

**Review Article**

**AUTISM: ASSESSMENT AND MANAGEMENT**

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**SUMMARY**

Autism and the related pervasive developmental disorders are characterized by patterns of delay and deviance in the development of social, communicative and cognitive skills, which arise in the first years in life. Differential diagnosis includes other psychiatric and pervasive developmental disorders, deafness and profound hearing loss. Autism is frequently associated with fragile X syndrome and tuberous sclerosis. Common co-morbidities include Mental Retardation, Seizure Disorder and Psychiatric disorders such as depression and anxiety. Early detection and intervention significantly improve outcomes, with about one third of autistic persons achieving some degree of independent living. Various treatment modalities administered by Multidisciplinary team are helpful. (e.g. Pharmacotherapy, special education, speech, communication therapy, and behavior modification). A Medline search dating back to 1970 was done, to look at the literature related to autism. Based on this search, up to date review of autism has been discussed with special emphasis on assessment and management.

**KEY WORDS:** Autism, Pervasive Developmental disorder, Developmental Disorder, Child Psychiatry, Children.

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**EPIDEMIOLOGY**

Autism is a neurodevelopmental disorder first described in a small case series by Leo Kanner in 1940's.¹ A series of epidemiological studies have been conducted and generally suggest rates of autism between 1 in 500 and 1 in 1000 children; children with difficulties falling within the broader PDD category are probably three or four times as common.² The frequency of the disorder appears to be changing, with recent survey of Center for Disease Control (CDC) indicating prevalence of 1 in 166 children in USA.³ Rates of autism in boys are three to four times higher than in girls but when females have autism; they tend to be more severely affected.²⁴

In most epidemiological based samples of autism, approximately 50% of cases exhibit severe or profound mental retardation, 30% have mild to moderate mental retardation and the remaining 20% of cases have IQ in the normal range.⁵

**ASSESSMENT AND DIAGNOSTIC METHODS**

Diagnosis is based on the identification of a particular pattern of deficits, together with consideration of whether this might be fully attributable to cognitive impairment or deafness etc. DSM- IV-TR Diagnostic Criteria for Autistic Disorder (Table I) and ICD-10 Crite-
ria for Childhood Autism (Table II) are helpful for making diagnosis of autism.\(^6\)^7

Indicators for formal developmental evaluation include no babbling, pointing or other gestures by 12 months of age, no single words by 16 months of age, no two words spontaneous phrases by 24 months of age and loss of previously learned language or social skills at any age.\(^8\) Parental concerns about delayed speech and language development typically noticed at about 18 months of age should always be taken seriously.

Making a certain diagnosis usually involves a complex multidisciplinary assessment of development, cognitive functioning, and speech and language skills. Special educational needs require assessment by an educational psychologist and observation of the child’s behavior and skills in several settings is necessary.

Historical information:

Clinical presentation: The diagnosis of Autism requires deficit in three areas including social interactions, communication, and restricted, repetitive behaviors and interests and also requires recognition of some type of abnormality before 36 months of age. There is variability in the age at which children present with features essential for the diagnosis.\(^9\) Predictors of ultimate outcome include the presence of communicative speech by age 5 and overall cognitive ability (IQ).\(^10\)

Social deficits: Areas of social disturbances include marked impairment in nonverbal behaviors, in social interactions, failure to develop peer relations as appropriate for developmental level, lack of seeking to share enjoyment or interest and a lack of social and emotional reciprocity.\(^11\) Children with autism are less likely to comfort others (such as bringing baby sister a blanket)

Communication: Fifty percent of all individuals with autism never speak at all, and those who do speak use awkward and often monotonous speech.\(^12\) Typically development of speech is delayed in children with autism, with most children having no single words until after age two and no phrase speech after age three. Pronoun reversal is common (e.g. referring to themselves as ‘you’ or ‘he’) and speech may be characterized by echolalia or delayed echolalia, in which previously heard words or phrases are repeated at a later time.\(^12\) The social cues that guide conversation are poorly understood by children with autism, so that speech may be too loud or too soft, rapid, halting or devoid of inflection.\(^13\)

