Original Article

Autism - Experiences in a Tertiary Care Hospital

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Abstract. Pervasive developmental disorders (PDD) or Autistic Spectrum Disorders (ASD) include Autistic Disorder (commonest), Asperger’s syndrome, Childhood Disintegrative Disorders, Rett’s syndrome and PDD –NOS (not otherwise specified). Objective: Autism is an important cause of social disability and reported more often from the developed world than from the developing countries. The present study was aimed to establish the diagnosis of autism amongst children with derangements of language, communication and behavior; ascertain and treat the co-morbidities; identify underlying cause and create a sensitivity and awareness among various health care professionals. Methods: Sixty-two of the seventy-five referred patients fulfilled the DSM-IV (Diagnostic and Statistical Manual of Mental Disorder) criteria for autism. Evaluation included a detailed history, clinical examination, IQ assessment, Connor’s scoring for hyperactivity and Fragile-X screening. Management of co-morbidities was done. A follow up of these patients was done. Parents’ assessment of the child was also done. A registry for autistic children was established at the Department of Pediatrics with other major institutions of Delhi. Results: The male: female ratio was 8:1 and missed diagnosis was common. Professional awareness is merited. Behavioral modification by early intervention and stimulation improved the core symptoms of autism. Important co-morbidities included mental retardation (95%), hyperactivity (53%) and seizures (10%) cases. Control of co-morbidities in these children facilitated child’s periodic assessment and implementation of intervention programmes. In the registry initiated 62 patients were enrolled at AIIMS and 6 were identified from other hospitals. Conclusion: Autism does occur in Indian children too. Diagnosis is often missed. Capacity building among health professionals by a more structured teaching of developmental disabilities in the medical curriculum is required. The need to attend to co-morbidities and associated symptoms was clear. The initiation of the registry and beginning of networking was important. [Indian J Pediatr 2005; 72 (3) : 227-230] E-mail : Kalra_veena@hotmail.com

Key words: Autism; Autistic spectrum disorders (ASD); Pervasive developmental disorders (PDD); Networking; Co-morbidities

Autism, the commonest and severest form of pervasive developmental disorders (PDD) is an important cause of neuromorbidity and referral to tertiary hospitals. It is characterized by alterations of social interaction, problems of communication and restricted range of activities and interests. There is wide variability in the prevalence data from the West. The reasons for the variability include lack of recognition of the disorder, errors in diagnosis and lack of ability to distinguish it from disorders that mimic autism. The studies point to the fact that diagnostic criteria need to be applied to avoid misdiagnosis.

The objectives of the present study were to establish a diagnosis of autism amongst children with derangements of language, communication and behavior; ascertain and treat the co-morbidities; identify underlying cause and create a sensitivity and awareness among various health professionals.

MATERIAL AND METHODS

Subjects
Seventeen hundred new children enrolled and 3920 children already registered came for follow up to the Pediatric Neurology Clinic in the past one year. Of these 75 patients were referred to the clinic with the conglomerate of following complaints- poor language development, regression in the isolated domain of language, abnormal behavior, attention deficit, mental subnormality or that the child was different from rest of the children of similar age groups. They were screened for autistic features according to the DSM IV Criteria for autism.

A proforma for evaluating such patients was designed based on clinical profile and DSM IV criteria. Children meeting the DSM IV criteria were subject to a detailed work up comprising evaluation of perinatal events and developmental milestones, cause of referral, details of birth order, family structure, history of similar illness in the family and presence of co-morbidities. History of seizures was enquired into. Stigmata of Tuberous Sclerosis were looked for. A formal Development Quotient (DQ) / Intelligence Quotient (IQ) / Social Quotient (SQ) was done according to the Stanford Binet Intelligence Scale or Vineland Social Maturity Scale. Connor’s scale was used to evaluate hyperactivity. Hearing assessment was done in all using Brainstem Evoked Response Audiometry (BERA). Fragile-X screen was done where consent was available.
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Interventions

