Pervasive Developmental Disorders

Pervasive Developmental Disorders are characterized by severe and pervasive impairment in several areas of development: reciprocal social interaction skills, communication skills, or the presence of stereotyped behavior, interests, and activities. The qualitative impairments that define these conditions are distinctly deviant relative to the individual's developmental level or mental age. This section contains Autistic Disorder, Rett's Disorder, Childhood Disintegrative Disorder, Asperger's Disorder, and Pervasive Developmental Disorder Not Otherwise Specified. These disorders are usually evident in the first years of life and are often associated with some degree of Mental Retardation, which, if present, should be coded on Axis II. The Pervasive Developmental Disorders are sometimes observed with a diverse group of other general medical conditions (e.g., chromosomal abnormalities, congenital infections, structural abnormalities of the central nervous system). If such conditions are present, they should be noted on Axis III. Although terms like "psychosis" and "childhood schizophrenia" were once used to refer to individuals with these conditions, there is considerable evidence to suggest that the Pervasive Developmental Disorders are distinct from Schizophrenia (however, an individual with Pervasive Developmental Disorder may occasionally later develop Schizophrenia).

299.00 Autistic Disorder

Diagnostic criteria for 299.00 Autistic Disorder

A.A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):1.qualitative impairment in social interaction, as manifested by at least two of the following:a.marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction

b.failure to develop peer relationships appropriate to developmental level

c.a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)

d.lack of social or emotional reciprocity

2.qualitative impairments in communication as manifested by at least one of the following:a.delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)

b.in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others

c.stereotyped and repetitive use of language or idiosyncratic language

d.lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

3.restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:a.encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus

b.apparently inflexible adherence to specific, nonfunctional routines or rituals

c.stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)

d.persistent preoccupation with parts of objects

B.Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

C.The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

Diagnostic Features

The essential features of Autistic Disorder are the presence of markedly abnormal or impaired development in social interaction and communication and a markedly restricted repertoire of activity and interests. Manifestations of the disorder vary greatly depending on the developmental level and chronological age of the individual. Autistic Disorder is sometimes referred to as early infantile autism, childhood autism, or Kanner's autism.

The impairment in reciprocal social interaction is gross and sustained. There may be marked impairment in the use of multiple nonverbal behaviors (e.g., eye-to-eye gaze, facial expression, body postures and gestures) to regulate social interaction and communication (Criterion A1a). There may be failure to develop peer relationships appropriate to developmental level (Criterion A1b) that may take different forms at different ages. Younger individuals may have little or no interest in establishing friendships. Older individuals may have an interest in friendship but lack understanding of the conventions of social interaction. There may be a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., not showing, bringing, or pointing out objects they find interesting) (Criterion A1c). Lack of social or emotional reciprocity may be present (e.g., not actively participating in simple social play or games, preferring solitary activities, or involving others in activities only as tools or "mechanical" aids) (Criterion A1d). Often an individual's awareness of others is markedly impaired. Individuals with this disorder may be oblivious to other children (including siblings), may have no concept of the needs of others, or may not notice another person's distress.

The impairment in communication is also marked and sustained and affects both verbal and nonverbal skills. There may be delay in, or total lack of, the development of spoken language (Criterion A2a). In individuals who do speak, there may be marked impairment in the ability to initiate or sustain a conversation with others (Criterion A2b), or a stereotyped and repetitive use of language or idiosyncratic language (Criterion A2c). There may also be a lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level (Criterion A2d). When speech does develop, the pitch, intonation, rate, rhythm, or stress may be abnormal (e.g., tone of voice may be monotonous or inappropriate to context or may contain questionlike rises at ends of statements). Grammatical structures are often immature and include stereotyped and repetitive use of language (e.g., repetition of words or phrases regardless of meaning; repeating jingles or commercials) or idiosyncratic language (i.e., language that has meaning only to those familiar with the individual's communication style). Language comprehension is often very delayed, and the individual may be unable to understand simple questions or directions. A disturbance in the pragmatic (social use) of language is often evidenced by an inability to integrate words with gestures or understand humor or nonliteral aspects of speech such as irony or implied meaning. Imaginative play is often absent or markedly impaired. These individuals also tend not to engage in the simple imitation games or routines of infancy or early childhood or do so only out of context or in a mechanical way.

