

**DIFFERENTIAL DIAGNOSIS OF AUTISM SPECTRUM
DISORDERS - VALIDATION OF DSM IV CRITERIA**

Register No: 05SLP001

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(Dedicated to.....

... .The Almighty

*Thank you for covering me
and protecting my heart*

You save me

You stay with me

You move-in spite of me

*Only the lonely play with me,
but you set aside kings for my company*

Oh How I'm Loved!

*by your enormous brilliance of mercy
when I feel like the remnants of the day-
there's your love and assurance*

sitting on my shoulders

Perfect peace

is promised

while I keep

you on my mind

You allow me to shine.

CERTIFICATE

This is to certify that this dissertation entitled "*Differential diagnosis of Autism Spectrum Disorders - Validation of DSM IV criteria*" is the bonafide work submitted in part fulfillment for the degree of Master of Science (Speech Language Pathology) of the student (Registration No. 05SLP001). This has been carried out under the guidance of a faculty of this institute and has not been submitted earlier to any other University for the award of any other Diploma or Degree.

Mysore,
April, 2007


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CERTIFICATE

This is to certify that this dissertation entitled "*Differential diagnosis of Autism Spectrum Disorders - Validation of DSM IV criteria*" has been prepared under my supervision and guidance. It is also certified that this dissertation has not been submitted earlier in any other University for the award of any Diploma or Degree.

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DECLARATION

This is to certify that this dissertation entitled "*Differential diagnosis of Autism Spectrum Disorders - Validation of DSM IV criteria*" is the result of my own study under the guidance of Prof. K. C. Shyamala, Head of the Department, Department of Speech Pathology, All India Institute of Speech and Hearing, Mysore, and has not been submitted earlier to any other University for the award of any diploma or degree.

Mysore,
April, 2007

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INTRODUCTION

INTRODUCTION

Autism spectrum disorders (ASD) also known as Pervasive Developmental Disorders (PDD) are set of neuropsychiatric disorders with social and communication dysfunction at its core. American Psychiatric Association (APA) in 1994 defined PDD as a broad and general classification for a group of severe pervasive impairments affecting social interaction and communication often accompanied by stereotyped behaviors, interests and activities. Recently, PDD is also known as Autism Spectrum Disorders (Wing 1988), Since then, both the terms are used interchangeably.

According to DSM-IV (Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition), 1994, the category of PDD includes Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, and Pervasive Developmental Disorder - Not Otherwise Specified.

Autism was first named by Kanner (1943), who described a group of children with an unusual pattern of behavior present from birth or before 30 months. He called this as "Early Infantile Autism." His essential criteria for diagnosis were social aloofness and elaborate repetitive routines. Subsequent epidemiological studies have shown that autism is not, as Kanner first thought, a unique and separate condition occurring in children of otherwise normal development; but, that it is closely related to a range of developmental disorders. These include Asperger's Syndrome, a condition characterized by borderline or normal IQ, social isolation or naive, inappropriate social interaction, intensive interest in

only one or two subjects, a narrow, repetitive life style, limited or inappropriate intonation and body language and poor motor coordination.

Heller (1908) described a rather rare condition called Childhood Disintegrative Disorder (CDD) many years before autism but has only recently been 'officially' recognized. In CDD children develop a condition which resembles autism but only after a relatively prolonged period (usually 2 to 4 years) of clearly normal development. This condition apparently differs from autism in the pattern of onset, course, and outcome (Volkmar, 1994).

Rett's Disorder was first described by an Austrian physician, Andreas Rett, in (1966). Prior to 1983, however, little was known about the syndrome because its occurrence is quite rare. Although Rett's Disorder was thought, at first, to result from the destruction or degeneration of brain tissue, genetic research has indicated that it is caused by the failure of the infant's brain to develop normally. Rett's Disorder has a distinctive onset, course and most commonly seen in girls. The child with this syndrome will develop normally during the first five months of life. After the fifth month, head growth slows down and the child loses whatever purposeful hand movements that had developed during the first five months. After 30 months, the child frequently develops repetitive hand-washing or hand-wringing gestures. 50%-80% of children with the disorder will eventually develop epilepsy. RS is also associated with severe or profound mental retardation.

The clinical picture of autism has been found to vary between and even within individuals, according to intellectual ability and age, so Wing (1988) used the label 'Autistic Spectrum Disorders'. The term ASD was used to introduce the concept of spectrum of disorders in autism and to capture the idea of range of manifestations of the same fundamental disorders. The notion of ASD, apart from including number of conditions also covers the fact that Kanner's original description of the syndrome may not represent 'pure' and simply defined conditions. This description has been modified over time with the recognition that same fundamental disability will have different effects in the same individual over time.

Wing and Gould (1979) conducted research in Camberwell, looking at all the children referred for psychiatric help. They were able to select a group of these children who are socially impaired to others who had equally severed learning difficulties but with no social impairment. They found that these three following areas of development associated with the social impairment, forming the cluster of features that provide the diagnostic criteria for autism.

Wing's (1979) triad symptoms in autism:

- Social - impaired, deviant and extremely delayed social development - especially interpersonal development. The variation may be a form of 'autistic aloofness' to 'active but odd' characteristics.
- Language and communication - impaired and deviant language and communication - verbal and non verbal. Deviant semantic and pragmatic aspects of language.

- Thought and behavior - rigidity of thought and behavior and impoverished social imagination. Ritualistic behavior, reliance on routines, extreme delay or absence of 'pretend play'.

This became the triad of impairments, by which autistic - like conditions were diagnosed. Some of these characteristics are out of keeping with Kanner's original description and children so diagnosed may not have what is known as Kanner's syndrome but they do fall into what Wing (1988) described as the 'Autistic Spectrum Disorders'.

The diagnostic category of ASD refers to a group of disorders characterized by delays in the development of socialization and communication skills. Parents may note symptoms as early as infancy, although the typical age of onset is before 3 years of age. Symptoms may include problems with using and understanding language, difficulty relating to people, objects, and events, unusual play with toys and other objects, difficulty with changes in routine or familiar surroundings, and repetitive body movements or behavior patterns. Autism is the most characteristic and best studied ASD. Children with ASD vary widely in abilities, intelligence, and behaviors. Some children do not speak at all, others speak in limited phrases or conversations, and some have relatively normal language development. Repetitive play skills and limited social skills are generally evident. Unusual responses to sensory information, such as loud noises and lights, are also common.

The assessment of the characteristic features of Autism Spectrum Disorders, and to differentially diagnose these spectrum of disorders, using validate tools is indispensable to work out an individual plan for therapy. Various classificatory systems have evolved over years, the most popularly used being Diagnostic and Statistical Manual of Mental Disorders (DSM) and International Classification of Diseases (ICD).

The ICD has become the international standard diagnostic classification for all general epidemiological and many health management purposes. These include the analysis of the general health situation of population groups and monitoring of the incidence and prevalence of diseases and other health problems in relation to other variables such as characteristics and circumstances of the individuals affected.

International Classification of Diseases - Tenth Edition (ICD-10) was endorsed by the Forty-third World Health Assembly in May 1990 and came into use in World health organization (WHO) Member States as from 1994. The classification is the latest in a series which has its origins in the 1850s. The ICD - 10 (1992) criteria includes childhood autism, atypical autism, Rett's Disorder other Childhood Disintegrative Disorder, Overactive disorder with Mental Retardation and stereotyped movements, Asperger's Syndrome, other Pervasive Developmental Disorder and Pervasive Developmental Disorder unspecified.

DSM is a handbook for mental health professionals that lists different categories of mental disorders and the criteria for diagnosing them, according to the publishing

organization, the APA which included causes of morbidity for the first time. The DSM has gone through five revisions since it was first published in 1952. The last major revision was the DSM - IV published in 1994, although a "text revision" was produced in 2000. In DSM - III (APA, 1980), autism was accorded diagnostic status for the first time. The inclusion reflected the body of work on autism which had accumulated over the period of decade. In DSM - III, the condition, termed 'infantile autism' was included in a new class of disorders, the PDD. As a result of concerns about Diagnostic and Statistical Manual of Mental Disorders - III Revised edition (DSM - III R) definition of autism and an awareness of categories and criteria pending in ICD 10, a large multisite field trial, which included ratings of nearly 1000 cases over 100 clinicians of varying backgrounds and experience (Volkmar et.al., 1994) was undertaken for DSM - IV.

In DSM - IV edition the PDD included, Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, and Pervasive Developmental Disorder - Not Otherwise Specified.

As there is ever prevailing tussle between labeling and profiling of these disorders along with challenging perspectives of ASD as being a spectrum of disorders rather than discrete entities, it indicates the need for a diagnostic system which should show how and where the individual is situated in ASD spectrum. We at AIISH, have a tendency to diagnose these conditions, under the broad descriptive label of 'Autistic Features' but it is very essential to differentially diagnose the ASD.

