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Diagnosis and definition of autism and other pervasive developmental disorders

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Introduction

Autism and other pervasive developmental disorders (PDDs) are a phenomenologically related set of neuropsychiatric disorders. These conditions are characterized by patterns of both delay and deviance in multiple areas of development; typically their onset is in the first months of life (APA, 1994; Volkmar & Klin, 2005). Although often associated with some degree of mental retardation, the pattern of developmental and behavioral features differs from that seen in children with mental retardation not associated with PDD in that certain sectors of development, such as social interaction and communication, are most severely affected whereas other areas, such as nonverbal cognitive abilities, may be within normal limits. While the validity of autism has been relatively well established, issues of syndrome boundaries remain the topic of some debate (Bailey *et al.*, 1996). In this chapter, the development of autism as a diagnostic concept, current definitions of the condition and of related diagnostic concepts, and their differentiation from other disorders will be reviewed.

Development of diagnostic concepts

Over the past 150 years, a major point of controversy has been the continuity, or discontinuity, of the severe psychiatric disorders of childhood with the adult psychoses. For example, the great British psychiatrist Maudsley suggested that children, like adults, could exhibit “insanity” (1867). Similarly Kraepelin’s

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concept of dementia praecox, or what we would now term schizophrenia, was extended to children (dementia praecocissima) (DeSanctis, 1906). While some investigators like Potter (1933) urged the use of more stringent definitions for childhood schizophrenia, there was a general assumption of a fundamental continuity of child and adult “psychosis.” The presumption of continuity was based largely on the severity of these conditions with rather little appreciation of the importance of developmental factors in children’s understanding of reality. These issues contributed to the confusion and controversy that surrounded Kanner’s initial (1943) description of the autistic syndrome.

Kanner’s description of autism

Kanner’s description of 11 children with “autistic disturbances of affective contact” was atheoretical and quite lucid; present definitions of the autistic syndrome remain profoundly influenced by it. Kanner reported that his patients exhibited a disorder characterized by a profound lack of social engagement starting from, or shortly after, their birth; as they reached toddlerhood they also exhibited a range of communication problems and unusual responses to the inanimate environment. These children might, for example, not be particularly responsive to the comings and goings of their parents but be exquisitely sensitive to a small change in the inanimate environment or routine, such as a kitchen cupboard left open. Three of the 11 children were mute, but the language of those who did speak was remarkable for echolalia, literalness, and pronoun reversal.

Although not cooperative with formal psychometric assessment, these children could sometimes be engaged on certain subtests of cognitive or developmental tests and seemed to do well. Because of this, and because of their usually attractive appearance, Kanner speculated that they probably had good intellectual potential. In his original report, he suggested that the disorder was different from other psychiatric conditions and not associated with specific medical conditions. Kanner also mentioned that in many instances, parents, usually the fathers, were remarkably successful and that interactions with the child often seemed strange or odd; on the other hand, his emphasis on the apparently congenital nature of the problem made it difficult to attribute the disorder exclusively to parent–child disturbance.

In Kanner’s view, the essential feature of autism was the children’s inability to relate. He put this observation within a developmental context citing the work of Gessel on early social development. The use of Bleuler’s (1911/1950) term “autism” for the idiosyncratic, self-centered thinking observed in schizophrenia was intended by Kanner to suggest the notion of the child’s living in his or her

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own world. However, his use of this word and certain aspects of his original report, when interpreted literally, proved to be false leads.

Early studies and false leads for research

Kanner's use of the term "autism" immediately suggested schizophrenia; this was consistent with the very broad and inclusive views of schizophrenia which then were dominant. Many clinicians and researchers did not question longstanding assumptions about severe psychiatric disturbance in children. Unfortunately this meant that there was considerable confusion about autism with some investigators, e.g. Bender (1947), assuming continuity of "psychotic" conditions in younger children with more typical adult forms of schizophrenia. As this has proved not to be the case, it makes it difficult to interpret much of the early work on autism. The issue of continuity of autism and schizophrenia was only resolved in the 1970s as the work of Rutter (1970) and Kolvin (1971) suggested important differences between the conditions, e.g. in course, family history, and clinical features.

