A retrospective study of toddlers with autism spectrum disorder: Clinical and developmental profile

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Abstract

Objective: To retrospectively examine the developmental and clinical characteristics of children with autism spectrum disorders (ASD) in the first 2 years of life in order to narrow the interval between parental concern and getting a reliable diagnosis of autism. **Materials and Methods:** The case records of 21 children in whom a diagnosis of ASD was made in the first 2 years of life and confirmed 6 months to 1 year later were examined. The inclusion criterion was absence of neurological, metabolic, or genetic disorders and sensory or motor impairments. These case records were maintained in the Pediatric Psychology Clinic at the Department of Pediatrics of a tertiary care teaching hospital in North India. **Results:** The average age at presentation to the clinic was 21.23 months (SD = 2.18). The clinical characteristics that were found in two-thirds or more children included lack of speech, inability to follow verbal commands, lack of pretend play, no index finger pointing, difficulty in playing with toys in a constructive manner, lack of joint attention, and motor stereotypies. The mean IQ was 66.62 (SD = 15.11) and the mean SQ as measured by the Vineland Social Maturity Scale was 80.43 (SD = 17.45). **Conclusions:** Given the validity of early diagnosis over time, clinicians should be encouraged not only to make an early diagnosis but also to initiate early interventions in children with ASD.

Key Words

Autism spectrum disorders, development, India, toddlers

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Introduction

Autism spectrum disorders (ASD) are neurodevelopmental disorders which are characterized by qualitative deficits in areas of reciprocal social interaction, verbal and non-verbal communication, and a preference for repetitive, stereotyped behaviors and interests. These characteristics must be present before 3 years of age in order to meet the Diagnostic and Statistical Manual of Mental Disorders (DSM) IV criteria for an ASD diagnosis.[1] Recent reviews of ASD epidemiological studies have reported higher estimates of incidence and prevalence than earlier studies and the current median ASD prevalence estimate is about 62 in 10,000.[2,3]

One of the key research priorities in autism research is on the identification of early biological and behavioral signs or symptoms of ASD, as it is hoped that this would facilitate the identification of underlying genetic and neurobiological mechanisms, improve early screening, and provide an impetus to research focused on behavioral or biological treatment.[4-6] Moreover, considerable evidence has accumulated to support the finding that early interventions in children with ASD lead to immense improvement in language, cognition, and behavior functioning.[7]

Despite the fact that symptoms of autism present early, the age of diagnosis is typically not before 4 years. In India, most children with ASD are diagnosed between 3 and 6 years of age.[8-10] For instance, Daley, in a study of 98 ASD children in India, found that parents reported recognizing symptoms at an average age of 26 months and spent on an average of 2 years between the time they initially recognized the child's problems and took the child to a clinician and had it diagnosed. Parents, on an average, reported consulting 3.5 clinicians before receiving a reliable diagnosis.[10] Moreover, studies indicate that many children may get wrong diagnostic labels such as Attention Deficit Hyperactivity Disorder (ADHD) or mental retardation.[10,11] A number of factors have been implicated

in delayed diagnosis, including limited knowledge of ASD among physicians, lack of awareness regarding screening and diagnostic ASD instruments for young children, and doubts about the validity of an early diagnosis of ASD.[8,10,11] Most parents also express early concerns about the development of their children and notice behavioral abnormalities generally in the first 2 years of life.[12-14] However, confirmation of diagnosis does not occur till 3-4 years of age or later, and this often results in extended period of time before referring families of children with ASD to specialists for assessment.[10,15]

There is no study from India which has examined the clinical and developmental profile of autism in toddlers. Keeping this in mind, the aim of the study was to retrospectively examine the developmental and clinical characteristics of children with ASD in the first 2 years of life in order to narrow the interval between parental concern and getting a reliable diagnosis of autism.

