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Determination of genetic cause in cases of developmental delay: A retrospective study in Indian cohort

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Statement of the Problem: To elucidate the genetic component in patients with developmental delay (DD) through 5 year retrospective data analysis. After the initial workup by the pediatrician, the patients were referred to the department for cytogenetic and molecular testing.

Methodology & Theoretical Orientation: Most detrimental form of developmental delay is global DD which is presented as overall absence of the required milestone in the child including mental, cognitive as well as motor delay. The advent of cytogenetic and molecular biology techniques has been helpful in understanding the genetic etiology of these conditions. DD is affecting 1:150 children. Cases with following indications like attention deficit disorder, attention deficit hyperactivity disorder, global DD, pervasive developmental disorder, pervasive developmental disorder-not otherwise specified, syndromic, intellectual disability, learning disability, specific DD, speech delay etc. were all included in the data analysis.

Findings: A total of 199 cases (age ranging from 5 months to 22 years) were studied. Cytogenetic analysis was performed on 142 patients and chromosomal abnormalities were observed in 8.5% (12/142) of the cases. FISH assays (For Prader-Willi/Angelman syndrome, DiGeorge syndrome, etc.) were performed in 59 cases, positive results were obtained in 3.3% (2/59). Molecular analysis (Fragile X analysis, Rett syndrome, etc.) was done for 44 subjects. One case 2.4% (1/44) revealed an intermediate phenotype for Fragile-X studies. Chromosomal abnormalities were found in 8.4% (12/142) cases. These observations can be further classified as structural abnormalities in 66.7% (8/12), numerical abnormalities in 25% (3/12) and a combination of both abnormalities was seen in 8.3% (1/12) of the cases. Polymorphic variants were found in 4.2% (6/142) of the cases.

Conclusion & Significance: Cytogenetic and molecular tests could successfully elucidate the genetic components implicated in 7.5% of the total cases. As chromosomal analysis can detect the anomalies up to 5MB resolution, advanced techniques like microarray studies would be more helpful in detecting the cryptic subtle rearrangements in unsolved cases.

Biography

Namrata Londhe has completed her Doctoral degree in Human Molecular Genetics at Bhabha Atomic Research Centre, Mumbai. She is currently working in Kokilaben Dhirubhai Ambani Hospital and Medical Research Centre as an expertise in cytogenetics and FISH based assays in constitutional, hematological and solid tumors. During her academic and industrial tenure, she has worked on several cytogenetic and molecular biology techniques and has presented her work at several national and international conferences.

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