An understanding of the fundamental difficulties faced by autistic children were crucial to develop a curriculum and teaching programme for the child. The parents and other family members were educated about the disorder. Each child was observed carefully and intervention in the form of early intervention and stimulation was planned according to the needs and behavior of the child. The Autism Treatment Evaluation Checklist (ATEC) was used to measure the baseline and follow up status in autistic children in the domains of speech/language/communication, sociability, sensory/cognitive awareness and health/physical/behavior. An IQ assessment was done, behavioral therapy and individualized educational training was initiated. Problematic behavior was modified by positive reinforcement i.e. the desired behavior was acknowledged with some reward or reinforcement to increase the frequency of desired behavior. Other behavior techniques used included ‘prompt’ (giving a visual, auditory or tactile clues to help a child complete an activity); ‘fading’ (gradually withdrawing a prompt); ‘modeling’ (becoming a model in front of the child so that the child observes the skill) and ‘chaining’ (teaching a behavior or skill by breaking down to the smallest steps so that the child can handle the behavior). Management of the co-morbidities of autism like seizures, hyperactivity and mental retardation was done. Speech therapy was given to all children. A registry for autistic children was initiated at AIIMS, which circulated the diagnostic criteria among five partner institutions from Delhi. These patients were followed in their parent institutions and notified at the AIIMS registry (e-mail address -autismAIIMS@hotmail.com). The registry is functional and six patients of autism have been notified from Kalawati Saran Hospital.

Follow up

These children were followed up regularly at frequent intervals of 4-6 weeks both by the pediatrician and child psychologist. Parameters during follow up included enquiry into the parents’ assessment of the child as a whole, level of hyperactivity (repeat Connor’s scoring was done at intervals of 3-6 months) after behavioral and pharmacological intervention. Seizure frequency were enquired into and antiepileptics dose modification done if needed. Details in change of social interaction, communication, activities, interests was enquired into. ATEC was used to assess the improvements made in the individual domains of speech/language/communication, sociability, sensory/cognitive awareness and health/physical/behavior every 6 months.

RESULTS

Of the 75 referrals 62 fulfilled the DSM IV criteria for autism. Majority (50%, n=31) of children were referred for language delay or regression in language. (30%, n=20) had deviation from the normal pattern of development or behavior. Additional (20%, n=12) were referred for Attention Deficit Hyperactivity Disorder (ADHD) / Mental Retardation (MR) /problems in school.

The male: female ratio was 8:1. The mean age of presentation and diagnosis at AIIMS was as follows: [<3 yr 15% (n=9), 3-6 yr 78%(n=49) and >6yr 7%(n=4)]. Majority were diagnosed between 3 –6 years of age. No adverse perinatal event was identified in any of the patient. In the present cohort of autistic children 30 children were living in nuclear families.

Based on the DSM IV criteria for diagnosis of autism a simple questionnaire was designed and identified the main presenting features in the present cohort of patients (Table 1).

| Table 1. Main Presenting Features Observed in Autistic Patients at AIIMS |
|-------------------------|---------------------|
| Feature | Percentage % |
| Social Interaction | |
| Poor eye contact | 90% |
| Prefers to be alone | 80% |
| Problems in communication/language | |
| Speech delay | 70% |
| Pretends to be deaf | 85% |
| Repetitive behavior/apparent obsessions | |
| Stereotyped behavior | 60% |
| Extreme restlessness/hyperactivity | 60% |

The mean baseline scores as observed in the ATEC were: in speech/language/communication domain the baseline score was 24-26, in the domain of social interaction the score was 34-36 and in the domain of cognition awareness and physical behavior the baseline scores were 34-36 and 42-44 respectively.

IQ assessment was done in 93% (n=56) cases. Mild to moderate mental retardation (IQ between 31-70) was present in 63% (n=35) cases; borderline intelligence (IQ between 71-89) was present in 32%(n=18) cases and 3 children had normal intelligence. Of these 5 children are going to special schools.

Hyperactivity assessed by Connor’s rating scale was >12 in 53% (n=32) patients. Drug used to control of hyperactivity was Thoridazine (n=30 patients, 0.5-2mg/kg/day) or Methylphenidate (n=2 patients: 0.3-1 mg/kg/day) besides behavioral therapy. Thoridazine was used in patients with co-existing mental subnormality.

Seizures were present in 10% (n=6) of patients and were generalized in nature. They were easy to control with first line antiepileptics (Phenytoin, 5-6 mg/kg/day in single doses or sodium valproate 10-25mg/kg/day) except in one case where Clobazam (1mg/kg/day) was used.