Materials and Methods

Participants
The participants in the study included 21 (19 boys, 2 girls) children in whom a diagnosis of ASD was made at age 2 years or less. The inclusion criteria for all children were a chronological age of 2 years or less, absence of neurological, metabolic, or genetic disorders, and absence of sensory or motor impairment. The case records of these children were maintained in the Pediatric Psychology Clinic at the Department of Pediatrics of a tertiary care teaching hospital in North India. All the children had received multidisciplinary evaluations that included neurological, diagnostic, cognitive, and behavioral assessments. Information regarding socioeconomic and demographic variables such as household income, number of years of parental education, IQ scores, scores on the Childhood Autism Rating Scale (CARS), scores on the Vineland Social Maturity Scale, and scores on the Developmental Profile II (DP II) was available for all the participants, as these measures are routinely administered to all children in whom a diagnosis of ASD is made. Scores on the Modified Checklist for Autism in Toddlers (M-CHAT) were available for only those children in whom a diagnosis of ASD was made after 2003 (*n* = 16). All the scales were individually administered to the parents by the first author. A description of the sample characteristics is presented in Table 1. All analyses were done using the SPSS 13.0 statistical software.

Table 1: Sample characteristics

Characteristic		SD
Mean age (months)	21.10	2.14
Males (%)	90.5 (*n*=19)	
Maternal education (%)		
College graduate and above	80.95 (*n*=17)	
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College graduate and above	80.95 (*n*=17)	
First born (%)	61.9 (*n*=13)	
IQ (Mean)	66.62	15.11
SQ (Mean)	80.43	17.45
CARS score (Mean)	37.21	3.54
Severe ASD (%)	62 (*n*=13)	

Measures used

DP II
A detailed developmental assessment was done using DP II,[16] which is a 186-item inventory to assess the child's developmental status from birth to 9½ years. The DP II assesses child's developmental age in five domains, namely, physical, social, self-help, academic, and communication. One of the most important features of DP II is that its academic scale can be converted into an IQ score. The academic scale assesses a range of skills necessary for success in school, including language, cognition, and scholastic accomplishments. The IQ calculated from the academic scale has been found to have moderate to high correlation with conventional measures of intelligence. In the present study, the IQ of the subjects was calculated from the academic subscale of DP II.

Vineland Social Maturity Scale
The adaptive behavior of the child was measured by the Indian adaptation of the Vineland Social Maturity Scale.[17] The scale yields two scores, social age and social quotient (SQ). The SQ is a measure of a child's social competence and the ability to perform daily living tasks.

CARS
The CARS is a 15-item rating scale which elicits information on the child's behavior with respect to socialization, communication, emotional responses, and sensory sensitivities.[18] The child is rated on each item based on the clinician's observation of the child's behavior throughout the evaluation, as well as on the report of the parent. Each item is scored on a range from normal, scored as 1, to severely abnormal, scored as 4. Scores for all the 15 items are summed to yield a total score that ranges from 15 to 60. The cut-off score for diagnosis of autism is 30. Scores from 30 to 37 are categorized as mildly/moderately autistic and scores above 37 are categorized as severely autistic.

M-CHAT
The M-CHAT is a parent-report 23-item questionnaire that is used to screen for autism in children aged between 18 and 30 months.[19] Each item is to be scored as yes or no. M-CHAT is considered failed (screen positive) if any 3 of the 23 items failed, or any 2 of the 6 critical items failed and these include items concerning joint attention (proto-declarative pointing, bringing to show, following a point), interest in other children, responding to name, and imitation.

Kuppuswamy socioeconomic status scale
The revised Kuppuswamy socioeconomic status scale was used to assess the socioeconomic status (SES) of the family.[20] The scale assigns different scores to different levels of education, occupation, and income of the family members, with higher scores indicating higher SES. Based on the total score of each family, five categories of socioeconomic groups are identified: Upper, upper middle, lower, middle, upper lower, and lower.

Diagnosis of autism
The diagnosis of autism was made independently by the two authors, a psychologist and a pediatric neurologist, both of whom have considerable experience in the diagnosis of autism

and related developmental disorders. The authors used DSM IV criteria[1] to base their clinical judgment. Clinical judgment by experienced clinicians is considered to be the gold standard for an ASD diagnosis.[21,22] Only those cases were labeled as ASD in whom there was a consensus between the two authors regarding the diagnosis.

