "associated with a neurodevelopmental, mental, or behavioral problem" can be applied to indicate problems (e.g., irritability, sleep problems, self-injurious behavior, or developmental regression) that contribute to the functional formulation or are a focus of treatment. Additional neurodevelopmental, mental, or behavioral disorders should also be noted as separate diagnoses (e.g., attention-deficit/hyperactivity disorder; developmental coordination disorder; disruptive behavior, impulse-control,

and conduct disorders; anxiety, depressive, or bipolar disorders; tics or Tourette's disorder; feeding, elimination, or sleep disorders).

Catatonia can occur as a comorbid condition with autism spectrum disorder. In addition to classic symptoms of posturing, negativism (opposition or no response to instructions or external stimuli), mutism, and stupor, an increase or worsening of stereotypy and self-injurious behavior may form part of the symptom complex of catatonia in the setting of autism spectrum disorder.

Diagnostic Features

The essential features of autism spectrum disorder are persistent impairment in reciprocal social communication and social interaction (Criterion A), and restricted, repetitive patterns of behavior, interests, or activities (Criterion B). These symptoms are present from early childhood and limit or impair everyday functioning (Criteria C and D). The stage at which functional impairment becomes obvious will vary according to characteristics of the individual and his or her environment. Core diagnostic features are evident in the developmental period, but intervention, compensation, and current supports may mask difficulties in at least some contexts. Manifestations of the disorder also vary greatly depending on the severity of the autistic condition, developmental level, chronological age, and possibly gender; hence, the term *spectrum*. Individuals without cognitive or language impairment may have more subtle manifestation of deficits (e.g., Criterion A, Criterion B) than individuals with accompanying intellectual or language impairments and may be making great efforts to mask these deficits. Criterion A deficits in social communication will be more subtle if an individual has better overall communication skills (e.g., is verbally fluent, does not have intellectual impairments). Similarly, Criterion B deficits (i.e., restricted patterns of behavior and interests) may be less obvious if the interests are closer to age-typical norms (e.g., Ancient Egypt or trains as compared to wiggling a string). Autism spectrum disorder encompasses disorders previously referred to as early infantile autism, childhood autism, Kanner's autism, high-functioning autism, atypical autism, pervasive developmental disorder not otherwise specified, childhood disintegrative disorder, and Asperger's disorder.

The impairments in social communication and social interaction specified in Criterion A are pervasive and sustained. Diagnoses are most valid and reliable when based on multiple sources of information, including clinician's observations, caregiver history, and, when possible, self-report. Verbal and nonverbal deficits in social communication have varying manifestations, depending on the individual's age, intellectual level, and language ability, as well as other factors such as treatment history and current support. Many individuals have language deficits, ranging from complete lack of speech through language delays, poor comprehension of speech, echoed speech, or stilted and overly literal language. Even when formal language skills (e.g., vocabulary, grammar) are intact, the use of language for reciprocal social communication is impaired in autism spectrum disorder.

Deficits in social-emotional reciprocity (i.e., the ability to engage with others and share thoughts and feelings) may be shown, for example, in young children with little or no initiation of social interaction and no sharing of emotions, along with reduced or absent imitation of others' behavior. What language exists is often one-sided, lacking in social reciprocity, and used to request or label rather than to comment, share feelings, or converse. In older children and adults without intellectual impairments or language delays, deficits in

social-emotional reciprocity may be most apparent in difficulties processing and responding to complex social cues (e.g., when and how to join a conversation, what not to say). Individuals who have developed compensation strategies for some social challenges still struggle in novel or unsupported situations and suffer from the effort and anxiety of consciously calculating what is socially intuitive for most individuals. This behavior may contribute to lower ascertainment of autism spectrum disorder in these individuals, perhaps especially in adult women. Thus, longer assessments, observation in naturalistic settings, and inquiring about any tolls of social interaction may be needed. If asked about the costs of social interaction, for example, these individuals might respond that social interactions are exhausting for them, that they are unable to concentrate because of the

mental effort in monitoring social conventions, that their self-esteem is adversely affected by being unable to be themselves, and so forth.

