

Autism Spectrum Disorder

Developmental disabilities such as Autism, Attention Deficit hyperactivity, dyslexia, conduct disorder affect an estimated 12 million children in the US, namely about one child in five. Though there are no extensive Indian studies, similar concerns are seen amongst our children too.

The latest focus has been on Autism. It is a behaviorally defined developmental disability, a life-long disorder that interferes with the ability to understand what is seen, heard and touched. This can cause profound problems in personal behavior and ability to relate to others. The behaviors can be broadly classified as defective social behavior, language deficits, stereotypy mannerisms and lack of imaginative play.

These may manifest in infancy and become apparent by 3 years of age. The common presentations are not being able to respond to typical interactions of infancy such as contacting caregiver's eyes, smiling or cuddling. They prefer inanimate objects to people, speech is commonly delayed or absent, and if it does emerge, it comes as a monotone and sounds very odd. They cannot initiate social games, they fail to imitate, hence do not have pretend play, which is an important red flag. They often keep themselves occupied by repetitive stereotypy movements of hands and fingers.

It is interesting to note that no two children are same in their clinical presentation, and hence Autism has been addressed as Autism Spectrum Disorder (ASD) where we get to see a variety of presentations from mild to severe.

The present diagnostic criterion is as per Diagnostic Statistical Manual IV (DSM IV) under the heading of Pervasive Developmental Disorder (PDD), though moves for a developmental diagnostic model are underway.

How big is the problem?

It is of great concern to the practicing pediatricians because of the astonishing rise in the number of cases of Autism between 1991-1997 to a prevalence rate higher than that of Spina bifida, Cancer or Down syndrome. It is possible that there are real differences in prevalence of Autistic Spectrum Disorder in different parts of the world, even in different parts of the same country and at different times. An epidemic of encephalitis, for example, could increase the number of affected children. However, it is very likely that some, even most, of the variations is due to difference of definition and the difficulty of defining the border of sub-groups within the whole Autism spectrum. There are no sharp boundaries separating typical Autism from other Autistic disorders including Asperger's Syndrome.

The individuals who present to the clinician with Autistic symptoms tend to fall into three broad categories – classical Autism, secondly, those with double syndrome – a dual diagnosis of a known medical syndrome plus features conforming to Autism. Then there is a third category where individuals express autistic features for temporary periods in their stages of development.

Autism has been mystifying scientists for more than half a century. It appears to be melting the edges of neurology, psychiatry and pediatrics blending into a new medical discipline.

There has been a gradual shift in the understanding of Autism from psychoanalytic etiology to more accurate descriptions of genetic, infectious and toxic etiologies. Multiple factors are incriminated but it is possible that we are dealing with 2 hit hypotheses, which predicts that in many genetic diseases, a second genetic or environmental factor is necessary for the expression of pathology. There is now compelling evidence that Autism is not a single disease entity. Hence, it is thought to be a syndrome rather than a spectrum.

Why we should diagnose it early?

Presently, there is no definite treatment but what has been realized is that early diagnosis and intervention are likely to be associated with better longterm outcomes. All this makes it imperative that pediatricians must increase their fund of knowledge regarding the disorder. Earlier studies estimated the prevalence of Autism to be 4 to 5 in 10,000 persons. Recent estimates are around 1:1000. Currently, there is no available data for the Indian population.

The challenging task of this problem is that there is no pathognomic sign or laboratory test to detect it. Thus the physician must make the diagnosis on the basis of the presence or absence of a constellation of symptoms, parent report, clinical judgement and recognition of early red flags.

Need of the hour!

Presently, the need of the hour is that pediatricians must take the responsibility of early diagnosis of Autism and involve the parents in providing comprehensive care of the child with ASD. This would include provision of care with appropriate educational, rehabilitation, social and special pediatric services. Pediatricians need to create the optimum support system for parents by placing the issue in its proper perspective so that there is no unnecessary intra-professional exchange of these cases or pursuing alternative therapies, and prevent delay in initiating management.

It is essential that pediatricians are familiar with management issues to put this problem in a proper perspective so that there is no delay in treatment. It has been clearly stated that ASD is not infantile schizophrenia and that it is a neurodevelopmental problem and that the behavioral issues change with time and age of their child. It is imperative that pediatricians involve themselves in finding the causative factors like genetics and the environment.

Pediatricians should consider using screening

tools specific for ASD and any language delay should undergo an audiometric evaluation, with a diagnostic otoacoustic emission (OAE) and brain stem evoked responses (BERA). What we should know about Autism is exactly how and when the pathology is triggered leading to the subsequent neurodevelopmental malfunctions. Such understanding and infant screening is the best hope for future prevention and treatment.

IAP's perspective and contribution

Towards this goal, the Indian Academy of Pediatrics has taken a path-breaking Programme on Childhood Autism under the IAP Vision 2007 where the pediatrician, as the first-line professional to every child, is best placed to provide the guiding light. The program aims at increasing awareness of Autism among medical professionals, facilitation of services and rehabilitation of cases with Autism, as outlined below:

- (a) Improve awareness of Autism among medical professionals, especially pediatricians who as frontline doctors may pick up the cases early;
- (b) Survey and epidemiological study;
- (c) Facilitation of services for management and rehabilitation of cases with Autism,
- (d) Serve as a support group for the parents of children with Autism
- (e) Contribute to the research and study of Autism.

We sincerely hope this initiative from IAP side will go a long way in decreasing the misery of children with Autism Spectrum Disorders.

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