The present study endeavors to validate DSM - IV criteria to differentially identify ASD. DSM - IV criteria is standardized for American children and it is not available in Indian settings. Thus the need of this study is to validate DSM - IV criteria in Indian context and to see whether it is possible to differentially diagnose ASD using DSM - IV criteria. Though DSM IV - Text Revision is the recent version developed in 2000, this study does not consider it, as there is no change in diagnostic criteria for differential diagnosis of ASD in this edition.

REVIEW OF LITERATURE

REVIEW OF LITERATURE

He wandered about smiling, making stereotyped movements with his fingers, crossing them about in the air. He shook his head from side to side, whispering or humming the same three- note tune. He spun with great pleasure anything he could seize upon to spin... When taken into a room, he completely disregarded the people and instantly went for objects, preferably those that could be spun...He angrily shoved away the hand that was in his way of the foot that stepped on one of his blocks...' (Kanner 1943)

DEVELOPMENT OF CONCEPT OF AUTISM SPECTRUM DISORDERS

The above description of a five year old boy called Donald was written over 60 yrs ago. Kanner saw Donald and made these observations in 1938, and they appear in his landmark paper, published in 1943. The paper was based on the study of 11 cases that Kanner had encountered in his work as a child psychiatrist in USA. He introduced the term 'Early Infantile Autism' for this disorder, hitherto unrecognized as a clinical entity. Kanner (1943) described these 11 cases as what he called as 'autistic disturbances of affective control'. As a result of this study, he decided that these children were sufficiently different from other children he saw, to warrant the identification of a separate condition. He noted that these children had in common 'extreme aloneness from the beginning of life and an anxiously obsessive desire for the preservation of the sameness'.

In 1944 an Austrian physician, Hans Asperger also identified similar group and it was not until 1981 that it received its name as a named syndrome (Wing 1981). He suggested the concept of 'Autistic Psychopathy' in 1994, which is now known as the Asperger's Disorder. By a remarkable coincidence, Asperger and Kanner independently described the same kind of disturbed child. This led to questions about the exclusivity of the two 'syndromes' or whether there were two syndromes at all.

The relationship between the syndromes

a) Evidence for separation of the syndromes:

Kanner never referred to Asperger's writing in any of his papers on autism. Asperger on the other hand, in his paper published in 1979, did discuss Kanner's early 'Infantile Autism' and its relationship with 'Autistic Psychopathy'. Kanner first described language peculiarities, such as echolalia, pronoun reversals and difficulties in generalizing word meanings. The children, Asperger first described, apparently, did not show these features but had clever - sounding language, invented words and generally spoke more like grown ups. These comments suggest that there is something not quite right in the way they used language.

Van Krevelen (1971) produced a scheme of points which he considered as the major distinguishing features that made it unmistakably clear, that Early Infantile Autism and Autistic Psychopathy are two entirely different nosological syndromes. These points were:

1. Early Infantile Autism is manifested in the first month of life, whereas Autistic Psychopathy is not manifested until the third year of life or later;
2. The child with Early Infantile Autism walks earlier than he speaks, speech is retarded or absent, and language is not used to communicate, whereas in Autistic Psychopathy the child walks late but speaks earlier and tries to communicate although in a one - sided manner;
3. In Early Infantile Autism, eye contact is poor because, for the child, other people do not exist and he lives in a world of his own. Whereas the child with Autistic Psychopathy evades eye contact and lives in our world but in his own way;
4. Early Infantile Autism is a psychotic process and the social prognosis is rather poor, whereas Autistic Psychopathy is a personality trait and the social prognosis is rather good.

The major problem with the formulation is that, in clinical practice, the features are not neatly divided as in this account. Most individuals manifest elements from both the lists. Van Krevelen went on to suggest that Autistic Psychopathy is a familial trait. It is transmitted via the male line, whereas, Early Infantile Autism occurs when a child who inherits the trait, also suffers from organic brain damage. Thus he recognized a relationship between the two conditions even though he considered them to be entirely different.

b) Evidence for close relationship:

Bosch (1962) reported that, among people, diagnosed as having typical Asperger's Disorder, there were some, who, if had been seen in their early years would at that time, certainly have been regarded as having Autistic Disorder. Conversely, some children, diagnosed as having Early Infantile Autism began to make progress gradually revealed to have more and more a picture of 'autistic' or 'schizoid psychopathy'. Bosch view seemed to be that Kanner tended to describe more severely affected cases while Asperger described those with milder problems, but there is an overlap between the two.

Schopler (1985) reported that Asperger's Disorder, higher level autism and some cases of 'learning disability' converged in their clinical pictures, and emphasized that no behavioral distinction between the first two has yet been demonstrated.

De Long and Dwyer (1988) reported that there was a high incidence of Asperger Syndrome, in the families of children with IQs above 70, but not in those with lower IQs. Fifteen of the nineteen probands with IQs above 70 had the clinical picture of Asperger's Disorder and showing the DSM - III criteria for Autism or PDD. The authors suggested that high- and low- functioning autism are different conditions. Asperger's Disorder and high functioning autism are largely equivalent and have a predominantly familial etiology, in contrast to low- functioning autism in which there is a high incidence of evidence of neuropathology.

Some have argued that the differential diagnosis of the Asperger's Disorder depends only on IQ, which tend to be low or very low in Kanner's Autism but normal or high (occasionally very high) in Asperger's Disorder. Some appear to believe that the two syndromes exist on a social deficit continuum with Kanner's autism again on a lowermost portion and Asperger's Disorder on the higher range. Even though both of these continuum approaches have considerable clinical credibility, autism is sometimes, albeit very rarely, diagnosed in cases with low IQ and Asperger's Disorder in cases with high IQ.

Tryon et.al. (2006), conducted a study, where parents of 26 children with diagnoses of Asperger's Disorder completed a symptom checklist. This was done to determine whether the children met Diagnostic and Statistical Manual of Mental Disorders-Fourth Edition, Text Revision (DSM-IV-TR) (American Psychiatric Association, 2000) criteria for Autistic Disorder, Asperger's Disorder or Pervasive Developmental Disorder - Not Otherwise Specified. Results showed that almost all (20) met criteria for autism, and a DSM-IV-TR diagnosis of Asperger's disorder could not be confirmed in any child. Further, 95% of the parents whose children did not have a DSM-IV-TR diagnosis of Asperger's disorder agreed with their child's diagnosis of Asperger's disorder. Findings suggest that the hierarchical DSM-IV-TR criteria are not applied by clinicians to diagnose Asperger's disorder because most children with diagnoses of Asperger's disorder actually met DSM-IV-TR criteria for autism, which precludes a

diagnosis of Asperger's disorder. Most experts now agree that autism is a spectrum disorder and Asperger's disorder is actually high-functioning autism.

Wing (1981) used the term Autistic Continuum and later in 1988 she used the label *Autism Spectrum Disorders* to emphasize the wide range of social and communication difficulties. The solitary withdrawn child with little emotional expression is at the one end of the continuum, the passive child who does not resist social interaction lies in the middle, and at the other extreme end of continuum is the child who interacts actively but in an odd way without relating to needs or concerns of the person approached.

The PDD are a phenomenologically related set of neuropsychiatric disorders, characterized by patterns of both delay and deviance in multiple areas of development, their onset typically in the first months of life. The term 'PDD' first came into usage in 1980s to describe a class of disorders, which essentially have in common following characteristics (Mauk, 1993):

1. Impairments in social interaction
2. Impairments in verbal and non verbal communication
3. Impairments in imaginative activity
4. Limited number of interests, and repetitive activities.

The various Autism Spectrum Disorders

a) Asperger's Disorder

Asperger suggested that the condition he observed was seen only in males. He reported these children had relatively strong language and cognitive skills. Unusual, idiosyncratic interests are common. Asperger thought that the syndrome he identified came to attention only after 3 or 4 years of age. Asperger's Disorder is often associated with much higher verbal IQ (Volkmar et.al., 1994).

The main features of children with Asperger's Disorder is that they tend to speak fluently by the time they are five, even if their language development was slow to begin with, and if their language is noticeably odd in its use for communication. Some of these children show dramatic improvement despite having had severe autistic symptoms as toddlers. As they grow older they often become quite interested in other people and thus belie the stereotype of aloof and withdrawn autistic child. Nevertheless they remain inept in their approaches and interactions.

By adolescence many will vaguely realize that they are different from their peers and there is a whole sphere of personal relationships from which they are excluded (Kanner 1971). They may learn many facts about the world, but their knowledge seems to remain curiously fragmented. They somehow fail to put their experience and knowledge together to derive useful meaning from these often unconnected bits of information. It is

a frequent complaint of parents, however that their children, despite sometimes high academic abilities, lack common sense.

