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Research Article

Clinical profile of children with autism spectrum disorder in tertiary care centre

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ABSTRACT

Background: Autism spectrum disorders (ASDs) present with major impairments social relationships, abnormal language development and rigid patterns of behaviour. There is variable clinical presentation of each subgroup of autism spectrum. This study aim at describing and analyzing the clinical and developmental profile of children with autism.
Methods: This cross-section observational study was conducted at the neurodevelopmental clinic at a tertiary care hospital in Mumbai, India over a period of two years. Consecutive 112 children (<12 yrs) diagnosed as autism spectrum disorder (as per DSM-IV) were assessed in detailed about perinatal events, developmental milestones and behavioral issues. Complete physical, neurological examination and neurodevelopmental assessment was done. MRI and CT brain done wherever possible.
Results: The clinical spectrum in our study includes ,autism 82(73.20%) cases, PDD-NOS -20(18.8%), 3 cases CDD (2.7%) and only 2 children with Rett's disease (1.8%) .The male: female ratio for Autism was 3.82:1. Common developmental concern observed were language delay in 93 (83.03%), lack of reciprocal social interaction in 66 (80.49%), lack of pretend play in one third, head banging and tantrums in 27 (32.93%) of children. Stereotypical movements, rocking, spinning were observed in almost 50% of study group. Unusual sensory responsiveness to smell, touch, sound was observed in 15(18.8%) of autistic kids. No clinico-radiological correlation found in behavioral patterns of Autism.
Conclusions: Core symptoms seen in Autism were deficits in social functioning, impaired language/communication skills, abnormal play, and unusual sensory affection. Early recognition of symptoms would help in appropriate therapeutic intervention resulting in favourable outcome.

Keywords: Autism, Clinical profile, Autism spectrum, Behaviour

INTRODUCTION

Autistic spectrum disorders (ASD) are a group of neurodevelopmental disorders with impaired social, communicative and cognitive development. Autism is one of five developmental disorders included under the pervasive developmental disorders (PDD).[1] In addition to Autism, other disorders in this group are Asperger's syndrome, Rett's disorder, childhood disintegrative disorder (CDD), and when full criteria for one of the above disorders is not met, a child may be diagnosed with pervasive developmental disorder-not otherwise specified (PDD-NOS).[1,2] Rett was included in the PDD category in DSM-IV and ICD-10, but subsequently, a specific

genetic etiology has been determined, as a consequence, Rett's disorder is anticipated to be removed from the DSM-V.[3,4]

Autism is characterized by deficits in social interaction and communication, and unusual and repetitive behavior. Although autism is defined by a certain set of behaviors, children exhibit any combination of the behaviors in any degree of severity. Children with autistic spectrum disorders differ from one another in individual behavioral characteristics and cognitive abilities, in severity of the disorder and in the coexistence of other medical conditions as well as environmental influences. As with any child with special needs, early identification is essential to allow interventions to be implemented. With autism, this need to be started before deviation and delay from the normal pattern of development has progressed too far. Children with autism often display a characteristic need for sameness and structure. Their resistance to change can impede treatments because inappropriate behaviors have to be curtailed as well as more appropriate behaviors introduced.[5] Early interventions can improve communication skills and reduce out-of-control behaviors. It is generally felt that autism is less prevalent in India but our experience suggests that there is a lack of awareness about the condition among pediatricians and often these children are misdiagnosed or diagnosed late. Early diagnosis and prompt referral for multidisciplinary intervention is a need of time and that is possible with increased awareness regarding the core symptoms of Autism among practitioners and parents. Very little systematic data is available from India and other developing countries.[6,7] We conducted this study with the aim of describing and analyzing the clinical and developmental profile of Indian children with autism.

METHODS

This cross-section observational study was carried out at the neurodevelopmental clinic at a tertiary care hospital in Mumbai, in India over a period of two years. The study was initiated after obtaining approval from the institutional ethics committee and children were enrolled after obtaining informed consent from the parents or guardians.[8]