Autism is associated not only with difficulties in language but also with deficits in nonverbal communication, including use of gestures, such as pointing, nodding, and showing. Deficits also occur in imitation, social play, spontaneous imaginative role play, or play with dolls or action figures.\(^14,15\) Autism is most often associated with lower verbal than non-verbal IQ score.\(^16\)

Restricted, Repetitive Interests and Behaviors: This category includes encompassing preoccupations and interests; adherence to nonfunctional routines or rituals, stereotypies and motor mannerisms (e.g. hand or finger flapping, body rocking, moving in circles, walking on toes); and persistent preoccupation with parts of objects (AACAP Parameters.1999).\(^17\) These children may repetitively move or spin an object in front of their eyes, or examine it from different angles for long periods of time. These repetitive behaviors provide sensory self-stimulation.\(^18\)

Other Features: Autistic children are often overactive with a short attention span. Other associated features include aggression, self-injurious behaviors, and sleep difficulties. Autistic children have high rates of anxiety. Seventy percent of autistic children have mental retardation, with only 5% having an IQ above 100. Occasionally some have remarkable abilities in isolated areas for instance music, computation or remote memory. About 20 % develop epilepsy most likely during adolescent years.\(^19\)

Pregnancy, neonatal and developmental history:

This includes a review of pregnancy, labour and delivery and of early neonatal course. The developmental history should focus on early development of language, social behavior and motor skills. The clinician should establish whether the child met motor milestones on time, used single words before age two years,
and put words together in short phrases by age three. A typically developing baby will make eye contact with adults and children and will begin to babble between ages 4 and 6 months. By two years of age typical children use about 50 single words, although there will be some slight variability.\textsuperscript{20}

Medical history: It should include history of seizures, head injury, hearing or visual impairment, or any other relevant medical conditions like fragile X syndrome.

Family history: It should be reviewed for presence of autism, other developmental disorders, mental retardation, speech delays, learning disabilities and epilepsy.

Psychosocial history: Family situation, stressors and support systems should be identified during the assessment.

Intervention history: The current and past psychotropic medication should be reviewed along with child response to any educational program or behavioral interventions.

**Diagnostic Instruments:** Various rating scales can be useful for gathering information.

Helpful tools include:

* The Autism Behavior Checklist.\textsuperscript{21}
* The Childhood Autism Rating Scales.\textsuperscript{22}
* The Autism Diagnostic Interview.\textsuperscript{23,24}
* Vineland Adaptive Behavior scale\textsuperscript{25} and the Aberrant behavior checklist\textsuperscript{26} while not targeted towards PDD but identify developmental delay and frequent maladaptive behavior seen in children with developmental difficulties.
* Autism Diagnostic Observation Schedule

**Autism Behaviour Checklist:** (ABC) It is a valid and reliable parent rating scale consisting of 57 items that target autistic type behaviors in five domains: sensory, relating, body and ob-