As adult, Asperger's Disorder individuals can become, superficially at least, well adapted and some are exceptionally successful. On the whole they tend to remain supremely egocentric and isolated.

Diagnostic criteria:

Distinguishing Asperger's Disorder from autism may be an artificial venture and that the two conditions may, in long run, be treated as one. Nevertheless, so far, the evidence is not unequivocally in favor of such an association. Clinically there is something to be said for the use of the Asperger label for certain relatively high functioning individuals with autistic - type empathy deficits and superficially excellent language skills. Diagnostic criteria for Asperger's Disorder adapted from Gillberg (1991) is given below

1. Severe impairment in reciprocal social interaction (at least two of the following)
 - inability to interact lack of desire to interact with peers
 - lack of appreciation of social cues
 - socially and emotionally inappropriate behavior
2. All - absorbing narrow interest (at least one of the following)
 - Exclusion of other activities
 - Repetitive adherence

- More rote than meaning
3. Imposition of routines and interests (at least one of the following)
 - On self, in aspects of life
 - On others.
 4. speech and language problems (at least three of the following)
 - delayed development
 - superficially perfect expressive language
 - formal, pedantic language
 - odd prosody, peculiar voice characteristics
 - Impairment of comprehension including misinterpretations of literal/implied meanings.
 5. Non - verbal communication problems (at least one of the following)
 - Limited use of gestures
 - Clumsy/gauche body language
 - Limited facial expressions
 - Inappropriate expression
 - Peculiar, stiff gaze
 6. Motor clumsiness: Poor performance on neurodevelopment examination

All six criteria must be met for confirmation of diagnosis.

b) The Autistic Disorder

The term Autism was coined by Bleuler (1911) to designate a category of the thought disorder that is present in schizophrenic syndromes. When Kanner (1943) described infantile autism he used the term differently, although with special reference to schizophrenia which he at first thought was related to infantile autism but later tried to clearly distinguish from Kanner's syndrome. Autism is the most well-known ASD and is estimated to occur in one or two out of every 1,000 people.

Kanner in 1943 described 11 cases and reported the defining features of the syndrome being as follows:

1. Profound autistic withdrawal
2. An obsessive desire for the preservation of sameness
3. A good rote memory
4. An intelligent and pensive memory
5. Mutism or language without real communicative intent
6. Over sensitivity to stimuli
7. A skillful relationship to objects.

A further study of 23 children in 1946 led Kanner to elaborate on the above characteristics, particularly in relation to language. He noted then how apparently mute children might, under condition of stress, utter complete and well articulated phrase. Many of the children with language showed both immediate and delayed echolalia far in

excess of any found in normal language development. They were also characterized in the following ways.

1. Their language is also extremely literal
2. They used simple negation to avoid unpleasant events but not to deny
3. They did not affirm by saying 'yes' but by reputation
4. They showed metaphorical substitutions, transfer of meaning by substitute analogy of the whole for the part and the part for the whole;
5. Their speech showed 'pronominal reversals'

By 1950s Kanner had moved away from detailed descriptions of deviant behavior to produce broader defining characteristics (Eisenberg and Kanner, 1956) which were

1. Extreme isolation
2. Obsessive insistence on the preservation of the sameness
3. Onset of the condition within the first two years of life.

Shah and Firth (1983) reported that though not all, some show 'islets' of special abilities, particularly in the fields of rote memory, music, art and visuospatial skills.

Boucher (1981) reported that many have an impaired memory for recent events. Specifically their memory difficulties impair them to recall past activities in response to 'open' or 'uninformative' questions.

It is the unusual cognitive profile of children with autism that has given rise to widespread speculations that they are indeed of superior intelligence, and just hiding their

phenomenal capacity behind a shell of autism. Unfortunately, a large body of research is agreed that this view of autism is mistaken and that most children with autism, even those showing almost unbelievable splinter skills, are clearly mentally retarded. All have cognitive problems (Rutter 1983).

Diagnostic Criteria:

The earliest confusions however were between autism and childhood psychoses. For many years the term 'childhood psychosis' and 'childhood schizophrenia' were used synonymous for 'early infantile autism' even though autism was deterioration in social functioning but an 'autistic' aloofness from the start. Thus there has to appear lists of 'points' for diagnosis of autism, although it was often called 'childhood schizophrenia' at this stage.

Rutter et al. (1971) gave the following four essential diagnostic criteria

1. Delay in speech and language development
2. An 'autistic - like' failure to develop interpersonal relationships
3. Ritualistic and compulsive phenomena; and
4. Onset before 30 months

These 4 points were reformulated by Rutter (1978) to include a description of these social abnormalities and to take an account of greater understanding about the nature of difficulties in communication and behavior.

1. Failure to come for comfort or a cuddle;
2. Lack of eye gaze giving the appearance of aloofness or distance;
3. Relative failure to become attached to parents;
4. Little or no separation anxiety
5. Sometimes little variation in facial expressions
6. Apparent lack of interest in people
7. Tendency when a toddler to treat all adults in the same way; and
8. Failure to make friends and join group activities

These amended criteria formed the basis of the diagnostic criteria accepted by the WHO and were very similar to American formulations under DSM. The four criteria were

1. Delayed and deviant language development, which has certain defined features and is out of keeping with the child's intellectual level;
2. Impaired social development, which has number of special characteristics and is out of keeping with the child's intellectual level;
3. Insistence on sameness, as shown by stereotyped play patterns, abnormal preoccupations, or resistance to change; and
4. Onset before 30 months.

Each of these attempts at diagnosis so far looked for features that would pick out autism as a separate syndrome. However there are other interactions, which make this process even more difficult. Kanner had originally supposed that all children with autism

are fundamentally intelligent and that apparent delays in development were a direct result of autistic condition. Sadly, this one of the 'facts' about autism that Kanner got wrong and *Rainman* - like autistic children are very rare indeed, even within autistic population.

c) *Childhood Disintegrate Disorder*

Heller (1908) proposed the term 'dementia infantilis' or what now would be termed as Childhood Disintegrative Disorder, to account for children who develop normally for some period prior to profound developmental regression and the development of many autistic like features. More boys than girls appear to be affected. CDD is perhaps 10 times less common than more strictly defined than autism (Volkmar, 1994). Very few children who have an ASD diagnosis meet the criteria for CDD. An estimate based on four surveys of ASD found fewer than two children per 100,000 with ASD could be classified as having CDD. This suggests that CDD is a very rare form of ASD. It has a strong male preponderance.

The pattern of onset in children with CDD is a dramatic developmental deterioration and the onset of various autistic like features in previously apparently normal child is highly distinctive. The outcome appears to be worse than that in autism (Volkmar et.al., 1997). There is an increased rate of electroencephalographic abnormalities and seizure disorders similar to that in autism but specific medical conditions which might account for the regression are not usually identified,.

The following is prominent with the condition:

1. Loss of social skills.
2. Loss of bowel and bladder control.
3. Loss of expressive or receptive language.
4. Loss of motor skills.
5. Lack of play.
6. Failure to develop peer relationships.
7. Impairment in nonverbal behaviors.
8. Delay or lack of spoken language.
9. Inability to initiate or sustain a conversation.

Associated Features:

CDD is usually associated with severe mental retardation. But this may **not** always be present. There also appears to be an increased frequency of EEG abnormalities and seizure disorder

d) Rett's Disorder

Andreas Rett in (1966) described Rett's Disorder. He reported this unusual syndrome, observed only in girls. This disorder is characterized by a very brief period of normal development which is followed by decelerated head growth, and loss of purposeful hand movements and development of severe psychomotor retardation. Rett's Disorder is relatively rare, affecting almost exclusively females, one out of 10,000 to 15,000.

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5. Stereotypic hand movements always present
6. Progressive gait difficulties, with gait and truncal ataxia; some may become ambulatory
7. Language always absent
8. Eye contact present, sometimes very intense
9. Little interest in manipulating objects
10. Seizures in at least 70% in early childhood (various seizures types)
11. Bruxism, hyperventilation with air - swallowing and breath - holding common
12. Choreoathetoid movements and dystonia may be present.

e) PDD - Not otherwise specified

There are another set of children who have marked impairment in social interaction, communication impairment and stereotyped behaviors patterns or interest suggestive of autistic disorder but do not meet the criteria for any of the formally defined disorders in that class, these children are labeled as having Pervasive Developmental Disorders - Not otherwise specified.

EPIDEMIOLOGY OF AUTISM SPECTRUM DISORDERS

Initially several researchers have claimed that autism is either rare or nonexistent or if present is not reported in places such as in India (Lotter, 1980; Sanua, 1984). This notion probably stems from the fact that there hasn't been even a single attempt to find

out prevalence rates of PDD in general population in the Indian subcontinent. There are, however, a few clinic based prevalence studies, case series and case reports which show that health professionals do see cases of PDD.