Out of the children referred to the neurodevelopmental clinic consecutive 112 children fulfilling inclusion and exclusion criteria were enrolled for the study after obtaining parental consent. Children included in study were below 12 years of age with a primary diagnosis of autism spectrum disorder and without recognizable neurological condition wherein autism could be an associated feature to obviate the confounding effects of the neurological disorder on the neurodevelopmental profile of these children. The diagnosis of autism was made independently by the two authors based on the criteria delineated by the DSM IV which assesses three broad behavioural domains: social interaction, communication, and activities and interests. In addition, the childhood autism rating scale (CARS) was administered to all children above 3 years of age and modified checklist for autism in toddlers (M CHAT) below 3 years of age.[9,10] A detailed history about behavioural issues, prenatal and perinatal events, and developmental milestones was taken. Complete physical and neurological examination and neurodevelopmental assessment was done in all the children. Hearing and visual defects, other neurological deficits and disorders were excluded. The functioning of the child is expressed in developmental ages in months. The Vineland social maturity scale (VSMS, Indian adaptation) is used to assess the child's development level in looking after his practical needs and taking responsibility in daily living and social quotient (SQ). Neuroimaging (MRI Brain-plain, T1W, T2W and FLAIR images) was advised and studied in detail.

All the data of our study cases was analyzed for statistical significance, using Pearson Chi square test (including the one-way classification of Chi-square test, with or without Yate's correction for continuity). The results were considered statistically significant if p value was less than 0.05 (Note- at some places, although p value was in the significant range but it could not be interpreted well due to small number of cases).

RESULTS

The study population consisted 112 children (24 girls and 88 boys) diagnosed as Autism spectrum giving a sex ratio of M: F=3.67:1. The male: female ratio for Autism was 3.82:1 and for PDD-NOS, it was 3:1. The mean age at presentation for girls was 5.86+/-2.81 year, whereas boys presented at mean age of 7.70+/-2.82 yr. Most of these children were referred to our center for certification and their age represents their age of referral rather than age at which diagnosis was made. In most-67 (59.8%) patients, the first symptoms were identified at the age group of 2 to 4 years. In only two children, symptoms were identified above 6 years (1.8%).

In our study maximum children had features of autism 82 (73.20%) followed by PDD-NOS-20 (18.8%). We had only two children in category of Rett's disease (1.8%) and three in CDD (2.7%). In addition, boys accounted for majority of cases of Autism 65 (79.27%), PDD-NOS 15 (75%) Asperger's five (100%) and CDD 3 (100%) (Table 1).

In our study out of 112 children, 26 had macrocephaly, (23.21%) of which 25 were male, (96.15%) and only one was female child. This association shows statistically significant male preponderance of macrocephaly in the study population. Both the girls with Rett's syndrome had microcephaly.

Out of 82 children with autism, CARS was applied in 77 as per the age at presentation criteria (above 3 years), 40

of these i.e., 51.95% were severely autistic; with the CARS score in the range of 37-50. However, only 2 children with PDD-NOS i.e., 10.52% had scores in severely autistic features. Three children (60.00%) with Asperger had moderate autistic features. One child with CDD (50.00%) and 2 with Rett's disease (100.00%) were severely autistic (Table 1).

Table 1: Demographical profile of study population.

Total cases enrolled	**112**
Male	88
Female	24
Male: female ratio	3.67: 1
Clinical classification of cases	
Classical Autism	82(73.2%)
PDD-NOS	20(18.8%)
Asperger's syndrome	5(4.46%)
Childhood disintegrative disorder	3(2.70%)
Rett's syndrome	2(1.88%)
Age of recognition of symptoms	
Before 24 months	32(28.57%)
25-48 months	67(59.82%)
After 48 months	13(11.64%)
Severity of symptoms	
Mild (CARS score 15-30)	22(19.64%)
Moderate (CARS score 31-36)	29(25.89%)
Severe (CARS score 37-50)	51(45.53%)
Abnormalities of head size	
Normocephaly	71(63.39%)
Microcephaly	15(13.39%)
Macrocephaly	26 (23.21%)

Regarding the behavioral characteristics, lack of eye contact was a predominant feature in Autism and was also seen in all cases of Asperger and CDD. Lack of reciprocal social interaction was seen in 66 (80.49%) of autistic patients, 12 (60.00%) of PDD-NOS and all children with Asperger, Rett's and CDD. Language delay, a predominant feature in the study group, was seen in 93 (83.03%) of children. Echolalia was commonly seen in Asperger-3 (60.00%) and least common in the PDD-NOS group. Regression of language was seen in both the girls with Rett's syndrome and all children of CDD. Stereotypical movements were seen in all patients of Rett's and in 44 (53.66%) of autistic patients. It was observed least commonly with Asperger (Table 2).