Individuals with Autistic Disorder have restricted, repetitive, and stereotyped patterns of behavior, interests, and activities. There may be an encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus (Criterion A3a); an apparently inflexible adherence to specific, nonfunctional routines or rituals (Criterion A3b); stereotyped and repetitive motor mannerisms (Criterion A3c); or a persistent preoccupation with parts of objects (Criterion A3d). Individuals with Autistic Disorder display a markedly restricted range of interests and are often preoccupied with one narrow interest (e.g., dates, phone numbers, radio station call letters). They may line up an exact number of play things in the same manner over and over again or repetitively mimic the actions of a television actor. They may insist on sameness and show resistance to or distress over trivial changes (e.g., a younger child may have a catastrophic reaction to a minor change in the environment such as rearrangement of the furniture or use of a new set of utensils at the dinner table). There is often an interest in nonfunctional routines or rituals or an unreasonable insistence on following routines (e.g., taking exactly the same route to school every day). Stereotyped body movements include the hands (clapping, finger flicking) or whole body (rocking, dipping, and swaying). Abnormalities of posture (e.g., walking on tiptoe, odd hand movements and body postures) may be present. These individuals show a persistent preoccupation with parts of objects (buttons, parts of the body). There may also be a fascination with movement (e.g., the spinning wheels of toys, the opening and closing of doors, an electric fan or other rapidly revolving object). The person may be highly attached to some inanimate object (e.g., a piece of string or a rubber band).

The disturbance must be manifest by delays or abnormal functioning in at least one (and often several) of the following areas prior to age 3 years: social interaction, language as used in social communication, or symbolic or imaginative play (Criterion B). In most cases, there is no period of unequivocally normal development, although in perhaps 20% of cases parents report relatively normal development for 1 or 2 years. In such cases, parents may report that the child acquired a few words and lost these or seemed to stagnate developmentally.

By definition, if there is a period of normal development, it cannot extend past age 3 years. The disturbance must not be better accounted for by Rett's Disorder or Childhood Disintegrative Disorder (Criterion C).

Associated Features and Disorders

Associated descriptive features and mental disorders.

In most cases, there is an associated diagnosis of Mental Retardation, which can range from mild to profound. There may be abnormalities in the development of cognitive skills. The profile of cognitive skills is usually uneven, regardless of the general level of intelligence, with verbal skills typically weaker than nonverbal skills. Sometimes special skills are present (e.g., a 4 1/2-year-old girl with Autistic Disorder may be able to "decode" written materials with minimal understanding of the meaning of what is read [hyperlexia] or a 10-year-old boy may have prodigious abilities to calculate dates [calendar calculation]). Estimates of single-word (receptive or expressive) vocabulary are not always good estimates of language level (i.e., actual language skills may be at much lower levels).

Individuals with Autistic Disorder may have a range of behavioral symptoms, including hyperactivity, short attention span, impulsivity, aggressiveness, self-injurious behaviors, and, particularly in young children, temper tantrums. There may be odd responses to sensory stimuli (e.g., a high threshold for pain, oversensitivity to sounds or being touched, exaggerated reactions to light or odors, fascination with certain stimuli). There may be abnormalities in eating (e.g., limiting diet to a few foods, Pica) or sleeping (e.g., recurrent awakening at night with rocking). Abnormalities of mood or affect (e.g., giggling or weeping for no apparent reason, an apparent absence of emotional reaction) may be present. There may be a lack of fear in response to real dangers, and excessive fearfulness in response to harmless objects. A variety of self-injurious behaviors may be present (e.g., head banging or finger, hand, or wrist biting). In adolescence or early adult life, individuals with Autistic Disorder who have the intellectual capacity for insight may become depressed in response to the realization of their serious impairment.

Associated laboratory findings.

When Autistic Disorder is associated with a general medical condition, laboratory findings consistent with the general medical condition will be observed. There are group differences in some measures of serotonergic activity, but these are not diagnostic for Autistic Disorder. Imaging studies may be abnormal in some cases, but no specific pattern has been clearly identified. EEG abnormalities are common even in the absence of seizure disorders.

Associated physical examination findings and general medical conditions.

Various nonspecific neurological symptoms or signs may be noted (e.g., primitive reflexes, delayed development of hand dominance) in Autistic Disorder. The condition is sometimes observed in association with a neurological or other general medical condition (e.g., fragile X syndrome and tuberous sclerosis).

Seizures may develop (particularly in adolescence) in as many as 25% of cases. Both microcephaly and macrocephaly are observed. When other general medical conditions are present, they should be noted on Axis III.

Specific Age and Gender Features

The nature of the impairment in social interaction may change over time in Autistic Disorder and may vary depending on the developmental level of the individual. In infants, there may be a failure to cuddle; an indifference or aversion to affection or physical contact; a lack of eye contact, facial responsiveness, or socially directed smiles; and a failure to respond to their parents' voices. As a result, parents may be concerned initially that the child is deaf. Young children with this disorder may treat adults as interchangeable, may cling mechanically to a specific person, or may use the parent's hand to obtain desired objects without ever making eye contact (as if it were the hand rather than the person that is relevant). Over the course of development, the child may become more willing to be passively engaged in social interaction and may even become more interested in social interaction. However, even in such instances, the child tends to treat other people in unusual ways (e.g., expecting other people to answer ritualized questions in specific ways, having little sense of other people's boundaries, and being inappropriately intrusive in social interaction). In older individuals, tasks involving long-term memory (e.g., train timetables, historical dates, chemical formulas, or recall of the exact words of songs heard years before) may be excellent, but the information tends to be repeated over and over again, regardless of the appropriateness of the information to the social context. Rates of the disorder are four to five times higher in males than in females. Females with the disorder are more likely, however, to exhibit more severe Mental Retardation.