Results

A total of 33 children were diagnosed with ASD at 2 years or less (from 1999 to 2012). However, in the present study, the clinical and developmental profile of only 21 children, i.e. those without a history of significant medical and genetic problems, is being presented. This is primarily being done in order to study the early features of autism in a more homogenous and neurologically normal sample of young children. Twelve children were therefore excluded. Out of the excluded children, six had infantile spasms, one had hypothyroidism, one had impaired vision, and three had abnormal magnetic resonance imaging (MRI) findings. Out of the 21 ASD children included, majority were diagnosed as having Autistic Disorder ($n = 20$) and only 1 child was assigned the diagnostic label of Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS). The average age at presentation to the clinic was 21.23 months (SD = 2.18) and only one child was less than 18 months. The ASD diagnosis was confirmed 6-12 months after the initial evaluation. A little less than two-thirds of the toddlers with ASD had CARS score in the severe range (M = 37.21, SD = 3.54).

The frequencies of reported characteristics of ASD children of age 2 years or less for the total sample and by the severity of autism as assessed by the CARS, i.e. mild/moderate severity (total scores on CARS less than 37) and severe ASD (total scores on CARS 37 and above), are presented in Table 2. The clinical characteristics which were found in two-thirds or more children included no meaningful speech (95.2%), little interest in children (95.2%), no index finger pointing (90.5%), motor stereotypies (85.7%), lack of joint attention (81%), inability to follow verbal commands (81%), lack of pretend play (81%), inability of the child to follow language (81%), unusual play (71.4%), and poor eye contact (66.7%). Children with severe autism, as defined by the CARS scores, as compared to the

children with mild autism, were more likely to show poor response to name ($\chi^2 = 6.10$, $P = 0.013$), have delayed motor milestones ($\chi^2 = 5.25$ $P = 0.022$), and display lack of a social smile ($\chi^2 = 6.10$, $P = 0.013$).

The ASD children displayed a wide variety of repetitive stereotypic behaviors involving the body and objects, including rocking, hand flapping, hand gazing, spinning, and toe walking. Non-functional manipulation of objects was reported including spinning, mouthing, banging objects, licking surfaces, banging doors, twirling ropes, holding objects, lining and stacking, and repetitive manipulation of objects. Little more than one-fourth of the children (28.6%) displayed self-injurious behavior such as repetitive head banging or slapping self.

The M-CHAT scores were available for only 16 children. All the ASD children failed the M-CHAT. The mean number of total items failed was 13.31 (SD = 4.01) and the mean number of critical items failed was 4.25 (SD = 1.53). The IQ scores of the children were based on the DP II academic scale. The mean IQ was 66.62 (SD = 15.11). Twelve children (57.1%) were cognitively delayed (IQ less than 70), one had borderline cognitive abilities (IQ of 70-79), and eight (38.1%) had IQs in the average range of intellectual functioning (IQ of 80 and above). The mean SQ as measured by the Vineland Social Maturity Scale was 80.43 (SD = 17.45). Twelve (57.1%) children had SQs in the average range of adaptive functioning (SQ of 80 and above) and only 5 (23.8%) had SQs less than 70. The most common concerns expressed by the parents at initial presentation were delay in language (66.7%, $n = 14$), motor delay (23.8%, $n = 5$), and odd behavior (9.5%, $n = 2$). Problems related to poor social interaction were not reported spontaneously by any parent. Four parents (19%) also reported developmental regression in their children.

Discussion

In this study, we report on the clinical characteristics of children diagnosed with ASD at age 2 years or less. The diagnosis of ASD in very young children is challenging, although recent research has documented that the diagnosis of ASD in toddlers is valid and stable over time.[23,24] The clinical characteristics found in the majority of the ASD children included lack

Table 2: Frequencies (%) of reported characteristics for the total sample and by severity of ASD symptoms

