Brief Reports

A Descriptive Hospital Based Study of Children with Autism

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The clinical and neurodevelopmental profile of 51 children with Autistic disorder, from a referral population was studied. A detailed history and examination was undertaken. The development/intelligence quotient and social quotient were assessed. Standard diagnostic tests were applied in children above 2 years of age. Younger children were evaluated with a screening test and re-evaluated at 2 years. It was observed that a correct diagnosis had been made in only 5.8% of children prior to referral. The mean age of presentation was 3.28 years. Ninety-six percent of the autistic children had developmental delay. Qualitative impairment in social interaction and communication was more commonly observed than restricted interests and activities. Forty-seven (92.15%) children were severely autistic and 4 (7.84%) mildly to moderately autistic. All children less than 2 years were confirmed to have Autistic disorder later.

Key words: *Autism, comprehensive assessment, screening.*

AUTISTIC disorder (AD) is one of the pervasive developmental disorders (PDD), which also include Rett Syndrome, Childhood disintegrative disorder, Asperger's disorder and PDD not otherwise specified. AD is characterised by qualitative impairments in communication, social interaction and imagination, restricted interests and stereotyped repetitive behaviors(1). It is still a relatively underdiagnosed or misdiagnosed disorder(2). Parents may deny the existence or fail to recognize certain behaviors as abnormal or may believe that the child will outgrow them. Lack of awareness among medical professionals often leads to delay in establishment of a correct diagnosis. This has important implications, as early intervention improves the long-term prognosis(3). Genetic counselling is important since the recurrence rate is 2-7%(4).

The present study was planned with the aim of understanding the demographic background, different clinical presentations and neurodevelopmental profile of AD in a referral population of children.

Subjects and Methods

The study was conducted in the child development clinic of a tertiary care hospital over 2 years. All children diagnosed as having AD were included. Those with co-morbidities of motor, visual and hearing deficits were excluded. History of the developmental milestones, degree of social interaction, verbal and non-verbal communication skills (use of

spoken language, facial expressions or gestures), type of play and socio-economic background were taken. Any behavior problems, peculiarities or stereotypies were recorded. The children's daily routine was enquired into. Detailed physical and neurological examination was undertaken. The children were directly observed and assessed for features like the degree of eye-to-eye contact, social interaction with the parents and examiner, type of play, stereotypic behavior and reaction to the new environment.

The diagnostic criteria for Autistic disorder of the Diagnostic and Statistical Manual of Mental Disorders (DSM IV), Autism Behavior Checklist (ABC) and Childhood Autism Rating Scale (CARS)(1,5,6) were used for diagnosing children older than 2 years. Younger children were screened by the Checklist for Autism in Toddlers (CHAT) at 18 months of age(7). They were reassessed at 2 years. The 'Early Developmental Profile' (EDP) was used to assess the development quotient(8). The Stanford-Binet Scale (Indian modification) was applied to estimate the intelligence quotient in children older than 5 years(9). The Vineland Social Maturity scale (Indian adaptation) was used for assessment of social quotient(10).

Electroencephalograms (EEG) were done in children with seizures. Neuroimaging of the brain was performed in children with localizing neurological signs. Brainstem evoked response audiometry was done to evaluate the hearing status.

Results

Sixty-four children were initially enrolled. Three children were excluded due to optic atrophy and cerebral palsy, while ten children were lost to follow up. In effect, 51 children constituted the actual study population. Only four had been referred with a diagnosis of PDD; three as AD and one with Rett syndrome. Thirty-three children were referred as developmental delay, 7 as speech delay, 5 as mental retardation and 2 as attention deficit hyperactivity disorder. The demographic characteristics of the children are presented in *Table I*. The mean age was 3.28 years. The boy:girl ratio was 2.2:1. There was no significant family history in any child. Perinatal complications were reported in 14 patients.

