Leading Article

Autism – the hidden epidemic

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Introduction

Autism has currently become a subject of discussion to the extent that it overshadows all other developmental disabilities in children put together. Autism is a highly disabling condition with life long functional impairment, especially in the areas of interpersonal behaviour and communication. Early diagnosis and treatment is vital as it increases the probability of a good outcome, whereas delayed intervention compromises child’s quality of life. It is also imperative that the health and educational interventions have to be highly specialized, structured and intense and target multiple domains of developmental deficits.

Epidemiology

Many childcare professionals in different parts of the world believe that the incidence of autism has rapidly escalated in the last decade or more. This claim is based on increasing numbers of children seen in different clinical settings, who fulfill the diagnostic criteria for autism. In the 1960s, it was assumed that autism was rare and associated with serious handicaps and intellectual disability. Empirical research brought about a change to this concept when core clinical criteria for autism were found in children with normal intelligence, or rarely even superior intelligence. Also, for several decades, the best estimated prevalence was 4 to 5 per 10,000 children whereas, much higher figures are reported in more recent epidemiological studies. For example, a prevalence of 62.6 per 10,000 among preschool children was reported in the United Kingdom, and a rate of 12 per 1000 was reported from Scandinavian countries. More than a tenfold increase in prevalence is shown here. Among the findings in Asia is the result of a longitudinal study in Taiwan, which showed that when compared to birth years of 1996-1999, the rate of autism increased by 14% during the period 2000-2004. In contrast, the same study showed that newly diagnosed mental retardation decreased by 42% to 50% over the same period. Comparable figures were found also in studies conducted in Singapore and United Arab Emirates. One of the highest prevalence rates has come from the United States. A multi-site population study reported prevalence rates among 8 year olds that ranged from 1 in 3003 to 1 in 94 in 14 different states, with an average rate of 1 in 150 or 6.6 per 1000. The variation in prevalence figures reported in different studies is based on different selection criteria used. For instance, some studies have been more inclusive in using the broader definition of Autistic Spectrum Disorders (ASD) rather than the narrower concept of Autistic Disorder. Some authors however, do not agree that the prevalence of autism is increasing. These opponents too acknowledge the rise in numbers of children diagnosed with autism, but explain this on the use of broader diagnostic criteria, which enables more cases to be picked up. A greater public awareness about autism is also thought to contribute to more cases being seen in clinical practice. Yet other authors believe that the broader diagnostic concept is only partly responsible and a degree of a true rise in prevalence cannot be firmly excluded. This is because many questions about the multifactorial aetiology of autism still remain unanswered.

Diagnosis of autism

Autism is a developmental brain disorder, the signs of which begin before the age of 3 years and lasts throughout life. Deficits in development in three broad categories are required for diagnosis, namely impaired reciprocal social interaction, impaired communication and use of language, and restricted repertoire of activities and interests. Affected children show an unusual pattern of development that is evident from infancy. Many favour the use of the term Autistic Spectrum Disorder (ASD), which include children who fulfill the main diagnostic

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requirements but have different levels of impairments. Accordingly, some children may have relatively good verbal skills and minimal language delay, but have significantly impaired social skills. Others may never develop speech and have severely impaired communication and interaction with others. Diagnostic classifications use the broad umbrella term of Pervasive Developmental Disorder (PDD) under which is included autism, pervasive developmental disorder not otherwise specified (atypical autism) and Asperger disorder (AS). Mild forms of the disorder are said to be present in one in five first degree relatives of children with autism. Some developmental adversities and disorders superficially resemble autism in their clinical presentation and may therefore lead to diagnostic error. Such quasi-autistic features are seen in children brought up in severely depriving environments, in receptive language disorder and semantic pragmatic language disorder and in congenital blindness.

**Aetiology of autism**

Genetic basis for autism is well recognized through family and twin studies. Also, autism is associated with other genetic and chromosomal disorders such as Fragile X syndrome and Tuberous Sclerosis. Results of twin studies imply that, when taken together with the population base rate for autism, the heritability or the underlying genetic liability is about 90%. This is the highest figure known among all multifactorial child psychiatric disorders. An infant with an older sibling with autism has a 1 in 20 chance of developing the disorder. The inference from twin and family studies is that there are 3 to 12 susceptibility genes for autism acting synergistically. Interestingly, some ethnic differences in prevalence of autism have been identified in population studies. For example in Sweden, autism among the immigrant populations was found to be higher than host population. Also, autism in higher numbers is found in Afro-Caribbean group in the UK. The reason for these findings is not clear. Furthermore, the genetic basis does not explain why autism is seen four times more commonly in boys.

However, only about 10% of cases of autism can be explained by genetic syndromes and chromosomal abnormalities. There is very little insight into the possible aetiology in the other 90% of cases. This knowledge gap has hindered action on taking any effective measures in prevention. The main hypothesized cause of the mumps-measles-rubella (MMR) vaccine has been consistently found to be negative. The number of cases of autism has continued to rise despite the elimination of ethylmercury-containing preservative thimerosal from most childhood vaccines since 1997. Intraterine infections, exposure to toxins and certain metabolic disturbances have also been identified as aetologically important in autism, based on case reports and studies on clinical samples. More recently, hazardous environmental pollutants have been implicated as a possible direct or indirect cause in the development of autism. Similarly, the effect of prenatal exposure to organophosphate (OP) insecticides from agricultural and household use has also come under suspicion, causing serious concern in many parts of the world. The effect of OP in the pathogenesis of autism is thought to be mediated through prenatal exposure during the critical period of neurodevelopment, interacting with a genetic vulnerability in paraoxonase activity in the fetus.

**Implications for Sri Lanka**

It is clear that autism is present in far greater number than was initially thought. The health and education services in Sri Lanka are only just beginning to wake up to this reality, but ignorance is still widespread among health professionals and the general public. Autism is a highly disabling condition with life long functional impairments and a significant burden of care. The best options for Sri Lanka lie in detection of autism as early as 18 months, and appropriately intervene to reduce the severity of impairment. In this regard, the responsibility falls mainly on the paediatric and primary healthcare services. Checklist for autism in toddlers (CHAT) and the modified version (M-CHAT) are useful tools in early screening in primary healthcare setting. These tools have high specificity and positive predictive value for autism. Lack of joint attention behaviour has emerged as a key indicator of autism detectable at an early age and is now considered a “Red Flag" sign for very early screening. Parents’ concerns are reliable indicators that should not be dismissed. Anticipated development of Community Paediatric services is a valuable opportunity to set up multidisciplinary teams to provide early screening, recognition and intervention. Home based interventions have proved effective with participation of mothers trained to carry out structured programmes of work. Finally, scientific research into identifying the preventable environmental causes of autism that is relevant to Sri Lanka is of immense importance.
References


