Molecular diagnosis of Down syndrome cases

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Introduction: Down Syndrome (DS), the most frequent form of mental retardation, is characterized by well-defined and distinctive phenotypic features. Research DS-related genes is based on studying the genes located on chromosome 21. Understanding the genes involved may help to target medical treatment to individuals with DS. It is estimated that chromosome 21 contains 200 to 250 genes. It has been identified major genes involved in DS characteristics are normally in the region 21q21–21q22.3.

Methodology: Blood samples from 85 DS cases and 30 normal cases were analyzed for D21S11 marker gene. Fluorescent dye-labeled primers were used in PCR amplification of this marker. The PCR amplified product was analyzed by GeneScan software version 4.1 automatically.

Results: The short tandem repeat marker frequency was estimated in triallelic, diallelic, and monoallelic patterns. Seventy nine (92.94%) of 85 DS cases showed 3 distinct peaks for D21S11 marker and 5 (5.88%) DS cases showed two uneven peaks. One DS (1.17%) sample showed only one large peak. For healthy individuals, two peaks were observed in all cases.

Discussion: Polymorphic STR DNA markeris useful for determining the numbers of chromosome 21. The high sensitivity and automation of the procedures suggest a good prospect for use of this method in detection of DS. However, this is a preliminary investigation and a large-scale study is necessary to validate the clinical application of this protocol.

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