Hearing assessment was done using BERA and was found to be normal in all except one who had unilateral mild hearing loss. Fragile-X testing was done in 30%
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Preliminary analysis of the data revealed a male preponderance, majority being diagnosed between 3-6 years of age. Mental subnormality was present in 95%, hyperactivity in 53% and seizures in 10% of autistic children. Epidemiological studies reveal a wide variation in the prevalence rates for autism. Various studies between 1966-1990 placed the prevalence of autism around 4.3/10,000. Subsequent studies have placed the occurrence of autism at the rate of 1/500-1/1000 people. According to WHO reports it is 1 out of 10,000. If all shades (spectrum disorders) of autism are included it is more like 1 out of 1000. Such estimates require an appropriate set of criteria, which should be based on adequate standards of the population to be screened. In India a proper survey to find out the incidence of the disease is yet to be done.

The present study in concordance with literature revealed a male preponderance with a higher male: female ratio of 8:1. This could be related to the small number of female children either being referred or brought to tertiary institutes. Literature reveals that the disorder occurs in boys 3-4 times more often than in girls. The male preponderance of the disorder could be related to the X- chromosome.

In the present study, 20 families with an autistic child did not consider having another child for fear of having another autistic child. Parental guilt may have contributed. In 10 families a normal child was born after an autistic child. In the present sample population no family had two autistic children. The risk of an autistic sib birth after one autistic child is between 1.5-20%; much greater than the population prevalence of 0.1%.

Environmental factors also influence the autism phenotype indicating a multidimensional etiology including Congenital Rubella and Autism, Autistic Spectrum Disorders (ASD) and 1st trimester Thalidomide exposure, MMR and increased risk for ASD. Neurologic basis of the disease is supported by the association of ASD with seizures and mental retardation. The age at referral to AIIMS and confirmation of diagnosis was late as 78% cases presented between 3-6 years of age indicating lack of awareness and knowledge about the disorder leading to delay  in initiating early intervention programmes. Studies have shown that the diagnosis is often delayed until mid childhood although retrospective reports suggest that most parents identify the onset of first symptoms by 18 months of age. This clearly points out the need for professional orientation.

Tuberous Sclerosis complex has been strongly associated with autism. However, none of our identified autistic children had features of Tuberous Sclerosis. Various studies have shown that 17% - >60% patients of Tuberous Sclerosis complex are autistic. This association increases if there are coexisting mental retardation and seizures. Conversely the number of autistic individuals with Tuberous Sclerosis complex varies from 0.4-3% in various epidemiological studies. This association also increases to about 8-14% in presence of seizures.

The reported association of autism and Fragile X is highly variable. Early reports reported a strong association between fragile X and autism up to 25% in some studies. Other studies have shown the association to be as less as 3-7%. In some studies no Fragile X was found using cytogenetic techniques. In the present study Fragile X was done in 30% patients and was negative in all.

The coexistence of autism and Down’s syndrome is 2%. The present cohort did not show similar association.

Mental subnormality is present in 70% autistic children. In the present study 63% cases had mild to moderate mental retardation and 35% cases had borderline intelligence.

In the present cohort, seizures were present in 10% of autistic children. Two peaks of seizures observed in these children are in early childhood and again at adolescence. Mental retardation, with or without motor abnormalities and family history of epilepsy are significant risk factors for the development of seizures in autistic children. Seizures most commonly observed in these children are Complex Partial Seizures and psychologists and psychiatrists may often miss the presence of seizures as the seizure occurrence may be labeled as an unusual behavior of autism. Seizures could contribute to the regression seen in some children with autism Landau Kleffner syndrome (acquired epileptic
aphasia) is characterized by language regression and may be confused with regressive autism. However, EEG helps to differentiate between the two.

It is concluded that autism is a frequently missed neurobehavioral disorder as there is tremendous lack of awareness and knowledge about the disorder among health professionals. In the past one year we followed 62 patients of autism and there was either a doubt in diagnosis or misdiagnosis in all. Often the treating physician was afraid to label the child as autistic. Diagnosis is based on history, developmental status, core behavior pattern and by observation in several settings. Autism does not meet the criteria for screening but surveillance through preschool years by parent-professional partnership is recommended. Six cardinal features, most frequently reported by families, should alert the treating physician. These include poor eye contact, child’s preference to be alone, speech delay or total lack of speech, pretends to be deaf, repetitive stereotyped behavior or exhibits extreme restlessness / hyperactivity. Limitation of the study was nonavailability of CARS data. The initiation of a registry and beginning of networking were important. Scientifically conducted therapy trials to attend to co-morbidities and associated symptoms were clear in this pilot study. Benefits after pharmacological and behavioral intervention needs to be tried in larger multicentric cohorts.

REFERENCES