As cases of autism were followed for a decade it became apparent that Kanner's original assumption that autism was not associated with any medical condition was also not correct. Many children with autism exhibit signs of overt central nervous system dysfunction including, most strikingly, seizures. As many as 25% of cases developed epilepsy, particularly during adolescence (Volkmar & Nelson, 1990). Other signs of neurological dysfunction were also often observed. Some reports associated autism with diverse medical conditions including chromosomal abnormalities, prenatal infections, various brain abnormalities, and so on. When taken together with the data on neurological dysfunction, these findings seemed to suggest the importance of multiple potential insults which acted through one or more mechanisms to cause autism (Rutter *et al.*, 1994).

Similarly, Kanner's assumption that children with autism had normal intellectual potentials proved mistaken. This notion was based on the accurate observation that "splinter skills" were present in some children with autism and that the occasional individual with autism had truly unusual (and usually very isolated) abilities in one area (Hermelin, 2001). It took over two decades before clinicians and investigators realized that, if *all* aspects of a test of intelligence were administered, many children with autism scored in the mentally retarded range in terms of overall IQ. For many years, low IQ scores were mistakenly attributed to willful noncompliance. In any case, the notion of preserved intellectual skills in individuals with autism, who have otherwise very limited abilities, endures to the present and is the basis for unproven and sometimes fraudulent treatment. Conversely, recent epidemiological studies have indicated that a much higher

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proportion of children with autism are not mentally retarded than occurred in autism samples 30 to 40 years ago (Fombonne, 2005).

Perhaps, most unfortunately, Kanner's observation of high levels of parental achievement and unusual parent-child interactions led some clinicians to attribute the source of autism to problems in the parent-child relationship or deficits in child care, e.g. Bettelheim (1967). This view was congruent with the ethos of the time which tended to minimize the importance of biological factors in psychiatric conditions. The ideas seemed superficially consistent with the nature of the disorder, i.e. should it not be the case that a problem characterized by deficits in social interaction is caused by deficiencies in social interaction? However, as controlled studies were conducted, it became clear that the parents of children with autism did not exhibit high degrees of psychopathology (Cox *et al.*, 1975) nor specific deficits in infant caregiving (Cantwell *et al.*, 1978). A growing appreciation of the role of *the child* in parent-child interactions also suggested that it was just as reasonable to assume that deviance in such interaction might sometimes be a function of a basic disturbance in the child rather than in the parent, i.e. interactions with a child who produces highly deviant social behaviors will differ from interactions with children who do not have this disability. Similarly, the idea that parents of autistic children tended to have much higher levels of occupational and educational achievement proved mistaken. Studies that controlled for possible bias in case detection and referral (Schopler *et al.*, 1980a; Wing, 1980) have suggested that autism is seen in all social classes. It seems likely that the high levels of education in parents of Kanner's original cases resulted from an understandable referral bias, i.e. that the parents who were able to locate Kanner were those who had access to a greater array of resources. It must, however, be noted that good, recent epidemiological data on this issue have been lacking (Rutter, 2005). The lack of studies on potential cultural and ethnic differences is also noteworthy (Brown & Rogers, 2003).

"Nonautistic" pervasive developmental disorders

As noted above, the issue of continuity of adult and child forms of schizophrenia was, until quite recently, a major source of confusion. But over the past century, various diagnostic concepts other than autism have been proposed for patterns of disturbance in children with severe developmental disorders. Three of these conditions are now officially recognized in both DSM-IV and ICD-10.

Childhood disintegrative disorder

Heller (1908, 1930/1969) proposed the term "dementia infantilis," or what now would be termed childhood disintegrative disorder (CDD), to account

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for children who develop normally for some period prior to profound developmental regression and the development of many “autistic-like” features. This condition (also sometimes referred to as Heller’s syndrome or disintegrative psychosis), appears to be very uncommon; over 100 cases have been reported since Heller’s original (1908) report (see Volkmar *et al.*, 2005). It is, however, likely that cases have frequently been misdiagnosed (Volkmar & Klin, 2005).

Childhood disintegrative disorder is now included in both DSM-IV and ICD-10 (see Appendix 1.A, pp. 21–25). The condition is difficult to distinguish from autism once it develops. However, the pattern of onset – including a dramatic developmental deterioration and onset of various “autistic-like” behaviors in a previously apparently normal child – is very highly distinctive. The outcome appears to be worse than that in autism (Volkmar, 1992). The presumption in DSM-III and III-R had been that such cases typically resulted from the presence of a significant neuropathological process, e.g. a childhood “dementia”; however, a review of the published cases suggests that this is not the case. There is an increased rate of EEG abnormalities and seizure disorders similar to that in autism, but specific medical conditions that might account for the regression are not usually identified. The relationship, if any, of this condition and “late-onset” or “regressive” autism (see below) merits further study (Siperstein and Volkmar, 2004; Volkmar & Klin, 2005).