The earliest report of clinic prevalence of infantile autism was by Hoch, a trained Psychoanalyst, in 1967, she found 2.9% cases were of infantile autism. No diagnostic criteria were used. All the cases were diagnosed clinically (Hoch, 1967).

Subsequently, a series of seven cases were reported from a mental retardation clinic at NIMHANS, Bangalore. The sex ratio of the cases was 1.33:1 and they had an age range of 3 to 12 years (Narayanan, 1978). All the above studies have been from psychiatric units of a general or psychiatric hospitals.

Singhi and Malhi (2001) attempted for the first time in India to identify cases of autism in a paediatric clinic. They reviewed case notes of all the cases registered at the paediatric neurology clinic of Post Graduate Institute of Medical Education and Research, Chandigarh, over a 2 years period. They found 16 children <5 years of age meeting the DSM IV criteria for autism. The cases were referred to them for speech (n=10) and /or developmental delay (n=6). Cases were in a sex ratio of 1.3:1 which is less than that reported elsewhere. Authors had included only those children who were less than 5 years of age, which limits the generalizability of the socio clinical data of their cases.

Childhood Autism Rating Scale's score was in severely autistic range in 62.5% of cases. About 25% of cases had normal development up to at least 18 months of age and after that there was regression in the areas of language and behaviour. It is possible that other

PDDs like Rett's Disorder and CDD may have been misdiagnosed as autism (Singhi & Malhi, 2001).

Srinath and coworkers (Srinath et al, 1989) from NIMHANS, Bangalore did a retrospective review of cases registered from 1981 to 1984 in the child psychiatry clinic. A total of 31 cases met the ICD-9 criteria for autism. Out of these cases only 17 fulfilled the criteria laid down by DSM III, which probably reflects the difference in approach to the diagnosis between two classificatory systems. The cases ranged from 2.5 to 14 years of age. Males constituted the bulk (M: F = 7.5: 1), which is higher than that reported internationally. Most were from well to do families. None had faced or were facing adverse psychosocial situations. All developed symptoms by 30 months. Two patients had co-morbid seizure disorder (Srinath et al, 1989).

Apart from typical autism, there are a few published reports on other PDD, mainly CDD. Malhotra and Singh from PGIMER, Chandigarh published socio clinical and investigation findings of 5 cases of CDD, registered at child and adolescent psychiatry clinic from 1980 to 1989. These cases constituted 0.22% of the total cases (2259) registered during this period. Age, at first contact ranged between 5.5 to 12 years. Regression started at 7.4 years in one, while in all the other cases it started between 3 and 4.5 years of age. Unusually rapid onset (1 to 4 weeks) was a notable finding of this study. EEG showed seizure activity in one while nonspecific rhythm abnormalities were found in two cases. CT scan head was normal in all the cases (Malhotra & Singh, 1993). Jaydeokar (1997) published his observations of a case of CDD in a case report.

Malhotra and Gupta from PGIMER, Chandigarh in a retrospective review of cases registered at child and adolescent psychiatry clinic between 1989 and 1999 found 12 cases of CDD, which is about 0.45% of all the cases. Average age at onset was 3.76 years and average age of presentation to the clinic was 7.46 years. Sex ratio of 5:1 is consistent with other reports. Again a rapid onset of illness was noted which is unusual, as a gradual onset is much more widely seen and reported internationally (Malhotra & Gupta, 2002).

As regard to other PDD there is only one case series of three cases of Asperger's Disorder by Malhotra et al from PGIMER. The cases had been registered from 1989 to 1999 and they fulfilled ICD-10 diagnostic criteria. All the cases were males who exhibited classical symptoms in the form of qualitative abnormalities in social interaction and reciprocity. ADHD was co-morbid in two and MR in one case.

Malhotra et al (2002) reported two cases of RS in the period between 1989 and 1999 at the CAP (Child and Adolescent Psychiatry Clinic), PGIMER, Chandigarh. Both were females with a period of normal development up to 12 and 18 months respectively. Both the cases showed classical symptoms.

In a recently published study Malhotra et al (2003) compared the socio-demographic and clinical profile of PDD patients registered at CAP Clinic, PGIMER, and Chandigarh between 1989 and 1999. Out of 2942 cases 46 cases (1.6%) met ICD-10 criteria for different PDD. 22 cases were of typical autism, 12 cases each were of CDDs

and other PDDs. 5 cases met criteria for atypical autism, 4 were of Asperger's Disorder and the rest were of Rett's Disorder (n=2) and PDD unspecified (n=1). Relatively high proportion (26%) of cases of CDD is notable as it has been suggested that CDD is only about one-tenth as common as autism. Seventy eight percent of the total sample was male, all exhibited classical symptoms, and temperamental variations were noted in the areas of activity, rhythmicity and attention span in most of the cases. Comparisons between the three groups of typical autism (n=22), CDD (n=12) and other PDDs (n=12) on various sociodemographic and clinical parameters showed significant differences on various socio clinical parameters like socioeconomic status, onset of illness, age at onset, temperamental variables, neurotic traits, delay in milestones and intelligence quotient (Malhotra et al, 2003). These findings lend credence to subtyping of PDDs as advocated by current classificatory systems

INTERNATIONAL CLASSIFICATION FOR AUTISM SPECTRUM DISORDERS

The category of PDD was included in classification system in 1980, when it became clear that autism was not psychotic disorder and could not be classified with childhood psychosis. The *Diagnostic and Statistical Manual of Mental Disorders, Third Edition (DSM-III)*, published in 1980, offered the first official definition and description of autism in the United States.

In the *International Statistical Classification of Disease 9th revision (ICD 9)*, the category psychoses with onset in childhood included the subgroup of infantile autism

among others. The use of term 'psychosis' in ICD 9 perpetuated the unfortunate impression that autism was related to adult schizophrenia.

a) Diagnostic and Statistical Manual of Mental Disorders (DSM)

It is a handbook for mental health professionals, that lists different categories of mental disorder and the criteria for diagnosing them, according to the publishing organization the APA. It is used worldwide by clinicians and researchers. It has attracted controversy and criticism as well as praise.

The DSM has gone through five revisions, since it was first published in 1952. The last major revision was the DSM - IV published in 1994, although a "text revision" was produced in 2000. The DSM-V is currently in consultation, planning and preparation, due for publication in approximately 2011.

DSM was developed from an earlier classification system adopted in 1918 to meet the need of the federal Bureau of the Census for uniform statistics from psychiatric hospitals, from categorization systems in use by the US military; and from a survey of the views of 10% of APA members. The manual was 130 pages long and contained 106 categories of mental disorder. The DSM-II was published in 1968, listed 182 disorders, and was 134 pages long. These manuals reflected the predominant psychodynamic psychiatry. Symptoms were not specified in detail for specific disorders, but were seen as reflections of broad underlying conflicts or maladaptive reactions to life problems, rooted in a distinction between neurosis and psychosis (roughly, anxiety/depression broadly in

touch with reality, or hallucinations/delusions appearing disconnected from reality). Sociological and biological knowledge was also incorporated, in a model that did not emphasize a clear boundary between normality and abnormality.

The diagnostic classification is the list of the mental disorders that are officially part of the DSM system. "Making a DSM diagnosis" consists of selecting those disorders from the classification that best reflect the signs and symptoms that are afflicting the individual being evaluated. Associated with each diagnostic label is a diagnostic code, which is typically used by institutions and agencies for data collection and billing purposes.

For each disorder included in the DSM, a set of diagnostic criteria that indicate what symptoms must be present (and for how long) in order to qualify for a diagnosis (called inclusion criteria) as well as those symptoms that must not be present (called exclusion criteria) in order for an individual to qualify for a particular diagnosis. Many users of the DSM find these diagnostic criteria particularly useful because they provide a compact encapsulated description of each disorder. Furthermore, use of diagnostic criteria has been shown to increase diagnostic reliability (i.e., likelihood that different users will assign the same diagnosis). However, it is important to remember that these criteria are meant to be used as guidelines to be informed by clinical judgment and are not meant to be used in a cookbook fashion.

Finally, the third component of the DSM is the descriptive text that accompanies each disorder. The text of DSM-IV systematically describes each disorder under the following headings: "Diagnostic Features"; "Subtypes and/or Specifiers"; "Recording

Procedures"; "Associated Features and Disorders"; "Specific Culture, Age, and Gender Features"; "Prevalence"; "Course"; "Familial Pattern"; and "Differential Diagnosis."

DSM-IV, published in 1994 was the last major revision of the DSM. It was the culmination of a six-year effort that involved over 1000 individuals and numerous professional organizations. Much of the effort involved conducting a comprehensive review of the literature to establish a firm empirical basis for making modifications. Numerous changes were made to the classification (i.e., disorders were added, deleted, and reorganized), to the diagnostic criteria sets, and to the descriptive text based on a careful consideration of the available research about the various mental disorders.

