Fragile X Syndrome (FRA X), Genetic test

General:

The fragile X syndrome (Martin Bell syndrome) is one of the most frequent causes of a genetically caused mental retardation and affects mainly males. The frequency is indicated as 1:4000 in males and as 1:8000 in women. It is caused by a mutation in the FMR-1 gene (fragile X mentally retardation gene) of the X-chromosome (region Xq27.3). This mutation leads to a repetitive extension of a triplet (cytosine-guanine-guanine; CGG). The FMR-1 function is impaired by increasing CGG-repeat-expansion. The disease can present with different grades of mental retardation, from learning disturbances up to severe mental disability. In infancy the following factors are predominant: mental development disturbance, speech and language dis-turbances, frequently hyperactive behavior. In adolescence and adulthood specific physical characteristics might appear. Intelligence reduction in-creases with age. The clinical diagnosis of the fragile X-syndrome is con-firmed by DNA analysis.

Indication:	mental retardation, analysis of the transmitter status of affected families.
Material:	2 x 10 ml EDTA blood
TAT:	2-3 weeks*
Method:	Southern Blot, PCR
Ref.- range:	see report

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit
http://www.fml-dubai.com/parameter-listings/

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