Deficits in nonverbal communicative behaviors used for social interaction are manifested by absent, reduced, or atypical use of eye contact (relative to cultural norms), gestures, facial expressions, body orientation, or speech intonation. An early feature of autism spectrum disorder is impaired joint attention as manifested by a lack of pointing, showing, or bringing objects to share interest with others, or failure to follow someone's pointing or eye gaze. Individuals may learn a few functional gestures, but their repertoire is smaller than that of others, and they often fail to use expressive gestures spontaneously in communication. Among young people and adults with fluent language, the difficulty in coordinating nonverbal communication with speech may give the impression of odd, wooden, or exaggerated "body language" during interactions. Impairment may be relatively subtle within individual modes (e.g., someone may have relatively good eye contact when speaking) but noticeable in poor integration of eye contact, gesture, body posture, prosody, and facial expression for social communication, or in difficulty maintaining these for sustained periods or when under stress.

Deficits in developing, maintaining, and understanding relationships should be judged against norms for age, gender, and culture. There may be absent, reduced, or atypical social interest, manifested by rejection of others, passivity, or inappropriate approaches that seem aggressive or disruptive. These difficulties are particularly evident in young children, in whom there is often a lack of shared social play and imagination (e.g., age-appropriate flexible pretend play) and, later, insistence on playing by very fixed rules. Older individuals may struggle to understand what behavior is considered appropriate in one situation but not another (e.g., casual behavior during a job interview), or the different ways that language may be used to communicate (e.g., irony, white lies). There may be an apparent preference for solitary activities or for interacting with much younger or older people. Frequently, there is a desire to establish friendships without a complete or realistic idea of what friendship entails (e.g., one-sided friendships or friendships based solely on shared special interests). Relationships with siblings, coworkers, and caregivers are also important to consider (in terms of reciprocity).

Autism spectrum disorder is also defined by restricted, repetitive patterns of behavior, interests, or activities (as specified in Criterion B), which show a range of manifestations according to age and ability, intervention, and current supports. Stereotyped or repetitive behaviors include simple motor stereotypies (e.g., hand flapping, finger flicking), repetitive use of objects (e.g., spinning coins, lining up toys), and repetitive speech (e.g., echolalia, the delayed or immediate parroting of heard words; use of "you" when referring to self; stereotyped use of words, phrases, or prosodic patterns). Excessive adherence to routines and restricted patterns of behavior may be manifest in resistance to change (e.g., distress at apparently small changes, such as taking an alternative route to school or work; insistence on adherence to rules; rigidity of thinking) or ritualized patterns of verbal or nonverbal behavior (e.g., repetitive questioning, pacing a perimeter). Highly restricted, fixated interests in autism spectrum disorder tend to be abnormal in intensity or focus (e.g., a toddler strongly attached to a pan or piece of string; a child preoccupied with vacuum

cleaners; an adult spending hours writing out timetables). Some fascinations and routines may relate to apparent hyper- or hyporeactivity to sensory input, manifested through extreme responses to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or spinning objects, and sometimes apparent indifference to pain, heat, or cold. Extreme reaction to or rituals involving taste, smell, texture, or appearance of food or excessive food restrictions are common and may be a presenting feature of autism spectrum disorder.

Many individuals with autism spectrum disorder without intellectual or language impairments learn to suppress repetitive behavior in public. In these individuals, repetitive behaviors like rocking or finger flicking may serve an anxiolytic or self-soothing function.

Special interests may be a source of pleasure and motivation and provide avenues for education and employment later in life. Diagnostic criteria may be met when restricted, repetitive patterns of behavior, interests, or activities were clearly present during childhood or at some time in the past, even if symptoms are no longer present.

Criterion D requires that the features must cause clinically significant impairment in social, occupational, or other important areas of current functioning. Criterion E specifies that the social communication deficits, although sometimes accompanied by intellectual developmental disorder (intellectual disability), are not in line with the individual's developmental level; impairments exceed difficulties expected on the basis of developmental level.

Standardized behavioral diagnostic instruments with good psychometric properties, including caregiver interviews, questionnaires and clinician observation measures, are available and can improve reliability of diagnosis over time and across clinicians. However, the symptoms of autism spectrum disorder occur as dimensions without universally accepted cutoff scores for what would constitute a disorder. Thus, the diagnosis remains a clinical one, taking all available information into account, and is not solely dictated by the score on a particular questionnaire or observation measure.