**Table-I: DSM- IV-TR Diagnostic Criteria for Autistic Disorder**

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

Table II: ICD-10 Criteria for Childhood Autism

Childhood Autism
A. Abnormal or impaired development is evident before the age of 3 years in at least one of the following areas:
   1. receptive or expressive language as used in social communication;
   2. the development of selective social attachments or of reciprocal social interaction;
   3. functional or symbolic play;
B. A total of at least six symptoms from (1), (2) and (3) must be present, with at least two from (1) and at least one from each (2) and (3):
   (1) Qualitative abnormalities in reciprocal social interaction are manifest in at least two of the following areas:
      a. failure adequately to use eye-to-eye gaze, facial expression, body posture, and gesture to regulate social interaction;
      b. failure to develop (in a manner appropriate to mental age, and despite ample opportunities) peer relationships that involve a mutual sharing of interests, activities, and emotions;
      c. lack of socioemotional reciprocity as shown by an impaired or deviant response to other people’s emotions; or lack of modulation of behavior according to social context; or a weak integration of social, emotional, and communicative behaviors;
      d. lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. a lack of showing, bringing or pointing out to other people objects of interest to the individual).
   (2) Qualitative abnormalities in communication are manifest in at least one of the following areas:
      a. a delay in, or total lack of, development of spoken language that is not accompanied by an attempt to compensate through the use of gesture or mime as an alternative mode of communication (often preceded by a lack of communicative babbling);
      b. relative failure to initiate or sustain conversational interchange (at whatever level of language skills is present, in which there is reciprocal responsiveness to the communications of the other person;
      c. stereotyped and repetitive use of language or idiosyncratic use of words or phrases;
      d. lack of varied spontaneous make-believe or (when young) social imitative play.
   (3) Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities are manifest in at least one of the following areas:
      a. an encompassing preoccupation with one or more stereotyped and restricted patterns of interest that are abnormal in content or focus; or one or more interests that are abnormal in their intensity and circumscribed nature though not in their content or focus;
      b. apparently compulsive adherence to specific, nonfunctional routines or rituals;
      c. stereotyped and repetitive motor mannerisms that involve either hand or finger flapping or twisting, or complex whole body movements;
      d. Preoccupations with part-objects or nonfunctional elements of play materials (such as their odor, the feel of their surface, or the noise or vibration that they generate).
C. The clinical picture is not attributable to the other varieties of pervasive development disorder: specific developmental disorder of receptive language with secondary socioemotional problems; reactive attachment disorder or disinhibited attachment disorder, mental retardation with some associated emotional or Behavioral disorder; schizophrenia of unusually early onset; and Rett’s syndrome.


ject use, language and social/self help. Examples of items from the ABC include, whether the child twirls and spins repeatedly, whether the child seems not to hear when spoken to, or whether the child does not imitate other children at play. Item scores are weighted and summed. The total score indicates whether the child shows serious autistic symptomatology.
The childhood autism rating scale: (CARS)
It is one of the most widely used rating scale for behaviours associated with autism. The child is rated by the clinician on 15 behaviours during the observation period of the assessment. The items are totaled to yield a score that indicates whether a child’s behavior is suggestive of PDD.

The autism diagnostic interview-revised (ADI-R)
It is a semi structured, clinician administered interview. The interview incorporates both developmental history and parent report regarding current behavior. The ADI-R consists of 111 items divided into four domains: communication, social reciprocity, restricted and repetitive behaviors and imagination. Domain scores are summed and compared against established cutoffs scores to determine whether the individual meets the criteria for autism in each domain.

The vineland adaptive behavior scale: (VABS)
It is a valid, standardized and reliable instrument used to collect information about day to day functioning in four domains: communication, daily living, socialization and motor skills. The data is collected in an interview format, in which the parent is asked to provide examples of day to day living with the child. The standard scores obtained reflect how the child functions in each domain compared to same age peers.

Autism diagnostic observation schedule: ADOS is a structured play session in which the examiner carries out a series of play or social activities to assess social interactions. The ADOS consists of four modules, each intended for children or adults with different levels of expressive language. The tasks within each module of ADOS, both play and conversation contain a series of scripted interactions, in which examiner leads with either a verbal or nonverbal cue. The responses are then scored and autism is diagnosed by using an algorithm.

The aberrant behavior checklist: It is a 57 item rating scale that can be completed by parents, teachers, or professional caretakers. It provides information regarding a broad range of behaviors that are relevant to developmental disabilities.

Observation/Interview of the child: The first component in assessment is one or several observation periods and play sessions with the child. Children with PDD are less likely to relate to a clinician or they relate in an unusual way. They actively avoid eye contact or bids for interactive play. Initiating play with a child is also an excellent way to assess social relatedness. Interactive games, simple make believe and more sophisticated pretend play can be used to observe the child’s ability to enjoy interaction, use language and think symbolically.

Medical Assessment: A medical history and physical examination should be obtained. Routine laboratory studies including Lead levels due to high levels of Pica in these children should be done. Any history of relevant medical condition would guide further laboratory studies if needed. EEG may be relevant in history suggestive of Seizures.

Audio logical and visual examination as well as neurological assessment may be helpful.

Speech/Language/Communication Assessments: This should cover vocabulary, actual use of language (receptive and expressive), articulation and oral motor skills, and social use of language and communication skills.

Occupational and Physical therapy Assessment: Assessment is helpful especially if there is some degree of hyposensitivity or hypersensitivity or difficulties in motor development.