The major clinical symptoms observed are described in *Table II*. The commonest complaints were delayed development, speech delay and 'being lost in one's own world'. Difficulties in toilet training were reported in 61% of children. Majority of the children had impairment in 'joint attention' (the ability to use eye contact and pointing for the social sharing of experiences). Each one played abnormally. While 53% of children were not interested in toys at all, 84% used toys inappropriately (banging, spinning tyres, lining up, etc). Imaginative or 'pretend' play was absent. Some children (33%) would go to great extents to obtain a desired object by themselves if it was out of reach, instead of simply asking for it. Echolalia was seen in two out of the three children who had meaningful speech.

Behavior problems like hyperactivity (25.4%), aggression (19.6%) and self-injury (9.8%) were reported. Pica was seen in 21.5%, with children eating carpet threads to pieces of rubber tyres. Seizures were present in 16 (31.3%) children. Five (9.8%) of them had generalised tonic clonic seizures with generalised discharges seen on EEG. Eleven (21.56%) children had myoclonic seizures with EEG changes consistent with myoclonic seizures in 3, hypsarrhythmia in 3 and normal discharges in 5.

Twenty-one patients (41.2%) had normal head circumference. The rest had micro-

TABLE I–*Demographic Characteristics of Autistic Subjects (n = 51).*

Characteristic	Sub-group	Number	Per cent
Age at initial presentation	< 2 years	10	19.6
	2 - 5 years	38	74.5
	> 5 years	3	5.8
Sex	Male	35	68.6
	Female	16	31.3
Religion	Hindu	40	78.4
	Muslim	7	13.7
	Sikh	4	7.8
Socio-economic Status	Upper	9	17.6
(Modified Kuppuswamy Scale)	Upper Middle	17	33.3
	Lower Middle	18	35.3
	Upper Lower	2	3.9
	Lower	5	4.8
Type of Family	Nuclear	27	52.9
	Joint	24	47.1

cephaly. One patient had tuberous sclerosis. Hypotonia and toe walking were commonly seen. Hearing was normal in all children.

All the children fulfilled the diagnostic criteria for AD of the DSM IV. Forty children had ABC scores above 67, considered to be suggestive of AD(11). The mean score was 72.4 ± 14.2. All children had CARS scores in the autistic range, with 47 (92.15%) children being severely autistic and 4 (7.84%) mildly to moderately autistic.

Autistic features noted in the ten children younger than 2 years included lack of social smile, failure to identify with own name, lack of pointing, poor eye contact, and presence of stereotypies. All failed screening by CHAT, suggesting a risk of AD. On reassessment at 2 years, all met the DSM IV criteria and had ABC and CARS scores in the autistic range.

There was global developmental delay in 46 children on developmental testing; mild-5.8%, moderate - 33.3%, severe - 29.4% and profound - 27.4%. The average global development quotient was 32.54 ± 16.67. Only 2 children had normal values. The 3 children, older than 5 years had severe mental retardation. The mean social quotient was 34.4 ± 20.13. The difference between the global development quotient and social quotient was not significant.

Discussion

Fifty-one children were diagnosed as AD. Forty-eight (94.12%) among these were referred with other diagnoses. This reflects the lack of awareness about autism among medical professionals.

In the present study the average age of presentation was 3.28 years. The age of presentation was significantly earlier in first-born children (2.28 years) as compared to later-born children (3.6 years). This is in contrast to another study where no significant association was found with the birth order(2). This observed difference might be attributable to parents spending more time with first-born children.

