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"ATM Gene Mutations Detection in Iranian Ataxia-Telangiectasia Patients "

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Abstract:

Ataxia-Telangiectasia (AT) is an autosomal recessive disorder involving cerebellar degeneration, immunodeficiency, radiation sensitivity and cancer predisposition. The ATM gene on human chromosome 11q22.3 has recently been identified as the gene responsible for ataxia-telangiectasia (AT). The gene mutated in AT, which has been designated as the ATM gene, encodes a large protein kinase with a PI-3 kinase-related domain. More than 100 mutations are broadly distributed throughout the ATM gene. The large size of the ATM gene (66 exons spanning ~150kb of genomic DNA) together with the diversity and broad distribution of mutations in AT patients, greatly limits the utility of direct mutation screening as a diagnostic tool. In this study, 20 families with at least one affected child clinically suspected to have ataxia-telagiectasia were examined and their DNA was extracted and amplified with standard methods. Sequencing methods were used to detect the new point mutation. Four exons which were hot spots for point mutations in ATM gene were detected by PCR-SSCP or PCR-RFLP.

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Ataxia-Telangiectasia

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top