Mutations In Ataxia Telangiectasia (Atm) Gene

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Technology description

Ataxia telangiectasia (AT) is a rare autosomal recessive disease resulting from mutations in ataxia telangiectasia (ATM) gene. It is a neurodegenerative disease resulting in impaired motor function, immune deficiency, radiation sensitivity, and increased cancer risk. The disease onset is in early childhood with symptoms of developmental delays in motor coordination progressing to speech difficulties and difficulty swallowing. ATM gene has been identified through linkage analysis consisting of ~200 families. The gene encodes for a large kinase involved in DNA repair and cell cycle checkpoint. 0.5% to 1.4% of the US population is thought to be heterozygote for the mutation. The carrier state of dysfunctional ATM gene has been linked to increased sensitivity to radiation and cancer risk, such as breast cancer. Numerous mutations in ATM gene have been identified all across the gene making targeted genetic testing for AT disease screening and carrier identification difficult.

Technology Description:

The researchers at Benaroya Research Institute have identified over 70 novel mutations in ATM gene corresponding to AT disease state that can be detected without DNA sequencing. Majority of mutations resulted in early termination or major truncations. These mutations may be detected with single stranded confirmation polymorphism, PCR, or SDS-PAGE analysis of in vitro translation-transcription product based on isolated mRNA. Some of the mutations are ethnically linked and can be identified rapidly using specially designed primers.

Business Opportunity:

The loss of functional ATM protein results in increased radiation sensitivity in the AT patients and those carrying a copy of dysfunctional ATM. This suggests that introduction of dysfunctional ATM gene through tissue specific gene therapy may induce radiation sensitivity to cancer cells therefore increasing effectiveness of radiation therapy with less risk to healthy cells. The mutations may also be introduced into non-human mammals as transgene to be used as a model system to further study and understand the AT disease progress and testing potential therapies.

Application area

tissue specific gene therapy may induce radiation sensitivity to cancer cells therefore increasing effectiveness of radiation therapy with less risk to healthy cells.

Institution

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