Associated Features

Many individuals with autism spectrum disorder also have intellectual and/or language impairment (e.g., slow to talk, language comprehension behind production). Even those with average or high intelligence usually have an uneven profile of abilities. The gap between intellectual and adaptive functional skills is often large. It is common for individuals with autism to have theory-of-mind deficits (i.e., to have difficulty seeing the world from another person's perspective), but these are not necessarily present in all cases. Executive function deficits are also common but not specific, as are difficulties with central coherence (i.e., being able to understand context or to "see the big picture," and thus tending to overfocus on detail).

Motor deficits are often present, including odd gait, clumsiness, and other abnormal motor signs (e.g., walking on tiptoes). Self-injury (e.g., head banging, biting the wrist) may occur, and disruptive/challenging behaviors are more common in children and adolescents with autism spectrum disorder than other disorders, including intellectual developmental disorder. Some individuals develop catatonic-like motor behavior (slowing and "freezing" mid-action), but these are typically not of the magnitude of a

catatonic episode. However, it is possible for individuals with autism spectrum disorder to experience a marked deterioration in motor symptoms and display a full catatonic episode with symptoms such as mutism, posturing, grimacing, and waxy flexibility. The risk period for comorbid catatonia appears to be greatest in the adolescent years.

Prevalence

Frequencies for autism spectrum disorder across the United States have been reported to be between 1% and 2% of the population, with similar estimates in child and adult

samples. However, prevalence appears to be lower among U.S. African American (1.1%) and Latinx children (0.8%) compared with White children (1.3%), even after the effect of socioeconomic resources is taken into account. The reported prevalence of autism spectrum disorder may be affected by misdiagnosis, delayed diagnosis, or underdiagnosis of individuals from some ethnoracial backgrounds. Prevalence across non-U.S. countries has approached 1% of the population (0.62% median global prevalence), without substantial variation based on geographic region or ethnicity and across child and adult samples. Globally, the male:female ratio in well-ascertained epidemiological samples appears to be 3:1, with concerns about underrecognition of autism spectrum disorder in women and girls.

Development and Course

The age and pattern of onset also should be noted for autism spectrum disorder. The behavioral features of autism spectrum disorder first become evident in early childhood, with some cases presenting a lack of interest in social interaction in the first year of life. Symptoms are typically recognized during the second year of life (age 12–24 months) but may be seen earlier than 12 months if developmental delays are severe, or noted later than 24 months if symptoms are more subtle. The pattern of onset description might include information about early developmental delays or any losses of social or language skills. In cases where skills have been lost, parents or

caregivers may give a history of a gradual or relatively rapid deterioration in social behaviors or language skills. Typically, this would occur between ages 12 and 24 months.

Prospective studies demonstrate that in most cases the onset of autism spectrum disorder is associated with declines in critical social and communication behaviors in the first 2 years of life. Such declines in functioning are rare in other neurodevelopmental disorders and may be an especially useful indicator of the presence of autism spectrum disorder. In rare cases, there is developmental regression occurring after at least 2 years of normal development (previously described as childhood disintegrative disorder), which is much more unusual and warrants more extensive medical investigation (i.e., continuous spike and waves during slow-wave sleep syndrome and Landau-Kleffner syndrome). Often included in these encephalopathic conditions are losses of skills beyond social communication (e.g., loss of self-care, toileting, motor skills) (see also Rett syndrome in the section “Differential Diagnosis” for this disorder).

First symptoms of autism spectrum disorder frequently involve delayed language development, often accompanied by lack of social interest or unusual social interactions (e.g., pulling individuals by the hand without any attempt to look at them), odd play patterns (e.g., carrying toys around but never playing with them), and unusual communication patterns (e.g., knowing the alphabet but not responding to own name). Deafness may be suspected but is typically ruled out. During the second year, odd and repetitive behaviors and the absence of typical play become more apparent. Since many typically developing young children have strong preferences and enjoy repetition (e.g., eating the same foods, watching the same video multiple times), distinguishing restricted and repetitive behaviors that are diagnostic of autism spectrum disorder can be difficult in preschoolers. The clinical distinction is based on the type, frequency, and intensity of the behavior (e.g., a child who daily lines up objects for hours and is very distressed if any item is moved).

Autism spectrum disorder is not a degenerative disorder, and it is typical for learning and compensation to continue throughout life. Symptoms are often most marked in early childhood and early school years, with developmental gains typical in later childhood in at least some areas (e.g., increased interest in social interaction). A small proportion of individuals

deteriorate behaviorally during adolescence, whereas most others improve. While it was once the case that only a minority of individuals with autism spectrum disorder lived and worked independently in adulthood, as diagnosis of autism spectrum

disorder is made more frequently in those who have superior language and intellectual abilities, more individuals are able to find a niche that matches their special interests and skills and thus are productively employed. Access to vocational rehabilitation services significantly improves competitive employment outcomes for transition-age youth with autism spectrum disorder.