Lining of toys in obsessive manner was more commonly seen in the Asperger group-3 (60.00%) and none in Rett's. Lack of pretend play was seen in one third autistic children, all children with CDD and 20% of Asperger group. Solitary play and aloofness was a predominant feature of the Asperger, seen in 4 (80.00%) and least commonly seen with PDD-NOS-5 (25%). Head banging and tantrums were reported in 16 (19.51%) and 27 (32.93%) of children with autism respectively and none with CDD and Rett's disease. Unusual sensory responsiveness to smell, touch, sound was a feature seen in 15(18.8%) of autistic kids but seen more commonly in the PDD-NOS group-8 (40.00%) and none in the Asperger, CDD and Rett's disease. Lack of adaptation to change in surroundings was a predominant feature seen in autism and the PDD-NOS group-10 (50.00%) (Table 2).

Table 2: Behavioral characteristic and type of ASD.

Behavioural characteristic	**Clinical types of ASD**					**p value**	**Remark**
	Classical autism	**PDD-NOS**	**Asperger syndrome**	**Childhood disintegrative disorder**	**Rett's syndrome**		
Lack of eye contact (n=85)	63 (76.83%)	13 (65%)	5 (100%)	3 (100%)	2 (100%)	0.330	Not significant
Lack of social reciprocity (n=88)	66 (80.49%)	12 (60%)	5 (100%)	3 (100%)	2 (100%)	0.577	Not significant
Language delay (n=93)	70 (85.37%)	14 (70%)	4 (80%)	3 (100%)	2 (100%)	0.423	Not significant
Echolalia (n=40)	29 (35.36%)	6 (30%)	3 (60%)	1 (33%)	1 (50%)	0.555	Not significant
Absent pretend play (n=45)	31 (37.8%)	9 (45%)	1 (20%)	3 (100%)	2 (100%)		Not significant
Resistance to change (n=37)	25 (30.49%)	10 (50%)	1 (20%)	1 (33%)	----		Not significant
stereotypical movements (n=57)	44 (53.66%)	8 (40%)	1 (20%)	2 (66%)	2 (100%)		Not significant

Out of 112 children, MRI brain was available in 67 children. MRI findings were normal in 39 children with autism (78.00%) and seven children with PDD-NOS (87.50%). All children with Asperger's, CDD and Rett's disease showed normal findings. Abnormalities were seen in 11 (22.00%) children with autism and in 1 (12.50%) child with PDD-NOS. Various abnormalities noted in children with autism on MRI were periventricular leucomalacia (4), agenesis/thinning of corpus callosum (3), followed by generalized atrophy and delayed myelination (1), dysplastic changes in left temporal lobe (1), infarct in right lentiform nucleus (1), hypoplastic corpus callosum (1). No significant difference was noted in MRI brain findings and different types of ASD.

DISCUSSION

Leo Kanner first described the prototypic disorder in this group, autistic disorder, in 1943 as an "inborn disorder of affective contact" or "early infantile autism".[1] Until the mid-1980s, autism was wrongly viewed as a psychiatric disorder caused by parents who did not adequately love their children.[1] There is no one single unified theory that explains the etiology of autism. Possible etiologies include genetic factors, perinatal stress, and a combination of genetic and environmentally induced brain damage.[11]

Few cases of children with Autism were reported in the Indian scenario in the 60's and 70's.[12-15] There are very few studies in India about behavioral characteristics on autism.[15,16]

There has been a documented increase in the prevalence of autism worldwide.[17,18] There is no data available from India regarding prevalence rates of autism in the general population. However, authors of recent case series reports of 16 and 62 autistic children from tertiary hospitals in Chandigarh and New Delhi, respectively, have stated that, autism is not uncommon in India.[19] Its diagnosis is frequently missed, as there is tremendous lack of awareness and knowledge about the disorder among health professionals.[20] According to the recent survey 1 in 250 children in India are likely to be autistic.[4]

A recent meta-analysis of 37 prevalence studies of autism reported from USA, UK, European countries and Japan has estimated that boys are affected more often than girls and the average male: female ratio is 3.8:1.21 In our study M:F ratio of 3.67:1 was consistent with the literature. Higher prevalence of autism in men than in women, has led to the suggestion of strong genetic basis and possibility of relation with prenatal brain exposure to androgens. Most children-77 (68.75%) enrolled in the study late (7.31+/-2.91 year), for the purpose of certification although the parents identified the core symptoms between 2-4 year of age in most of the cases. Early recognition of symptoms also possible by 18-20 months.[22] In a similar studies from India and abroad reported the mean age of presentation 3.5 to 4.5 years with male:female ratio of 2.20:1.[22,23] Behavioral symptoms of ASD appear to emerge over time, beginning in the second half of the first year of life and continuing to develop for several years.[23]

Approximately 25% to 30% of children with ASDs begin to say words but then stop speaking, often between the ages of 15 and 24 months.[24] In our study; parents of 18% children reported regression in the language area without regression of motor milestones, seizures or any other physical illness.