DIFFERENTIAL DIAGNOSIS
It includes consideration of the various Pervasive Developmental Disorders (PDD), Mental Retardation not associated with PDD, specific Developmental Disorders (e.g. language disorders), Early Onset Psychosis, Deafness, OCD and Reactive Attachment Disorder.

Mental retardation with behavioural symptoms: Mentally retarded children usually relate to adults and other children in accordance with their mental age; they use the language to communicate with others, and they have a relatively even profile of impairments.
Schizophrenia with childhood onset: It is rare under the age of 5. There is low incidence of seizures and mental retardation and children have hallucinations and delusions.

Congenital deafness or severe hearing impairments: Deaf children only respond to loud sounds, whereas autistic children may ignore loud and normal sounds and respond to soft or low sounds. Children with deafness also usually relate to their parents and seek their affection.

Language related developmental disorders: Primary deficits are in the area of language/communication: social skills are usually preserved and restricted and repetitive behaviors of autism are not present.

Obsessive-compulsive disorder: Social and communication skills are well preserved in children and adolescents with OCD.

Reactive attachment disorder: There usually is history of severe neglect and social deficits remit in caring and suitable environment.

Co-morbid medical conditions: The prevalence of various co-morbid medical and neurodevelopmental conditions in autism is around 10%.28 Fragile X Syndrome is the most common form of mental retardation affecting about 1 in 4,000 males and 1 in 8,000 females.29 Approximately 15-25% of the cases with Fragile X syndrome also have autistic features.30, 31 Tuberous Sclerosis is an autosomal dominant disorder characterized by the classic triad of mental retardation, epilepsy and skin lesions.32 The prevalence of tuberous sclerosis in autism spectrum disorder estimated to be about 1-4%.33

The incidence of Down syndrome in children with autism is estimated to be as high as 11%.34,35 The most common cause of Down syndrome is trisomy21. Autistic patients have more than a 100 fold increased risk of developing neurofibromatosis 1 (NF1) as compared to the general population.36,3 Neurofibromatosis is characterized by multiple cafe-au-lait spots and neurofibromas. About 50% of people with NF1 also have learning disabilities. There are multiple reports linking the neurofibromatosis 1 (NF1) gene and autism.38

The features of autism including social deficits, stereotypies and narrow range of interests are strikingly common in some cases of congenital blindness.39,40 There are also high rates of autistic features in children who have deafness.41,42 For the physicians serving patients with autism, a referral for genetic testing should be considered when a patient with autism has dysmorphic features or unusual physical and developmental growth which is not typical for autism.

TREATMENT

The comprehensive treatment of autism include speech and language therapy, social skills training, appropriate school and educational services, structured environments, occupational therapy and behavioral interventions along with medication evaluation, and treatment and education about the illness and support to the parents. Early intervention is extremely important. All the interventions should be carefully planned with interdisciplinary and integrated approach. Involvement of the child and family is the key to success.

Speech/language therapy: Since presence of communicative speech by age 5 is the most important predictor of positive outcome, it’s important to have speech and language assessment and interventions as soon as possible. Some of the behavioral strategies to increase communication include teaching sign language or using picture representations.45

Social skills training: Social skills deficits are hallmark of autistic disorder. Various interventions such as Social Stories and Priming and Pivotal Response Training can be effective in this respect.44,45 Play can be important in improving peer interactions.

Educational Interventions: Appropriate educational setting is extremely important for appropriate learning. Educational curriculum has to be individualized and flexible according to the needs of the children. Usually small and structured educational environment with emphasis on child’s strength is beneficial.

Occupational Therapy: Most of the children with autism have sensory issues, like being sensitive
to touch, bright lights, certain sounds, particular clothing and foods. These children should have individualized occupational therapy plan including sensory integration approaches.\textsuperscript{46} 

**Behavioral Approaches:** Behavioral interventions should be one of the first interventions applied to reduce disruptive behaviors like aggression, temper tantrums and self injurious behaviors, and to promote skills that support normal development. Behavioral approaches for teaching daily life skills like toilet training, feeding, taking bath, brushing teeth are important aspects of day to day living which are important in improving quality of life. Various behavioral models are used with most popular being Applied Behavioral Analysis (ABA) and Lovaas model.\textsuperscript{47} 

**Family Involvement:** Family involvement at every stage of the process is key to successful treatment. Support groups for parents and siblings can make a huge difference for the families. Along with professionals, Internet and educational materials can be an important resource for providing basic information. 