In general, individuals with lower levels of impairment may be better able to function independently. However, even these individuals may remain socially naive and vulnerable, have difficulties organizing practical demands without aid, and are prone to anxiety and depression. Many adults report using compensation strategies and coping mechanisms to mask their difficulties in public but suffer from the stress and effort of maintaining a socially acceptable facade. Relatively little is known about old age in autism spectrum disorder, but higher rates of co-occurring medical conditions have been documented in the literature.

Some individuals come for first diagnosis in adulthood, perhaps prompted by the diagnosis of autism in a child in the family or a breakdown of relations at work or home. Obtaining detailed developmental history in such cases may be difficult, and it is important to consider self-reported difficulties. Where clinical observation suggests criteria are currently met, autism spectrum disorder may be diagnosed, particularly if supported by a history of poor social and communication skills in childhood. A compelling report (by parents or another relative) that the individual had ordinary and sustained reciprocal friendships and good nonverbal communication skills throughout childhood would significantly lessen the likelihood of a diagnosis of autism spectrum disorder; however, ambiguous or absent developmental information in itself is not sufficient to rule out a diagnosis of autism spectrum disorder.

Manifestations of the social and communication impairments and restricted/repetitive behaviors that define autism spectrum disorder are clear in the developmental period. In later life, intervention or compensation, as well as current supports, may mask these difficulties in at least some contexts. Overall, symptoms remain sufficient to cause current impairment in social, occupational, or other important areas of functioning.

Risk and Prognostic Factors

The best established prognostic factors for individual outcome within autism spectrum disorder are presence or absence of associated intellectual developmental disorder and language impairment (e.g., functional language by age 5 years is a good prognostic sign) and additional mental health problems. Epilepsy, as a comorbid diagnosis, is associated with greater intellectual disability and lower verbal ability.

Environmental. A variety of risk factors for neurodevelopmental disorders, such as advanced parental age, extreme prematurity, or in utero exposures to certain drugs or teratogens like valproic acid, may broadly contribute to risk of autism spectrum disorder.

Genetic and physiological. Heritability estimates for autism spectrum disorder have ranged from 37% to higher than 90%, based on twin concordance rates, and a more recent five-country cohort estimated heritability at 80%. Currently, as many as 15% of cases of autism spectrum disorder appear to be associated with a known genetic mutation, with different de novo copy number variants or de novo mutations in specific genes associated with the disorder in different families. However, even when a known genetic mutation is associated with autism spectrum disorder, it does not appear to be fully penetrant (i.e., not all individuals with that same genetic abnormality will develop autism spectrum disorder). Risk for the majority of cases appears to be polygenic, with perhaps hundreds of genetic loci making relatively small contributions. Whether these findings apply to all racial/ethnic populations equally is unclear, given the limited inclusion of communities of color in genetic research.

Culture-Related Diagnostic Issues

Cultural differences exist in norms for social interaction, nonverbal communication, and relationships, but individuals with autism spectrum disorder are markedly impaired against the norms for their cultural context. Culture influences the perception of autistic behaviors, the perceived salience of some behaviors over others, and the expectations for child behavior and parenting practices. Considerable discrepancies are found in age at diagnosis of autism spectrum disorder in children from diverse ethnoracial backgrounds; most studies find delayed diagnosis among socially oppressed ethnic and racialized children. In addition to being diagnosed later, African American children are more often misdiagnosed with adjustment or conduct disorder than are White children.

Sex- and Gender-Related Diagnostic Issues

Autism spectrum disorder is diagnosed three to four times more often in males than in females, and on average, age at diagnosis is later in females. In clinic samples, females tend to be more likely to show accompanying intellectual developmental disorder as well as epilepsy, suggesting that girls without intellectual impairments or language delays may go unrecognized, perhaps because of subtler manifestation of social and communication difficulties. In comparison with males with autism spectrum disorder, females may have better reciprocal conversation, and be more likely to share interests, to integrate verbal and nonverbal behavior, and to modify their behavior by situation, despite having similar social understanding difficulties as males. Attempting to hide or mask autistic behavior (e.g., by copying the dress, voice, and manner of socially successful women) may also make diagnosis harder in some females. Repetitive behaviors may be somewhat less evident in females than in males, on average, and special interests may have a more social (e.g., a singer, an actor) or “normative” focus (e.g., horses), while remaining unusual in their intensity. Relative to the general population, rates of gender variance have been reported to be increased in autism spectrum disorder, with higher variance in females compared with males.