Asperger’s disorder

In 1944, Hans Asperger, a Viennese medical student unaware of Kanner’s earlier report, suggested the concept of autistic psychopathy or what is now usually termed Asperger’s disorder. Asperger’s description resembled that of Kanner (1943) in some ways, e.g. in the use of the word autism/autistic to describe marked problems in social interaction. However, Asperger suggested that the condition he described was seen only in males, was observed in the face of relatively strong language and cognitive skills, and tended to run in families. Unusual, idiosyncratic interests were common, e.g. affected children would have marked interests in acquiring certain kinds of knowledge. Early research on the condition was largely confined to non-English speaking Europe until Wing’s (1981) influential literature review and case series. Subsequent work has yielded somewhat contradictory results (e.g. Pomeroy, 1991; Szatmari *et al.*, 1990; Tantam, 1991); however, this may be a function of the absence, until recently, of generally recognized definitions. While the continuity of Asperger’s disorder with autism remains the topic of debate (Klin *et al.*, 1995; Schopler, 1985) the condition has now been included in DSM-IV and ICD-10 (see Appendix 1.A, p. 21).

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The overlap of Asperger's disorder with other diagnostic concepts remains an important topic for research (see Klin *et al.*, 2005). One set of issues has to do with the validity of the diagnostic concept as different from "higher functioning autism." Available data on this question remain somewhat contradictory, again reflecting, at least in part, the inconsistencies in approaches to diagnosis (see Klin *et al.*, 2005). Careful reading of the original reports by Kanner (1943) and Asperger (1944; reprinted 1991) as well as a recent review of Asperger's subsequent cases (Hippler & Klicpera, 2003) suggest several possible points of differentiation. Kanner suggested that in autism the condition was congenital; Asperger thought that the syndrome he identified came to attention only after age 3 or 4 years. In Asperger's disorder language skills are usually an area of strength. In contrast, even in higher-functioning individuals with autism, verbal skills tend not to be advanced over nonverbal ones. Several studies have suggested that different IQ profiles characterize the two conditions. Asperger's disorder is associated with higher, often much higher, verbal IQ whereas in autism verbal IQ is either lower or roughly on a par with nonverbal IQ (Klin & Shepard, 1994; Volkmar *et al.*, 1994). Asperger's disorder may also be associated with a characteristic profile on neuropsychological testing referred to as a nonverbal learning disability (Rourke, 1989; Klin & Shepard, 1994). However, the most common finding in comparisons of autism and Asperger's disorder is that whatever feature is used to distinguish the two groups during ascertainment (e.g. severity of autistic characteristics, age of onset, strength of language skills, deficits in visuospatial and motor skills) continues to differentiate them. In the long run, the question will be whether it is scientifically or clinically helpful to classify individuals with these patterns into separate categories of autism and Asperger's disorder or whether it would be better to treat them as part of a greater continuum.

A major series of questions stems from a more complex issue – the broader phenotype of autism (Le Couteur *et al.*, 1996; Pickles *et al.*, 2000). Somewhat paradoxically, the achievement of a consensus on a rigorous definition of autism has led to an awareness of a broader range of difficulties in social interaction, communication, and behavior that affect relatives (see Rutter, 2005 and Chapter 5). As awareness of autism has grown so has awareness of the "broader spectrum" and the large group of individuals with social difficulties. There has been, on the part of the public and media, a tendency to equate such difficulties with "mild" autism and Asperger's disorder; this, and the awareness of the large number of individuals with some social oddity has tended to blur the boundaries between clinical disorders and eccentricity and oddity within the broad range of normal.