**Psychopharmacology:** Various psychotropic medications are used to treat problems associated with autism spectrum disorders. These medications generally are not helpful for the core symptoms of social issues and communication difficulties but can be effective for associated symptoms including aggression, self injurious behaviors, hyperactivity, impulsivity, stereotypies and repetitive behaviours, and sleep problems. Various surveys show that the frequency with which psychotropic medications are used, range from 45\% to 55\%, with antidepressants, antipsychotics, stimulants and antiepileptic medications the most commonly prescribed medications.\textsuperscript{48-50} Since patients with autism and other developmental disabilities are sensitive to side effects, the various psychotropic medications should be started on a very low dose and titrated very slowly. 

**Stimulants:** Stimulant medications are prescribed in autism spectrum disorders to manage hyperactivity, impulsivity and attention span problems. Studies have shown positive response with stimulants for these symptoms, but response rate is much lower in autism spectrum disorders as compared to children with ADHD.\textsuperscript{51,52} The Research Units on Pediatric Psychopharmacology (RUPP) Autism Network completed a double-blind, placebo-controlled, crossover trial of children aged 5 to 14 years with methylphenidate. Only half of the group was much improved or very much improved. About one in five children could not tolerate the medication.\textsuperscript{53} 

**Selective Serotonin Re-Uptake Inhibitors (SSRI’S):** This class of medications is one of the most commonly used in this population. Serotonin re-uptake inhibitors are often considered to treat stereotypies and repetitive behaviors that interfere with day-to-day functioning. SSRI’s can be helpful for co morbid anxiety as well as depression. There are double blind studies involving various SSRI’S including fluoxetine, fluvoxamine and clomipramine (a tricyclic antidepressant with significant serotonin reuptake inhibition) showing positive response in decreasing repetitive behaviors.\textsuperscript{54-56} 

**Atypical Antipsychotics:** Atypical antipsychotics use is widespread in autism spectrum disorders. There are positive studies showing reduction in aggression, self-injurious behaviors, impulsivity, hyperactivity and repetitive behaviors. Patients on atypical antipsychotics should be carefully monitored for weight gain and metabolic abnormalities. 

Risperidal, an atypical neuroleptic is the first psychotropic medication approved for treatment of irritability associated with autism by the Food and Drug Administration (FDA) in USA. This FDA approval was based on landmark multisite, randomized, double-blind study conducted by Research Units on Pediatric Psychopharmacology (RUPP) Autism Network, involving 101 autistic children with reduction in temper tantrums, aggression and self-injurious behavior.\textsuperscript{57} There are also small studies involving olanzapine, aripiprazole and ziprasiodone with positive response.\textsuperscript{58-60} 

**Anticonvulsants:** Autism spectrum disorders has high rate of co-morbidity with seizures. Good seizure control is essential in these children and adolescents. Anticonvulsants like
divalprox sodium and carbamazepine have an added advantage of helping with mood lability and behavioral issues.  

**Sympatholytics:** The alpha adrenergic agonists, clonidine and guanfacine are used in autistic children to help with hyperactivity, impulsivity and irritability.  

**CONCLUSION**  

General Practitioners, Pediatricians, Psychiatrists, as well as staff from various disciplines working with children are likely to encounter children with Pervasive Developmental Disorders (PDD). To work effectively with these children and families, they must know how these clinical syndromes differ from other childhood psychiatric disorders. Knowledge of typical impairments seen in children with autism and how these impairments are manifested at different developmental levels is crucial. In addition, assessment of family perspective and coping skills guide effective intervention.

**REFERENCES**  


17. Practice Parameters for Assessment and Treatment of Children, Adolescents and Adults with Autism and other Pervasive Developmental Disorders. Am Academy Child Adolesc Psychiatry (supplement) 1999;38:325-54S.