Association With Suicidal Thoughts or Behavior

Individuals with autism spectrum disorder are at greater risk for suicide death compared with those without autism spectrum disorder. Children with autism spectrum disorder who had impaired social communication had a higher risk of self-harm with suicidal intent, suicidal thoughts, and suicide plans by age 16 years as compared with those without impaired social communication. Adolescents and young adults with autism spectrum disorder have an increased risk of suicide attempts compared with age- and sex-matched control subjects, even after adjustments for demographic factors and psychiatric comorbidities.

Functional Consequences of Autism Spectrum Disorder

In young children with autism spectrum disorder, lack of social and communication abilities may hamper learning, especially learning through social interaction or in settings with peers. In the home, insistence on routines and aversion to change, as well as sensory sensitivities, may interfere with eating and sleeping and make routine care (e.g., haircuts, dental work) extremely difficult. Adaptive skills are typically below measured IQ. Extreme difficulties in planning, organization, and coping with change negatively impact academic achievement, even for students with above-average intelligence. During adulthood, these individuals may have difficulties establishing independence because of continued rigidity and difficulty with novelty.

Many individuals with autism spectrum disorder, even without intellectual developmental disorder, have poor adult psychosocial functioning as indexed by measures such

as independent living and gainful employment. Functional consequences in old age are unknown, but social isolation and communication problems (e.g., reduced help-seeking) are likely to have consequences for health in older adulthood.

Co-occurring intellectual developmental disorder, epilepsy, mental disorders, and chronic medical conditions may be associated with a higher risk of premature mortality for individuals with autism spectrum disorder. Deaths from injury and poisoning are higher than for the general

population, as are deaths from suicide. Drowning is the leading cause of accidental death in children with autism spectrum disorder.

Differential Diagnosis

Attention-deficit/hyperactivity disorder. Abnormalities of attention (overly focused or easily distracted) are common in individuals with autism spectrum disorder, as is hyperactivity. Moreover, some individuals with ADHD may exhibit social communication deficits such as interrupting others, speaking too loudly, and not respecting personal space. Although potentially difficult to discriminate ADHD from autism spectrum disorder, the developmental course and absence of restricted, repetitive behaviors and unusual interests in ADHD help in differentiating the two conditions. A concurrent diagnosis of ADHD should be considered when attentional difficulties or hyperactivity exceeds that typically seen in individuals of comparable mental age, and ADHD is one of the most common comorbidities in autism spectrum disorder.

Intellectual developmental disorder (intellectual disability) without autism spectrum disorder. Intellectual developmental disorder without autism spectrum disorder may be difficult to differentiate from autism spectrum disorder in very young children. Individuals with intellectual developmental disorder who have not developed language or symbolic skills also present a challenge for differential diagnosis, since repetitive behavior often occurs in such individuals as well. A diagnosis of autism spectrum disorder in an individual with intellectual developmental disorder is appropriate when social communication and interaction are significantly impaired relative to the developmental level of the individual's nonverbal skills (e.g., fine motor skills, nonverbal problem solving). In contrast, intellectual developmental disorder is the appropriate diagnosis when there is no apparent discrepancy between the level of social communicative skills and other intellectual skills.

Language disorders and social (pragmatic) communication disorder. In some forms of language disorder, there may be problems of communication and some secondary social difficulties. However, specific language disorder is not usually associated with abnormal nonverbal communication, nor with

the presence of restricted, repetitive patterns of behavior, interests, or activities.

When an individual shows impairment in social communication and social interactions but does not show restricted and repetitive behavior or interests, criteria for social (pragmatic) communication disorder, instead of autism spectrum disorder, may be met. The diagnosis of autism spectrum disorder supersedes that of social (pragmatic) communication disorder whenever the criteria for autism spectrum disorder are met, and care should be taken to enquire carefully regarding past or current restricted/repetitive behavior.

Selective mutism. In selective mutism, early development is not typically disturbed. The affected child usually exhibits appropriate communication skills in certain contexts and settings. Even in settings where the child is mute, social reciprocity is not impaired, nor are restricted or repetitive patterns of behavior present.