Characteristic	Total sample % (n)	Severity of autism (CARS scores)		χ^2	P value
		Mild/moderate (%)	Severe (%)		
Lack of eye contact	66.7 (14)	57.1	71.4	0.43	0.513
Lack of social smile	52.4(11)	14.3	71.4	6.10	0.013
Lack of joint attention	81.0 (17)	71.4	85.7	0.62	0.432
Delayed motor milestones	33.3(7)	0.0	50.0	5.25	0.022
Lack of responsiveness to name	52.4 (11)	14.3	71.4	6.10	0.013
Failure to use gestures	90.5 (19)	71.4	100	4.42	0.035
No meaningful speech	95.2 (20)	100	92.9	0.53	0.469
Inability to follow verbal commands	81.0 (17)	85.7	78.6	0.15	0.694
Motor stereotypies	85.7 (18)	85.7	85.7	0.0	1.00
Unusual play	71.4 (15)	83.3	71.4	0.32	0.575
Lack of imaginative play	81.0 (17)	85.7	78.6	0.15	0.694
No peer play	95.2 (20)	85.7	100	2.10	0.147
Self-injurious behavior	28.6 (6)	14.3	28.6	0.53	0.469

of joint attention, unusual play, lack of pretend play, no index finger pointing, difficulty in playing with toys in a constructive manner, inability of the child to follow language, motor stereotypies, and poor eye contact. However, we need to keep in mind that the sample size is relatively small and the results need to be replicated using larger samples. Our findings support previous research which had examined early signs and symptoms of autism in infants and toddlers using a retrospective methodology, including examining of home videotapes of children already diagnosed with ASD[25] or using retrospective questionnaires and interviews asking parents to recall the early abnormal behaviors of children with ASD.[26] These studies have shown that behaviors that consistently discriminate toddlers with autism from those with non-autism developmental delays or typical development are orienting to name, eye contact, social referencing, interest in other children, joint attention, affect sharing, and imitation.[27,28]

Nearly one-third of the toddlers with ASD had motor delays. History of delayed motor development was more prevalent in children with severe ASD. Our findings support previous studies reporting that 50-75% ASD children have significant motor delays compared to normative children.[29] For example, Provost *et al.*[29] assessed motor delay in ASD children aged 21-41 months, and compared their motor scores to those of children without ASD and found that all children with ASD had delays in gross motor skills, fine motor skills, or both.

An overwhelming majority of the ASD children of age 2 years or younger displayed unusual preoccupations, hand and finger mannerisms, and repetitive use of objects. These findings confirm the findings of previous studies.[30,31] For instance, Watt *et al.*[31] found that young ASD children aged 18-24 months demonstrated significantly higher frequency and longer duration of repetitive stereotypic behaviors as compared to children with developmental delay and children with typical development. In fact, repetitive stereotypic behaviors were found to predict developmental outcomes and severity of autism symptoms at 3 years. It seems then that early stereotypic behaviors in the second year are important not only for early identification but also for the prediction of developmental outcomes.

Most parents reported speech delay and inability to follow verbal commands as their main concern for seeking medical help. Previous studies have documented that the most common and first noted concerns are delays in speech and language development, followed by abnormal social responsivity level, medical problems, and behavioral difficulties related to sleeping, eating, and hyperactivity.[13] It is noteworthy that parental concerns related to core ASD symptoms such as social deficits were not raised spontaneously by any parent. Clearly then, parents may raise concerns which may not be very specific to the core symptoms of autism and by themselves may not be reliable markers of an ASD diagnosis.[13] Four different onset patterns in autism have been described in the literature, including very early onset of symptoms without skill loss, late onset of symptoms seen in children with loss of developmental milestones without early symptoms, early onset combined with later skill loss, and late onset without any skill loss. Therefore, it has been suggested in the literature that ASD children may reach the threshold for diagnosis at different ages in the first

3 years of life depending on early or late presentation of symptoms with or without regression.[32,33] Autistic regression has been reported in 20-40% of the cases in previous studies.[32,33] It is noteworthy that a significant proportion of the parents reported loss of developmental milestones in their children, particularly in the language and social domain.

Despite the growing evidence of diagnostic stability, providing reliable diagnoses of ASD can be challenging in very young children. Our study suggests that signs of autism emerge over the first 2 years of life for a large proportion of children with ASD. Given the validity of early diagnosis over time, clinicians should be educated regarding the early signs and encouraged to make a timely diagnosis. Earlier identification would enable ASD children to enroll in intervention services sooner, which would result in improved outcomes.

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