Martin Bell syndrome

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 FMR1 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 disturbances, frequently hyperactive behavior. In adolescence and adulthood specific physical characteristics might appear. Intelligence reduction increases with age. The clinical diagnosis of the fragile Xsyndrome is confirmed by DNA analysis.

Indication:	mental retardation, analysis of the transmitter status of affected families
Material:	2 x 10 ml EDTA blood
TAT:	5-7 days*
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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