TABLE II–*Major Clinical Symptoms of Autistic Patients.*

S. No.	Clinical features	No.	%
1.	Delayed development	49	96.1
2.	No meaningful speech	48	94.1
3.	Lost in one's own world	46	90.0
4.	Not responsive to people's facial expressions or feelings	45	88.2
5.	Does not reach out when reached for	44	86.2
6.	Inappropriate use of toys (spinning wheels, banging, mouthing, *etc.*)	43	84.3
7.	No visual reaction to a new person	43	84.3
8.	Avoids eye contact	38	74.5
9.	Doesn't cling when held in arms	33	64.7
10.	Difficulties with toilet training	32	62.7
11.	Will feel, smell or taste things in the environment	26	50.9
12.	Strong reactions to changes in environment or routine	23	45.0
13.	Rocking for long periods of time	19	37.2
14.	Is stiff and hard to hold	16	31.3
15.	Complicated rituals such as 'lining up'	11	21.5
16.	Using parent's hand as a tool	4	7.8

Most children were from the middle class (*Table I*). This is probably since upper class patients usually do not avail government hospital medical facilities. Parents from lower income groups may postpone seeking medical attention for disorders other than sickness.

The observed 2.2 : 1 boy-girl ratio was lesser than that reported in literature, which is 3-4 : 1(12). Male predominance is less marked in autistic patients with mental retardation. In the present study 96% children had developmental delay, whereas mental retardation has been reported in 75% of children with autism previously(4). Perinatal events were reported in 27.5% of children, which is in line with one of the theories of causation centering on pre and perinatal brain injury(13).

Fifty-six per cent of parents reported that their children felt, smelt or tasted things deviantly. Children with autism frequently react paradoxically to particular sensory stimuli; being sometimes hypersensitive and sometimes indifferent to certain stimuli or pain. The sensory processing abilities are aberrant in 42% to 88% of autistic individuals(14). Absence of imaginative play was seen universally. This is characteristic of autism. Motor stereotypes like hand flapping and rocking were commonly observed. Self-injurious behavior was seen in 10% of children. This has been attributed to increased levels of endorphins(15). A peculiarity noted in many was a ritualistic preference for walking or lying on narrow parapets or the edges of stairs.

Approximately 25% of children with AD have macrocephaly, which usually appears in early to mid-childhood(16). In the present study, 58.8 % children had microcephaly. Another study has also reported microcephaly in 15.1% of their subjects, where an association was observed with low-functioning autistic subjects(17). Further studies are warranted to see whether a correlation exists between microcephaly and the severity of autism or associated mental retardation.

Nine (17.6%) patients had hypotonia. This is considered to be due to abnormalities in

Key Messages

- All children should be formally monitored for developmental progress routinely.
- AD should be suspected in any child with global developmental delay who has a disproportionate lag in the social/emotional and language spheres.

cerebellar and parietal lobe functioning. Toe walking, seen in 11.7% of the children is frequently found in autistic individuals(15). Seizures were seen in 31.3%, which is in concordance with pre-existing data. A third of autistic individuals particularly those with mental retardation or motor deficits, develop epilepsy by adolescence(18). However, a high incidence of myoclonic seizures (21.56 %) was observed, which has not been reported earlier.

Three DSM IV criteria were not applicable in a majority of children. These were impaired development of peer relationships (which is difficult to gauge in young children) and impairment in initiation and sustaining conversation and stereotyped and repetitive use of language (which is only applicable when there is meaningful speech). Despite this all the children still had at least 6 positive criteria, essential for the diagnosis of AD.

The features of AD in children less than 2 years are subtle and difficult to define and need to be reassessed at 2 years(19). In the present study, all children in this age group suspected as having AD by CHAT were confirmed. Previous studies have demonstrated that autism can be diagnosed in young children with stability of diagnosis over time(20). Early identification is especially important, as early intervention programs has resulted in significant improvement in cognitive and behavioral functions(3).

The developmental testing by EDP revealed lower performance in social and language domains as compared to the performance in gross motor and activities of daily living. Significantly, delayed social skills relative to overall developmental functioning is one of the characteristic features seen in AD(16). However this difference was not significantly appreciable on assessment of social quotient by VSMS. It is probably because this scale predominantly includes activities of daily living skills in young children.

Since there is no pathognomonic sign or laboratory test to detect AD, the diagnosis is made on the basis of the presence or absence of a constellation of symptoms. Pediatricians must increase their knowledge and their ability to recognize this disorder for early diagnosis.

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