Etiological Evaluation of Global Developmental Delay

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To the Editor: Global developmental delay (GDD) is one of the most common chronic disorders of childhood and occurs in 2–3% of general population [1]. There is lack of studies regarding etiological evaluation in southern India, where consanguineous marriages are common. Etiological evaluation helps in the management and antenatal detection and thus, this study was planned.

The present study is a descriptive cross-sectional study. All children aged between 6 mo and 12 y with GDD: Developmental quotient (DQ)/ Intelligent quotient (IQ) lesser than 70, visiting the neurodevelopmental clinic at tertiary care center, Bangalore from January 2016 through January 2017 were enrolled. Detailed history, examination and relevant investigations were done. The DQ/IQ assessment was done. Informed consent from parents and ethical clearance from the institutional ethical committee was obtained. Specific laboratory investigations, CT or MRI of brain, thyroid function tests, arterial blood gas (ABG), ammonia, lactate, tandem mass spectrometry (TMS), gas chromatography/mass spectrometry (GC/MS), enzyme assay, karyotyping, chromosomal microarray, next generation sequencing and other tests were done wherever indicated.

A total of 82 children were analysed; most common age group was 6 mo to 5 y and the median age of presentation was 38 mo with male to female ratio of 1.4:1. The investigations showed abnormal MRI/CT brain in 38, abnormal TMS in 3 and abnormal karyotyping in 5 (Supplementary Table 1). Various causes for GDD were genetic in 33(41%) children; chromosomal anomalies, Inborn errors of metabolism (IEM)

and neurocutaneous syndromes - 11 children each (14%) followed by perinatal insult in 20(24%) children, congenital infections and miscellaneous causes in 6(7%) each and unknown in 10(12%) (Supplementary Table 2). The consanguineous marriage of parents was present in 80% of children with genetic etiology compared to 30% in other causes.

In this study, etiological diagnosis for GDD could be made in 72(88%) children. In the study by Jain et al. [1], etiological yield of 82.1% ($n = 83$) in GDD was noted; genetic 51(61.4%), perinatally acquired 17(20.4%), CNS malformations 10(12%), prenatal 3(3.6%) and postnatal acquired causes in 2(2.4%). In the study by Jauhari et al. [2], etiological yield was 54.1% ($n = 66$); of these, 17 were of prenatal origin, 38 were perinatal/neonatal, and 11 were post neonatal. This difference observed in etiology could be due to different geographic distribution of disease spectrum and improvement in genetic investigation compared to previous studies. Most of the causes of GDD are preventable, hence, antenatal screening and careful fetal monitoring are important. Genetic etiology can be partially decreased by avoiding consanguineous marriages in southern India as they are very common in this part of the country. In conclusion, genetic cause was the most common etiology of global developmental delay followed by perinatal insult.

Compliance with Ethical Standards

Conflict of Interest None.

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References

1. Jain S, Chowdhury V, Juneja M, et al. Intellectual disability in Indian children: experience with a stratified approach for etiological diagnosis. *Indian Pediatr.* 2013;50:1125–30.
2. Jauhari P, Boggula R, Bhavne A, et al. Aetiology of intellectual disability in pediatric outpatients in northern India. *Dev Med Child Neurol.* 2011;53:167–72.

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