Language delay was the commonest developmental concern in 93 (83.03%) in our study. Impairment of verbal and nonverbal communication was a major feature in all subgroups of children with ASD. High functioning children had language peculiarities like repetitive language and echolalia. Absence of speech, or abnormal speech, especially when it is associated with the lack of desire to communicate and lack of nonverbal compensatory efforts such as gestures is always a common finding in children with ASD. However, children with milder symptoms, especially those with normal cognitive skills, may have some speech. Their speech may not be functional or fluent and may lack communicative intent with abnormalities of semantics and pragmatics.[1,3,25]

Children with ASDs universally demonstrate deficits in social relatedness defined as the inherent drive to connect with others and share complementary feeling states.[3] Often parents fail to recognize the warning signs of social deficit in children younger than 2 years.[25,26] Children with ASDs often do not appear connected to others; they are content being alone, seldom make eye contact or do not often seek for other's attention with gestures or vocalizations.[3,25,26] In our experience similar social deficit was observed.

Stereotypical movements, rocking, spinning, hand flapping movements were observed as common motor mannerism in almost 50% of study group. Stereotypic behavior begins before the 3 years of age in children with autism or PDD-NOS. Excitement, stimulation, stress, anxiety, boredom, fatigue, sensory isolation, or social demands increase stereotypies. Forty-four percent of children with autism have at least one subtype of stereotypy.[27]

In contrast to typically developing children, Autistic children show specific impairments in symbolic play as early as 18 months of age. Abnormalities of play like solitary play, lack of domestic mimicry, absent pretend play (40.17%) and lining up of toys (33.03%) were observed in some of the children in our study. When

children with autism do acquire symbolic play skills, their level of symbolic play tends to remain below that of their language level and is often less diverse and elaborate compared to that of typically developing children.[28]

We could not significantly correlate neuroimaging findings with behavioral phenotype. Periventricular leucomalacia, thinning of corpus callosum, right lentiform nucleus infarct, dysplastic changes, delayed myelination, early posterior vermian atrophy, generalized atrophy were some of the findings in our study. In recent research, Functional MRI evidence had been used to postulate impaired "connectivity" between various cortical regions in the brains of people with ASDs. A MRI-based morphometric study of the total corpus callosum volume and its seven subdivisions showed reductions in the total volume of the Corpus callosum and several of its subdivisions in autism.[29] Associations were also seen between corpus callosal structures and clinical features including social deficits, repetitive behaviors, and sensory abnormalities.

Clinical profile and behavioral pattern of the autistic spectrum of disorders have a common a triad of impairments in social interaction, communication, and narrow and repetitive patterns of behavior. Hence the reason why development of language and the communicative competence of children with autism has been an important focus of interventions for children with autism and a number of language training interventions have been developed over the years. The period between initial recognition of a symptom and initial diagnosis is important, because it can serve as a window into broader cultural factors and allows for more immediate behavior-based intervention, which is associated with improvement in core areas, such as social functioning and communication.[3,20] Children participated in our study, after diagnosis were enrolled in symptom based personalized integrated interventions in the same centre. Interventions were based on facilitating communication, improving social relatedness, decreasing aggression and hyperactivity, control of stereotypy's and sensory integration. Regular follow up and clinical monitoring was done to see the improvement in behavioral and developmental profile of these children. Assessment at the end of 6 months and one year had shown better eye contact, improved social relatedness and attempts to communicate in 30% of children. Most of these children are still undergoing comprehensive therapy under expert guidance and relentless efforts are taken for gradual mainstreaming of these kids. Early intervention is crucial because it is likely to lead to an improved outcome.

CONCLUSION

Our study supports the fact that although various behavioral features differ characteristically in different sub-groups of ASD, all of these are bound together by unified core areas of impaired social functioning, ritualistic behavior and lack of communication skills. Increased awareness for early recognition of symptoms among parents and prompt referrals by physician after the initial suspicion of Autism would help in early diagnosis and appropriate therapeutic intervention resulting in favorable outcome.

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