In anticipation of the fact that the next major revision of the DSM (i.e., DSM-V) will not appear until 2010 or later (i.e., at least 16 years after DSM-IV), a text revision of the DSM-IV called DSM-IV-TR was published in July 2000. The primary goal of the DSM-IV-TR was to maintain the currency of the DSM-IV text, which reflected the empirical literature up to 1992. Thus, most of the major changes in DSM-IV-TR were confined to the descriptive text. Changes were made to a handful of criteria sets in order to correct errors identified in DSM-IV.

1. DSM-III:

In DSM - III (APA. 1980), autism was accorded diagnostic status for the first time. The inclusion reflected the body of work on autism which had accumulated over the period of decade. In DSM - III, the condition, termed infantile autism was included in a new class of disorders, the PDD. Several other conditions, including a separate category

for childhood onset PDD and another category, termed 'residual' autism, were included in this class as well. Although the term PDD was rather cumbersome, it achieved a wide acceptance. The DSM - III definition of infantile autism was much influenced by Rutter's earlier work and emphasized the early onset of serious disturbances in social and communicative development and unusual pattern of environmental responsiveness. The recognition of autism in DSM - III was a major advance, as was the availability of an officially recognized definition.

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 in fact, commonly associated there seems no reason why having autism would necessarily act to protect a person from subsequently developing schizophrenia. A few cases such as, i.e. of individuals with autism who then also develop schizophrenia, have, in fact have been observed (Volkmar & Cohen, 1991). Similarly, the term 'residual autism' was used in cases where the individual disorder has 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 did indeed emphasize the way the condition presented in early childhood, e.g. with more 'pervasive' social deficits.

The system did not adequately address the fact that the older children and adults continue to exhibit autism which changed somewhat in its expression over the course of development. The term 'residual' also had the most unfortunate effect of suggesting that

somehow children 'outgrew' autism; this clearly is not the case(Cohen et al., 1987) as a result of these concerns relatively major changes were made in the definition of autism in DSM - III -R(APA, 1987)

2. *DSM - III-R:*

In DSM - III - R, the term PDD 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 term DSM - III term 'infantile autism'. DSM - III - R included more criteria and polymeric definition, because of various concerns, age of onset was not included as essential diagnostic features although age of onset could be specified. Criteria in DSM - III - R was arranged developmentally and grouped into three broad categories relating to

Social development

Communication and play

Restricted activities and interest.

This last category reflects the earlier concept of 'insistence on sameness' included in previous diagnostic schemes. For a diagnosis of autism, an individual was required to exhibit atleast eight of the sixteen criteria with atleast two from social and one from each

of the remaining groups. In DSM - III - R, only autism and the 'subthreshold' category PDD - NOS were included in the PDD class. The strong development orientation of DSM - III - R was a major improvement. Unfortunately, it was quickly apparent that the new scheme resulted in a significantly broadened diagnostic concept. This broadening was a source of considerable concerns for many reasons. The task of interpreting research using DSM - III - R would be complicated.

3. *DSM-IV*

As a result of concerns about the DSM - III - R, definition of autism and an awareness of the categories and the criteria pending in ICD - 10, a large multisite field trial was undertaken for DSM - IV. Consistent with previous research, the DSM - III - R system was noted to have high false positive rate, particularly in individuals with high levels of mental handicap. This was true regardless of whether DSM - III - R was compared with the clinician's best judgment (clinical diagnosis) or clinical ratings of other diagnostic criteria (e.g. for DSM - III and ICD - 10), and when alternative method of judging the probability of 'caseness' were employed.

While the DSM - III definition (in its 'lifetime' sense) had some advantages, it was much less developmentally oriented than DSM - III - R the ICD - 10 systems worked reasonably well but tend to err on the side of over stringency. The large number and detail of the research criteria were of concern for a system like DSM - IV which was meant for both clinical and research use. Together, recent research and data collected in

the field trial indicated reduction in the number and details of the ICD - 10 criteria which still yielded good results even when less experienced clinicians were using the criteria (Lord et al., 1994). Thus a modified version of original ICD - 10 draft criteria for autism was proposed for both the American (DSM - IV) and the international (ICD - 10) systems. Although primarily focused on the definition of autism, the result of field trial also provided some support for the inclusion, as in ICD - 10, of RS, CDD and AS in the FDD class in DSM-IV.

For a diagnosis of autism, a total of at least six criteria (from impairments in social interaction, communication and restricted interests and activities) are required, with at least two social impairment criteria present. By definition the condition must have its onset before 3 years of age not be either due to Rett's Disorder or CDD. The choice of 3 years as an arbitrary cut - off point for the onset was consistent with previous research (e.g. Rogers & DiLalla 1990) and the results of the field trial of autism (Volkmar et al., 1994) which suggested that only very rarely do a behavioral syndrome resembling autism have its onset after 3 years. The convergence of the USA and the international definitions of autism should enhance clinical work as well as research.

b) International Classification of Diseases (ICD)

ICD-10 was endorsed by the Forty-third World Health Assembly in May 1990 and came into use in WHO Member States as from 1994. The classification is the latest in a series which has its origins in the 1850s. The first edition, known as the International List of Causes of Death, was adopted by the International Statistical Institute in 1893.

WHO took over the responsibility for the ICD at its creation in 1948 when the Sixth Revision, which included causes of morbidity for the first time.

The ICD has become the international standard diagnostic classification for all general epidemiological and many health management purposes. These include the analysis of the general health situation of population groups and monitoring of the incidence and prevalence of diseases and other health problems in relation to other variables such as the characteristics and circumstances of the individuals affected.

It is used to classify diseases and other health problems recorded on many types of health and vital records including death certificates and hospital records. In addition to enabling the storage and retrieval of diagnostic information for clinical and epidemiological purposes, these records also provide the basis for the compilation of national mortality and morbidity statistics by WHO Member States.

ICD-10:

The ICD 10 criteria includes childhood autism, atypical autism, Rett's Disorder, other CDD, overactive disorder with mental retardation and stereotyped movements, Asperger's Disorder, other PDD and PDD unspecified. The ICD 10 worked reasonably well but tended to err on the side of over stringency. Together recent research and data collected in the field trial indicated reductions in the number and details of ICD 10 criteria. As a result of concerns about DSM III R definition of autism and an awareness of categories and criteria pending in ICD 10, a large multisite field trial was undertaken

for DSM - IV. This field trial included ratings of nearly 1000 cases over 100 clinicians of varying backgrounds and experience (Volkmar et.al., 1994).

In DSM - IV edition the pervasive developmental disorders include, Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, and Pervasive Developmental Disorder - Not Otherwise Specified.

METHOD

METHOD

The present study endeavors to validate DSM-IV criteria in Indian context to differentially diagnose ASD.

PROCEDURE

The study was carried out in four phases:

Phase 1: Subject selection

SUBJECTS: In this phase, 100 subjects, with the male to female ratio of 4:1, between the age range 1 - 6 yrs, who were already diagnosed as having Autistic features, at All India Institute of Speech and Hearing were selected. Subjects selected would not have attended therapy for more than 6 - 12 months.

Phase 2: Construction of DSM questionnaire:

TOOLS USED: The DSM - IV criteria for ASD was taken up and all the criteria were listed irrespective of the disorder. The 26 listed criteria were then divided based on five domains as Medical history, Language skills, Social skills, Behavioral skills and others. Finally the 26 criteria were converted into interrogative form.

Phase 3: Administration of the test

Using DSM - IV questionnaire, information on presence of autistic features were collected through parental or caregiver interviews in addition to which information from already available material (case files) was considered.

Phase 4: Finalization of DSM - IV criteria

Based on the subject study, DSM - IV questionnaire was finalized.

This phase has 2 steps;

Step 1:

In DSM - IV there are certain criteria which were not overlapping among the disorders. These criteria were considered as 'crucial features' and other features, overlapping among the disorders were considered as 'overlapping features'. Based on these crucial features and overlapping features children were classified into different categories of ASD.

Step 2:

The overlapping features were listed. There were totally 14 features overlapping among the Asperger's Disorder, Autistic Disorder and Childhood Disintegrative

Disorder. (There are no overlapping features for Rett's Disorder; it has only 8 crucial features). The presence of each criterion in a number of children in each group (Asperger's Disorder, Autistic Disorder and CDD) was determined. This determined value was then subjected to Chi - square analysis. The above determined value was considered as observed frequencies from which the expected frequencies were drawn. Chi - square was done to find, if there exists any significant difference between the expected frequencies and observed frequencies. The null hypothesis drawn would be,

Null hypothesis 1 - There are no significant differences between the three categories of ASD (Asperger's Disorder, Autistic Disorder and CDD) based on the 14 overlapping features.