Prevalence

The median rate of Autistic Disorder in epidemiological studies is 5 cases per 10,000 individuals, with reported rates ranging from 2 to 20 cases per 10,000 individuals. It remains unclear whether the higher reported rates reflect differences in methodology or an increased frequency of the condition.

Course

By definition, the onset of Autistic Disorder is prior to age 3 years. In some instances, parents will report that they have been worried about the child since birth or shortly afterward because of the child's lack of interest in social interaction. Manifestations of the disorder in infancy are more subtle and difficult to define than those seen after age 2 years. In a minority of cases, the child may be reported to have developed normally for the first year (or even 2 years) of life. Autistic Disorder follows a continuous course. In school-age children and adolescents, developmental gains in some areas are common (e.g., increased interest in social functioning as the child reaches school age). Some individuals deteriorate behaviorally during adolescence, whereas others improve. Language skills (e.g., presence of communicative speech) and overall intellectual level are the strongest factors related to ultimate prognosis. Available follow-up studies suggest that only a small percentage of individuals with the disorder go on as adults to live and work independently. In about one-third of cases, some degree of partial independence is possible. The highest functioning adults with Autistic Disorder typically continue to exhibit problems in social interaction and communication along with markedly restricted interests and activities.

Familial Pattern

There is an increased risk of Autistic Disorder among siblings of individuals with the disorder, with approximately 5% of siblings also exhibiting the condition. There also appears to be risk for various developmental difficulties in affected siblings.

Differential Diagnosis

Periods of developmental regression may be observed in normal development, but these are neither as severe or as prolonged as in Autistic Disorder. Autistic Disorder must be differentiated from other Pervasive Developmental Disorders. Rett's Disorder differs from Autistic Disorder in its characteristic sex ratio and pattern of deficits. Rett's Disorder has been diagnosed only in females, whereas Autistic Disorder occurs much more frequently in males. In Rett's Disorder, there is a characteristic pattern of head growth deceleration, loss of previously acquired purposeful hand skills, and the appearance of poorly coordinated gait or trunk movements. Particularly during the preschool years, individuals with Rett's Disorder may exhibit difficulties in social interaction similar to those observed in Autistic Disorder, but these tend to be transient. Autistic Disorder differs from Childhood Disintegrative Disorder, which has a distinctive pattern of severe developmental regression in multiple areas of functioning following at least 2 years of normal development. In Autistic Disorder, developmental abnormalities are usually noted within the first year of life. When information on early development is unavailable or when it is not possible to document the required period of normal development, the diagnosis of Autistic Disorder should be made. Asperger's Disorder can be distinguished from Autistic Disorder by the lack of delay or deviance in early language development. Asperger's Disorder is not diagnosed if criteria are met for Autistic Disorder.

Schizophrenia with childhood onset usually develops after years of normal, or near normal, development. An additional diagnosis of Schizophrenia can be made if an individual with Autistic Disorder develops the characteristic features of Schizophrenia (see Schizophrenia) with active-phase symptoms of prominent delusions or hallucinations that last for at least 1 month. In Selective Mutism, the child usually exhibits appropriate communication skills in certain contexts and does not have the severe impairment in social interaction and the restricted patterns of behavior associated with Autistic Disorder. In Expressive Language Disorder and Mixed Receptive-Expressive Language Disorder, there is a language impairment, but it is not associated with the presence of a qualitative impairment in social interaction and restricted, repetitive, and stereotyped patterns of behavior. It is sometimes difficult to determine whether an additional diagnosis of Autistic Disorder is warranted in an individual with Mental Retardation, especially if the Mental Retardation is Severe or Profound. An additional diagnosis of Autistic Disorder is reserved for those situations in which there are qualitative deficits in social and communicative skills and the specific behaviors characteristic of Autistic Disorder are present. Motor stereotypies are characteristic of Autistic Disorder; an additional diagnosis of Stereotypic Movement Disorder is not given when these are better accounted for as part of the presentation of Autistic Disorder. Symptoms of overactivity and inattention are frequent in Autistic Disorder, but a diagnosis of Attention-Deficit/Hyperactivity Disorder is not made if Autistic Disorder is present.