It is clear that, compared to work on autism, there has been much less research on the problems of individuals with social difficulties that are more broadly

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defined. In addition, other diagnostic concepts, derived from diverse disciplines, have been developed; thus, terms like semantic–pragmatic language impairment (Bishop & Norbury, 2002), attention deficit disorder plus PDD (Hellgren *et al.*, 1994), right hemisphere learning problems (Weintraub & Mesulam, 1983), and schizoid disorder (Wolff, 2000) have called attention to specific features that overlap with those of PDDs. The specificity of all of these disorders is unclear. An increased understanding of the relationship of Asperger's disorder to all these conditions and the broader phenotype of autism remains an important area of study.

Rett's disorder

Rett (1966) reported an unusual syndrome, observed only in girls where a very brief period (months) of normal development is followed by decelerated head growth, loss of purposeful hand movements, and the development of severe psychomotor retardation. Characteristic symptoms such as breath-holding spells, air-swallowing, and mid-line hand-wringing or hand-washing stereotypies develop and language skills become severely impaired. By preschool, motor involvement is very significant; an apparent loss of social skills is most evident during this period although social interest will subsequently increase.

Rett's syndrome differs from autism in important ways (van Acker *et al.*, 2005); however, because of the regression, repetitive behaviors, and decrease in social interest, there is the possibility for confusion, particularly during the preschool years. Usually, however, the diagnosis is relatively straightforward (see Hagberg *et al.*, 1983). The condition was included within the PDD group in both ICD-10 and DSM-IV because of the potential for confusion and a concern that it should be listed somewhere in the manuals (Rutter, 1994). Given its distinctive pattern there was also a strong sense that a specific etiology might be found.

The condition is relatively rare, affecting perhaps 1 in 15 000 to 1 in 22 000 females (Fombonne, 2005); the condition almost exclusively occurs in females. A role for genetic factors was suggested by recurrence in family members and in monozygotic twins. The recent identification of methyl CpG binding protein 2 as the cause of a majority of cases of classic Rett's syndrome (Amir *et al.*, 1999) may turn out to provide important clues and promising approaches to genetic studies of other conditions such as CDD (Volkmar *et al.*, 2005).

Pervasive developmental disorder not otherwise specified: atypical autism

The term pervasive developmental disorder not otherwise specified (PDD-NOS) (also referred to as atypical personality development, atypical PDD, or atypical autism) was included in DSM-IV to encompass "subthreshold" cases. It is

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intended to describe individuals who have a marked impairment of social interaction and communication difficulties, and/or stereotyped behavior patterns or interests suggestive of a PDD but who do not meet criteria for any of the formally defined disorders in that class. ICD-10 (see Appendix 1.A, pp. 21–24) adopts a somewhat different approach in that the term can be used when a case meets behavioral criteria for autism but fails the onset criteria, or when the onset criteria are met but the behavioral criteria are not, or when individuals appear to have an “autistic-like” illness but meet neither the onset nor behavioral criteria. It seems likely that many possible subtypes are encompassed within this category (Towbin, 2005). Szatmari and colleagues (Mahoney *et al.*, 1998) and other investigators have suggested that this term is used for individuals who meet social and communication criteria for autism but do not have repetitive behaviors or interests. If this distinction is stable across development, it helps increase the homogeneity of a group of individuals with autism spectrum disorder (ASD). Although introduced to encompass “subthreshold” cases in official classifications, the concept also has its origins in earlier notions, e.g. Rank’s concept of atypical personality development (Rank, 1949).

The limited available evidence suggest that children with PDD-NOS probably come to professional attention rather later than those with autism. This may reflect the fact that intellectual and language skills tend to be more preserved (Towbin, 2005). Some have argued that PDD-NOS and Asperger’s disorder are synonymous terms. As with Asperger’s disorder, the validity of PDD-NOS as a separate disorder rather than as a “range” of behaviors will depend on its value in determining etiology or treatment. More research on continuities of all these conditions is needed.

Approaches to the diagnosis of autism

Definitions of autism subsequent to Kanner

The controversy surrounding the nature of autism and the dearth of careful research studies impeded progress for many years. Starting in the 1970s, however, there was a growing appreciation that autism was indeed a distinctive condition, *not* simply the earliest manifestation of childhood schizophrenia. For example, Rutter (1970) also noted the frequency of seizures and the considerable evidence for some degree of neurobiological involvement. In a classic series of clinical studies, Kolvin (1971) demonstrated that autism and childhood schizophrenia differed in clinical features, course, and family history. In addition, there was a growing tendency to de-emphasize the role of theory and establish reliable descriptions of the syndrome in research. As a result of these developments,

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there were various efforts to develop guidelines for the diagnosis of autism which would facilitate research; this approach paralleled attempts in adult psychiatry to develop precise definitions of syndromes for research purposes, e.g. Spitzer Williams (1988).