Based on the presence or absence of significant difference between expected frequencies and observed frequencies, null hypothesis would be accepted or rejected i.e., it would be concluded whether the three groups ASD (Asperger's Disorder, Autistic Disorder and CDD) can be identified as distinct groups based on the 14 overlapping features in DSM - IV criteria.

Similarly the presence of each criterion in a number of males and females i.e., with respect to gender would be determined. This determined value was considered as observed frequencies from which the expected frequencies were calculated. Chi - square was done to find if there is any significant difference between the expected frequencies and observed frequencies. Based on this, the null hypothesis - 2

Null hypothesis 2 - There are no significant differences between the three categories of ASD (Asperger's Disorder, Autistic Disorder and CDD) in terms of gender based on the 14 overlapping features.

From the presence or absence of significant difference between the expected frequencies and observed frequencies, null hypothesis would be accepted or rejected, i.e., it would be concluded whether, there exists any gender difference in grouping of children into different ASD using DSM - IV criteria.

RESULTS AND DISCUSSION

RESULTS AND DISSCUSSION

The study was embarked on with the purpose of validating DSM-IV, 1994 to differentially diagnose ASD. This study is a retrospective epidemiological study, as it determines the number of children in each category of ASD.

Results of the study are discussed in 2 steps, as follows

STEP 1:

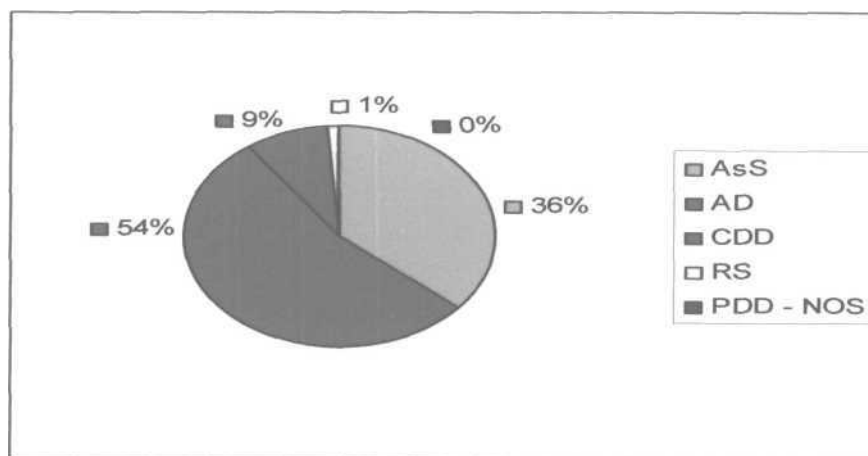
Using the DSM-IV questionnaire the 100 children (80 male subjects and 20 female subjects) were classified into different categories of Autism Spectrum Disorders (Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, and Pervasive Developmental Disorder - Not Otherwise Specified.). Table 1 shows the number and percentage of children in each category of ASD among 100 subjects. Similarly Pie Chart 1 shows the distribution of different categories of ASD

Table 1: Number and percentage of individuals in different categories of Autism

Spectrum Disorders:

Categories of ASD	Number of Subjects (100)	Percentage
Asperger's Disorder	36	36%
Autistic Disorder	54	54%
Childhood Disintegrative Disorder	9	9%
Rett's Disorder	1	1%
Pervasive Developmental Disorders Not otherwise specified	0	0%

Pie - chart 1: Distribution of the different categories of Autism Spectrum Disorders among 100 subjects:



AsS - Asperger Syndrome; AD - Autistic Disorder; CDD - Childhood Disintegrative Disorder; RS - Rett's Syndrome; PDD - NOS - Pervasive Developmental Disorders - Not otherwise specified.

Table 2 & 3 shows the number of males and females respectively in each category of ASD. Pie chart 2 & 3 shows the distribution of males and females respectively in different categories of ASD.

Table 2: Number and percentage of male subjects in different categories of Autism

Spectrum Disorders:

Categories of ASD	Number of Subjects (80)	Percentage
Asperger's Disorder	31	38.75%
Autistic Disorder	41	51.25%
Childhood Disintegrative Disorder	8	10%
Rett's Disorder	0	0%
Pervasive Developmental Disorders Not otherwise specified	0	0%

Pie - chart 2: Distribution of different categories of Autism Spectrum Disorders among the male subjects (80):

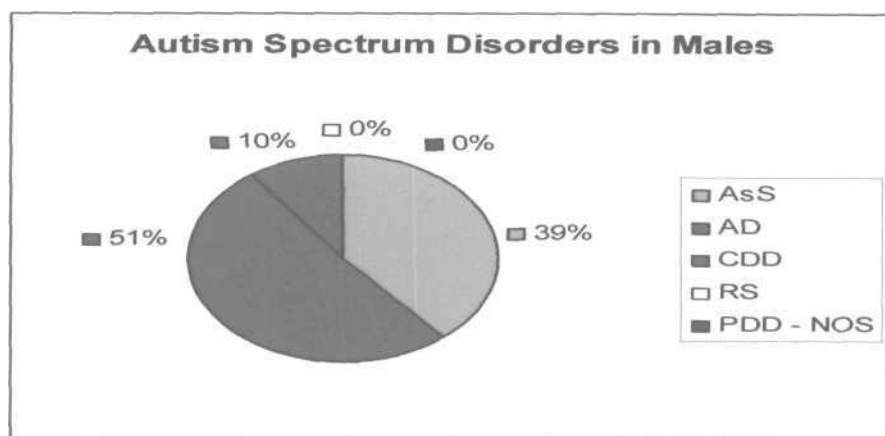
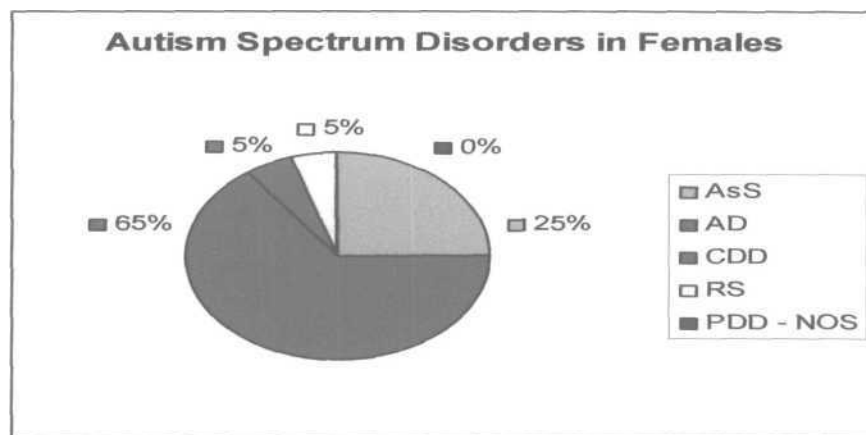


Table 3: Number and percentage of female subjects in different categories of Autism

Spectrum Disorders:

Categories of ASD	Number of Subjects (20)	Percentage
Asperger's Disorder	5	25%
Autistic Disorder	13	65%
Childhood Disintegrative Disorder	1	5%
Rett's Disorder	1	5%
Pervasive Developmental Disorders Not otherwise specified	0	0%

Pie - chart 3: Distribution of the different categories of Autistic Spectrum Disorders among the female subjects (20):



As seen in Table 1 and Pie chart 1 maximum number of children met the criteria of Autistic Disorder (54%) followed by Asperger's Disorder (36%), only few children fell under the category of CDD (9%) and among 100 subjects only 1 female subject was

diagnosed as having Rett's Disorder (1%). None of the children met the criteria to be diagnosed as having PDD - NOS, because all the children met the criteria for one of the formally defined disorders in that class of ASD.

Studies similar to this study have reported the epidemiology of ASD. Hoch, in 1967, found 2.9% cases were of infantile autism.

Srinath et al, (1989) conducted a retrospective review of cases registered from 1981 to 1984 in the child psychiatry clinic, NIMHANS, Bangalore, and reported that a total of 31 cases met the ICD-9 criteria for Autism. Out of this only 17 cases fulfilled the criteria laid down by DSM III.

Singhi and Malhi (2001) reported that 16 children <5 years of age met the DSM IV criteria for Autism.

(Malhotra & Gupta, 2002) found 12 cases of Childhood Disintegrative Disorder which is about 0.45% of all the cases and two female cases of Rett's Disorder in the period between 1989 and 1999 at the CAP (Child and Adolescent Psychiatry Clinic), PGIMER, Chandigarh.

Malhotra et al (2003) reported that of 2942 cases 46 cases (1.6%) met ICD-10 criteria for different PDDs. Among these 46 cases, 22 cases were of typical autism, 12 cases each were of CDD and other PDDs. 5 cases met criteria for atypical autism, 4 were

of AS and 2 were of Rett's Disorder and 1 was PDD unspecified (n=1). Relatively high proportion (26%) of cases of CDD is notable as it has been suggested that CDD is only about one-tenth as common as autism.

