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MOLECULAR SCREENING OF XQ27.3 CHROMOSOMAL REGION AND CYTOGENETIC ABNORMALITIES OF FRAGILE-X SYNDROME IN MENTAL RETARDATION PEOPLES OF TAMIL NADU, SOUTH INDIA

A. Sundaramoorthy[1], K. Sasikala[1], V.G. Abilash[2]

[1]Human Molecular Genetics Laboratory, Department of Zoology, Bharathiar University ; [2]Division of Bio Molecules and Genetics, School of Bio Science and Technology, VIT-University

Objective: Fragile X syndrome is the most common inherited form of X-linked mental retardation. The aim of this study was to screen and identify fragile X syndrome-affected individuals in Tamil Nadu (South India) using DNA-based molecular analysis and chromosomal abnormalities.

Materials and Methods: Genomic DNA extracted from 47 (29 males and 18 females) individuals with mental retardation, including 47 controls, was analyzed using polymerase chain reaction, DNA gel electrophoresis and sequence analysis. Individuals with mutation were determined according to the CGG repeat size in the FMR1gene on chromosome Xq27.3. At same set of mentally retarded individuals, including controls 3ml of blood were collected in Lithium heparin tubes. The blood samples were incubated in 72 hours under culture conditions and harvested for G-banding technique and results were guaranteed by SKY techniques.

Results: The sequence analysis shows that the CGG repeat was found in fragile x site which expressed Fragile X syndrome highly when compared to healthy controls. The major Chromosomal aberrations (CA) like deletion, translocation and mosaic were also found. However, when compare with experimental subjects, the controls exhibited low levels of CA ($P<0.05$).

Conclusion: Results of this investigation suggest that, deregulation of the FMR1 gene might have responsible for fragile X syndrome of mental retardation, and CA among the individuals. Further investigation of this study helps to predicts the causes of this syndrome in molecular level (RNA based FMR1gene – Protein).

Key words: Fragile X syndrome, mental retardation, cytogenetic, molecular analysis.

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