Of these early attempts to develop more precise categorical definitions, Rutter's (1978) is undoubtedly the most important. This definition was fundamentally grounded in Kanner's early phenomenological description of the condition, but also recognized the importance of subsequent research. Rutter suggested that there were four features essential for the diagnosis of autism:

- (1) an onset prior to 30 months of age;
- (2) impaired social development of a distinctive type which did not simply reflect associated mental retardation;
- (3) impaired communicative development, which again was distinctive and not simply the result of an overall cognitive delay;
- (4) the presence of unusual behaviors subsumed under the concept of "insistence on sameness," i.e. resistance to change, idiosyncratic responses to the environment, and so on.

This definition particularly shaped the first official categorical definition of autism.

The categorical definition of autism**DSM-III**

In DSM-III (APA, 1980) autism was accorded diagnostic status for the first time. This inclusion reflected the body of work on autism which had accumulated over the previous decade. In DSM-III, the condition, termed infantile autism, was included in a new class of disorders, the PDDs. Several other conditions, including a separate category for Childhood-onset pervasive developmental disorder, and another category, termed "residual" autism, were also included in this class. Although the term PDD was rather cumbersome, it achieved relatively wide acceptance. The DSM-III definition of infantile autism was much influenced by Rutter's earlier work and emphasized the onset of serious disturbances in social and communicative development and unusual patterns of environmental responsiveness in early childhood. The recognition of autism in DSM-III was a major advance, as was the availability of an officially recognized definition of the condition.

Unfortunately, the other categories proposed and some of the decisions made were less constructive. Partly in response to the early confusion about autism and schizophrenia, the two conditions were made mutually exclusive. While the available data suggested that the two conditions are not, in fact, commonly

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associated, there is no reason why having autism would necessarily act to *protect* a person from subsequently developing schizophrenia. A few such cases of individuals with autism who then also develop schizophrenia, have, in fact, been observed (Volkmar & Cohen, 1991). Similarly, the term “residual autism” was used in cases where the individual’s disorder had once met criteria for infantile autism but no longer did so. In essence, this approach reflected the fact that the criteria proposed for infantile autism emphasized the way the condition presented in early childhood, e.g. with more “pervasive” social deficits. The system did not adequately address the fact that older children and adults continued to exhibit autism which changed somewhat in its expression over the course of development. The term “residual” also had the unfortunate effect of suggesting that somehow children “outgrew” autism; this clearly is *not* usually the case (Howlin, 2005; Rumsey *et al.*, 1985). As a result of these concerns, major changes were made in the definition of autism in DSM-III-R (APA, 1987).

DSM-III-R

In DSM-III-R, the term pervasive developmental disorders was retained to describe the overarching diagnostic class to which autism was assigned. The more problematic diagnostic concepts, e.g. Childhood-onset PDD and residual autism, were eliminated. The DSM-III-R definition was specifically designed to be more developmentally oriented and to be appropriate to the entire range of syndrome expression over both age and developmental level. This was reflected in the new name “autistic disorder” rather than the DSM-III term “infantile autism.” DSM-III-R included more criteria and a polythetic definition. Because of various concerns, age of onset was not included as an essential diagnostic feature. Criteria in DSM-III-R were arranged developmentally and grouped in three broad categories relating to social development, communication and play, and restricted activities and interests. This last category reflects an expansion of the earlier concept of “insistence on sameness” included in previous diagnostic schemes. For a diagnosis of autism, an individual was required to exhibit at least 8 of the 16 criteria with at least 2 of the social and 1 from each of the two remaining groups. In DSM-III-R only autism and the “subthreshold” category PDD-NOS were included in the PDD class.

The strong developmental orientation of DSM-III-R was a major improvement. Unfortunately it was quickly apparent that the new scheme had resulted in a significantly broadened diagnostic concept (Volkmar *et al.*, 1988; Factor *et al.*, 1989; Hertzog *et al.*, 1990). This broadening was a source of considerable concern for many reasons. It complicated the task of interpreting research (see Rutter & Schopler, 1992 for a discussion). Differences from the pending revision of the