In the present study during classification of children into different categories of the ASD, crucial features helped in easy categorization of disorders.

The crucial features drawn for each disorder are listed below:

a) Asperger's Disorder - 2 crucial features

- 1) There is no clinically significant general delay in language (e.g., single words used by age 2 years, communicative phrases used by age 3 years).
- 2) There is no clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood.

b) Autistic Disorder -1 crucial feature

- 1) 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) Childhood Disintegrative Disorder - 2 crucial features.

- 1) Apparently normal development for at least the first 2 years after birth as manifested by the presence of age-appropriate verbal and nonverbal communication, social relationships, play, and adaptive behavior.
- 2) Clinically significant loss of previously acquired skills (before age 10 years) in at least two of the following areas:
 - expressive or receptive language
 - social skills or adaptive behavior
 - bowel or bladder control
 - play
 - motor skills

d) Rett's Disorder - 8 crucial features

- 1) Apparently normal prenatal and perinatal development
- 2) Apparently normal psychomotor development through the first 5 months after birth
- 3) Normal head circumference at birth
- 4) Deceleration of head growth between ages 5 and 48 months

- 5) Loss of previously acquired purposeful hand skills between ages 5 and 30 months with the subsequent development of stereotyped hand movements (i.e., hand-wringing or hand washing)
- 6) Loss of social engagement early in the course (although often social interaction develops later)
- 7) Appearance of poorly coordinated gait or trunk movements
- 8) Severely impaired expressive and receptive language development with severe psychomotor retardation.

All the above mentioned crucial features are mainly based on the age of onset of the behavior and the normal or deviant development of social skills, language skills and cognitive skills.

Children diagnosed as having Autistic Disorder had onset of the disorder prior to 3yrs of age with delay in development of social, language and cognitive skills. Whereas children with Asperger's Disorder had normal development of language, social and cognitive skills till 3yrs of age. Rett's Disorder and CDD are characterized by regression in language and motor skills. Children with Rett's Disorder lose their language and motor skills from the age of 5 months, whereas children with CDD would have normal development till 2 years, and would lose previously acquired skills anytime before 1 Oyr of age.

STEP 2:

In the DSM-IV criteria there were totally 14 overlapping features; these features were overlapping among Asperger's Disorder, Autistic Disorder and CDD. Among the 5 categories of ASD, Rett's Disorder stands out as it does not have any overlapping features and is characterized only by 8 crucial features.

The 14 overlapping features are listed below:

- 1) Marked impairment in the use of multiple nonverbal behaviors, such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
- 2) Failure to develop peer relationships appropriate to developmental level
- 3) A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
- 4) Lack of social or emotional reciprocity
- 5) 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)
- 6) In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
- 7) Stereotyped and repetitive use of language or idiosyncratic language

- 8) Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
- 9) Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
- 10) Apparently inflexible adherence to specific, nonfunctional routines or rituals
- 11) Stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting or complex whole-body movements)
- 12) Persistent preoccupation with parts of objects
- 13) The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.
- 14) The disturbance is not better accounted for by Rett's disorder or childhood disintegrative disorder.

The presence of each of these 14 criteria in a number of children in each group (Asperger's Disorder, Autistic Disorder and CDD) was found. This value was subjected to statistical analysis (Chi — square) to determine if these three categories of ASD can be differentiated into 3 distinct groups based on the 14 overlapping features in DSM - IV criteria. Table 4 shows the observed frequencies of overlapping features for the three categories of ASD. Table 5 shows the expected frequencies of overlapping features for the three categories of ASD.

Table 4: Observed frequencies of the overlapping criteria in different categories of Autism Spectrum Disorders.

Overlapping criteria	Asperger's Disorder	Autistic Disorders	Childhood Disintegrative Disorder	Total
1	36	54	9	99
2	35	54	9	98
3	36	53	-	89
4	33	48	8	89
5	-	52	9	61
6	-	8	2	10
7	-	46	9	55
8	-	52	9	61
9	35	53	9	97
10	13	23	-	36
11	36	53	9	98
12	17	40	-	57
13	36	54	9	99
14	36	54	9	99
Total	313	644	91	1048

Table 5: Expected frequencies of the overlapping criteria in different groups of Autism Spectrum Disorders.

Overlapping criteria	Asperger's Disorder	Autistic Disorders	Childhood Disintegrative Disorder
1	29.56	60.83	8.59
2	29.2	60.22	8.50
3	26.58	54.69	7.72
4	26.58	54.69	7.72
5	18.21	37.48	5.29
6	2.98	6.14	0.86
7	16.42	33.79	4.77
8	18.21	37.48	5.29
9	28.97	59.60	8.42
10	10.75	22.12	3.12
11	29.26	60.22	8.50
12	17.02	35.02	4.94
13	29.56	60.83	8.59
14	29.56	60.83	8.59

Chi - Square calculated value:

$$X^2 = 118.15$$

Chi - Square table value:

$$X^2_{26,0.05} = 38.39$$

Conclusion:

As the chi - square calculated value is greater than the Chi - square table values, the Null hypothesis - 1 [There are no significant differences between the three categories (Asperger's Disorder, Autistic Disorder and CDD) of ASD, based on the 14 overlapping features] is rejected.

So there seems to be significant difference between the three categories (Asperger's Disorder, Autistic Disorder and Childhood Disintegrative Disorder) of ASD based on the 14 overlapping features.

Among the 14 criteria, certain criteria were present in very less number of children compared to other criteria. Criteria number 6 (*In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others*) and criteria 11 (*Apparently inflexible adherence to specific, nonfunctional routines or rituals*) were found to be present only in 10% and 36% of individuals respectively whereas other criteria were present in more than 50% of individuals.

Similar to above procedure presence of each criterion in number of males and females was determined and subjected to statistical analysis (Chi - square) to find if there exists any gender difference based on the 14 overlapping features. Table 6 shows observed frequencies of the overlapping features of male and female subjects. Table 7 shows the expected frequencies of overlapping features of male and female subjects.

Table 6: Observed frequencies of the overlapping criteria in male and female subjects

Overlapping criteria	Males	Females	Total
1	80	19	99
2	78	19	97
3	71	18	89
4	72	17	89
5	47	15	62
6	8	2	10
7	42	13	55
8	46	15	61
9	78	19	97
10	31	5	36
11	79	19	98
12	46	11	57
13	80	19	99
14	80	19	99
Total	838	210	1048

Table 7: Expected frequencies of the overlapping criteria in male and female subjects

Overlapping criteria	Males	Females
1	79.16	19.83
2	77.56	19.43
3	71.16	17.83
4	71.16	17.83
5	49.57	12.42
6	7.996	2.003
7	43.97	11.02
8	48.77	12.22
9	77.56	19.43
10	28.78	7.21
11	78.36	19.63
12	45.57	11.42
13	79.16	19.83
14	79.16	19.83

Chi - Square calculated value:

$$\chi^2 = 2.997$$

Chi - Square table value:

$$\chi^2_{26,0.05} = 38.39$$

Conclusion:

As the chi - square calculated value is lesser than the Chi - square table value the Null hypothesis - 2 [There are no significant differences between the three categories (Asperger's Disorder, Autistic Disorder and CDD) of ASD in terms of gender based on the 14 overlapping features] is accepted.

So there is no significant difference between three categories (Asperger's Disorder, Autistic Disorder and Childhood Disintegrative Disorder) of ASD in terms gender, based on the 14 overlapping features

Based on the results in step - 1 and step - 2, it can be inferred that, by focusing more on the crucial features and the overlapping features (which occurred in more than 50% of children) in DSM - IV, it is possible to easily differentially diagnose children with ASD. As the DSM - IV criteria does not have gender differences on disorders, the same criteria can be applied to assess both male and female individuals with ASD. So to conclude the **Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM - IV), 1994** can be considered as a validate tool to differentially diagnose the **variety of Autism Spectrum Disorders in India.**

SUMMARY AND CONCLUSION

SUMMARY AND CONCLUSION

There have been enormous research studies in the area of ASD over the past 80 years, but still this class of disorders remains as a mystery to all speech language pathologists. There have been lots of misconceptions and controversies especially on the differential diagnosis of ASD among various groups of experts, considering the two schools of thoughts: one being that of precise, specific diagnostic labels, and the other adopting a more diffuse view of Autism as a spectrum of disorders. This study considers the latter view.

Considering Autism as a spectrum of disorders puts forth the important issue of differential diagnosis of ASD. So far the category of PDD was included in two classification systems namely, The *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV)*, published in 1994, and the *International Statistical Classification of Disease 10 th revision (ICD 10)*, 1992.

This study is an attempt to validate the *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV), 1994* in Mysore context (as clients come from all parts of country) to contribute to the existing literature of differential diagnosis of ASD on the Indian front. This study can also be considered as retrospective epidemiological study as it reports the number of children in each category of ASD.