Stereotypic movement disorder. Motor stereotypies are among the diagnostic characteristics of autism spectrum disorder, so an additional diagnosis of stereotypic movement disorder is not given when such repetitive behaviors are better explained by the presence

of autism spectrum disorder. However, when stereotypies cause self-injury and become a focus of treatment, both diagnoses may be appropriate.

Rett syndrome. Disruption of social interaction may be observed during the regressive phase of Rett syndrome (typically between ages 1 and 4 years); thus, a substantial proportion of affected young girls may have a presentation that meets diagnostic criteria for autism spectrum disorder. However, after this period, most individuals with Rett syndrome improve their social communication skills, and autistic features are no longer a major area of concern. Consequently, autism spectrum disorder should be considered only when all diagnostic criteria are met.

Symptoms associated with anxiety disorders. The overlap of anxiety symptoms with the core symptoms of autism spectrum disorder can make the classification of anxiety symptoms in autism spectrum disorder challenging.

For example, social withdrawal and repetitive behaviors are core features of autism spectrum disorder but may also be expressions of anxiety. The most common anxiety disorders in autism spectrum disorder are specific phobia (in up to 30% of cases), and social anxiety and agoraphobia (in as many as 17% of cases).

Obsessive-compulsive disorder. Repetitive behavior is a defining feature of both obsessive-compulsive disorder and autism spectrum disorder. In both conditions, repetitive behaviors are considered to be inappropriate or odd. In obsessive-compulsive disorder, intrusive thoughts are often related to contamination, organization, or sexual or religious themes. Compulsions are performed in response to these intrusive thoughts in attempts to relieve anxiety. In autism spectrum disorder, repetitive behaviors classically include more stereotyped motor behaviors, such as hand flapping and finger shaking or more complex behaviors, such as insistence on routines or lining up objects. Contrary to obsessive-compulsive disorder, repetitive behaviors in autism spectrum disorder may be perceived as pleasurable and reinforcing.

Schizophrenia. Schizophrenia with childhood onset usually develops after a period of normal, or near normal, development. A prodromal state has been described in which social impairment and atypical interests and beliefs occur, which could be confused with the social deficits and restricted fixated interests seen in autism spectrum disorder. Hallucinations and delusions, which are defining features of schizophrenia, are not features of autism spectrum disorder. However, clinicians must take into account the potential for individuals with autism spectrum disorder to be concrete in their interpretation of questions regarding the key features of schizophrenia (e.g., “Do you hear voices when no one is there?” ”Yes [on the radio]”). Autism spectrum disorder and schizophrenia can co-occur, and both should be diagnosed when criteria are met.

Personality disorders. In adults without intellectual developmental disorder or significant language impairment, some behaviors associated with autism spectrum disorder may be perceived by others as symptoms of narcissistic, schizotypal, or schizoid personality disorder. Schizotypal personality disorder in particular may intersect with autism spectrum disorder in unusual preoccupations and perceptual experiences, odd thinking and

speech, constricted affect and social anxiety, lack of close friends, and odd or eccentric behavior. The early developmental course of autism spectrum disorder (lack of imaginative play, restricted/repetitive behavior, sensory sensitivities) is most helpful in differentiating it from personality disorders.

Comorbidity

Autism spectrum disorder is frequently associated with intellectual developmental disorder and language disorder (i.e., an inability to comprehend and construct sentences with proper grammar). Specific learning difficulties (literacy and numeracy) are common, as is developmental coordination disorder.

Psychiatric comorbidities also co-occur in autism spectrum disorder. About 70% of individuals with autism spectrum disorder may have one comorbid mental disorder, and 40% may have two or more comorbid mental disorders. Anxiety disorders, depression, and ADHD are particularly common. Avoidant/restrictive food intake disorder is a fairly frequent presenting feature of autism spectrum disorder, and extreme and narrow food preferences may persist.

Among individuals who are nonverbal or have language deficits, observable signs such as changes in sleep or eating and increases in challenging behavior should trigger an evaluation for anxiety or depression, as well as for potential pain or discomfort from undiagnosed medical or dental problems. Medical conditions commonly associated with autism spectrum disorder include epilepsy and constipation.

Attention-Deficit/Hyperactivity Disorder

Attention-Deficit/Hyperactivity Disorder