The validation of DSM - IV criteria was done in 4 phases. It started with administration of DSM - IV questionnaire on 100 subjects (male to female ratio being 1:4) diagnosed as having autistic features by parental or caregiver interview along with information from case files. The children were then grouped into different categories of disorders based on the crucial and overlapping criteria. There were totally 14 features overlapping among Asperger's Disorder, Autistic Disorder and CDD (There are no overlapping features for Rett's Disorder; it has only 8 crucial features). The number of overlapping criteria for each child was determined, this value was subjected to statistical analysis (Chi - Square) based on which it was concluded that the three disorders (AS, Autistic Disorder and CDD) among ASD can be easily differentiated based on the 14 overlapping criteria in the DSM - IV. Similarly the number of overlapping criteria in the males and females subjects was determined and subjected to statistical analysis (Chi - Square), based on which it was determined that there exist no gender difference among the three disorders (Asperger's Disorder, Autistic Disorder and CDD) based on the 14 overlapping criteria.

Based on the results it was concluded that, by focusing more on crucial features and overlapping features which occurred in more than 50% of children, DSM - IV criteria can be considered as a validate clinical tool to differentially diagnose the five categories (Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, and Pervasive Developmental Disorder - Not Otherwise Specified) of Autism Spectrum Disorders in Indian context.

IMPLICATIONS OF THE STUDY

- This study would validate *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV), 1994* criteria in Indian context.
- Validation of DSM - IV criteria would help us to understand core features and characteristics of ASD, which will further help us to accurately subcategorize children with ASD and plan for intervention.

SUGGESTIONS FOR FUTURE RESEARCH

- The study would have given more interesting results, if children already diagnosed as having Asperger's Disorder, Autistic disorder and CDD, Rett's Disorder and PDD - NOS were taken and compared against DSM - IV criteria.
- Study of similar type can be done, to determine the epidemiology of ASD by randomly selecting cases from general and clinical population, during which various epidemiological features like language, sociocultural, economic and regional background can be studied.
- Co morbidity of features can be studied by taking subjects with ASD s and associated conditions.
- DSM-IV can be compared with ICD - 10 and other non - DSM IV tests, if any, used by different professionals.

- The study needs to be confirmed by larger group of sample with subjects from all over India.

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APPENDICES

APPENDIX -1

DSM - IV questionnaire for differential diagnosis of PDD/ASD

Name: _____ Case Number: _____ Age/Gender: _____
Diagnosis: _____ Therapy Duration: _____

Questions	Response	
	Yes	No
I. MEDICAL HISTORY		
1. Did your child have apparently normal Prenatal and perinatal development?	•	•
2. Did your child have normal head circumference at birth?	•	•
3. Did your child have apparently normal psychomotor development through the first 5 months after birth?	•	•
4. Does your child appear to have poorly coordinated gait or trunk movements?	•	•
5. Did he lose previously acquired purposeful hand skills between ages 5 and 30 months with the subsequent development of stereotyped hand movements (i.e., hand-wringing or hand washing)?	•	•
6. Did the child have deceleration of head growth between ages 5 and 48 months?	•	•

II. LANGUAGE SKILLS

7. Was there a clinically significant general delay in language {e.g., single words used by age 2 years, communicative phrases (2 word sentence) used by age 3 years }? • •
8. Was there any 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)? • •
9. Does your child have 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? • •
10. Does your child have marked impairment in the ability to initiate or sustain a conversation with others inspite of having adequate language ability? • •
11. Does your child exhibit stereotyped and repetitive use of language or idiosyncratic language? • •
12. Does your child have severely impaired expressive and receptive language development with severe psychomotor retardation? • •
13. Did your child have apparently normal development for at least the first 2 years after birth as manifested by the presence of age-appropriate verbal and nonverbal communication, social relationships, play, and adaptive behavior? • •

III. SOCIAL SKILLS

14. Does your child show social or emotional reciprocity? • •
15. Does your child fail to develop peer relationships appropriate to developmental level? • •
16. Does your child spontaneously seek to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)? • •
17. Did he lose of social engagement early in the course (although often social interaction develops later)? • •
18. Does your child lack varied, spontaneous make-believe play or social imitative play appropriate to developmental level? • •

IV. BEHAVIORAL SKILLS

19. Does your child encompass preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus? • •
20. Does your child exhibit apparently inflexible adherence to specific, nonfunctional routines or rituals? • •
21. Does your child exhibit stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting or complex whole-body movements)? • •
22. Does your child show persistent preoccupation with parts of objects? • •

V. OTHERS

23. Does he show any 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. • •
24. Does the disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning? • •
25. Is there clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood? • •
26. Did he exhibit clinically significant loss of previously acquired skills (before age 10 years) in at least two of the following areas? • •
- a) Expressive or receptive language
 - b) Social skills or adaptive behavior
 - c) Bowel or bladder control
 - d) Play
 - e) Motor skills

APPENDIX - II

DSM-IV (APA, 1994) classification of Pervasive Developmental Disorders (PDD)

299.00 Autistic Disorder

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

(1) qualitative impairment in social interaction, as manifested by at least two of the following:

(a) marked impairment in the use of multiple nonverbal behaviors, such as eye-to- eye gaze, facial expression, body postures, and gestures to regulate social interaction

(b) failure to develop peer relationships appropriate to developmental level

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

(d) lack of social or emotional reciprocity

(2) qualitative impairments in communication, as manifested by at least one of the following:

(a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)

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

(c) stereotyped and repetitive use of language or idiosyncratic language

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

(3) restricted, repetitive, and stereotyped patterns of behavior, interests, and activities as manifested by at least one of the following:

(a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus

(b) apparently inflexible adherence to specific, nonfunctional routines or rituals

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

(d) persistent preoccupation with parts of objects

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

C. The disturbance is not better accounted for by Rett's disorder or childhood disintegrative disorder.

299.80 Pervasive Developmental Disorder, Not Otherwise Specified

This category should be used when there is a severe and pervasive impairment in the development of reciprocal social interaction or verbal and nonverbal communication skills, or when stereotyped behavior, interests, and activities are present, but the criteria are not met for a specific pervasive developmental disorder, schizophrenia, schizotypal personality disorder, or avoidant personality disorder. For example, this category includes "atypical autism" —presentations that do not meet the criteria for autistic disorder because of late age of onset, atypical symptomatology, or subthreshold symptomatology, or all of these.

299.80 Asperger's Disorder

A. Qualitative impairment in social interaction, as manifested by at least two of the following:

(1) marked impairment in the use of multiple nonverbal behaviors, such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction

(2) failure to develop peer relationships appropriate to developmental level

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

(4) lack of social or emotional reciprocity

B. Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:

(1) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus

(2) apparently inflexible adherence to specific, nonfunctional routines or rituals

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

(4) persistent preoccupation with parts of objects

C. The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.

D. There is no clinically significant general delay in language (e.g., single words used by age 2 years, communicative phrases used by age 3 years).

E. There is no clinically significant delay in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood.

F. Criteria are not met for another specific pervasive developmental disorder or schizophrenia.

299.80 Rett's Disorder

A. All of the following:

- (1) apparently normal prenatal and perinatal development
- (2) apparently normal psychomotor development through the first 5 months after birth
- (3) normal head circumference at birth

B. Onset of all of the following after the period of normal development:

- (1) deceleration of head growth between ages 5 and 48 months
- (2) loss of previously acquired purposeful hand skills between ages 5 and 30 months with the subsequent development of stereotyped hand movements (i.e., hand-wringing or hand washing)
- (3) loss of social engagement early in the course (although often social interaction develops later)
- (4) appearance of poorly coordinated gait or trunk movements
- (5) severely impaired expressive and receptive language development with severe psychomotor retardation

299.10 Childhood Disintegrative Disorder

A. Apparently normal development for at least the first 2 years after birth as manifested by the presence of age-appropriate verbal and nonverbal communication, social relationships, play, and adaptive behavior.

B. Clinically significant loss of previously acquired skills (before age 10 years) in at least two of the following areas:

(1) expressive or receptive language

(2) social skills or adaptive behavior

(3) bowel or bladder control

(4) play

(5) motor skills

C. Abnormalities of functioning in at least two of the following areas:

(1) qualitative impairment in social interaction (e.g., impairment in nonverbal behaviors, failure to develop peer relationships, lack of social or emotional reciprocity)

(2) qualitative impairments in communication (e.g., delay or lack of spoken language, inability to initiate or sustain a conversation, stereotyped and repetitive use of language, lack of varied make-believe play)

(3) restricted, repetitive, and stereotyped patterns of behavior, interests, and activities, including motor stereotypies and mannerisms

D. The disturbance is not better accounted for by another specific pervasive developmental disorder or by schizophrenia.