



Mental Retardation and Learning Disabilities

Fragile X DNA Test

The possibility of fragile X syndrome should be considered for any male or female with mental retardation or learning disabilities of unknown etiology. The Fragile X DNA Test provides definitive diagnosis of fragile X syndrome and accurate carrier detection. The test detects the fragile X mutation which is an expanded sequence of CGG repeats in the FMR1 gene. Fragile X DNA testing should also be offered to all individuals with a family history of fragile X syndrome or mental retardation of unknown cause and to all women with premature menopause.

Indications for Fragile X DNA Testing:

- Any male or female with mental retardation, developmental delay, or learning disabilities of unknown cause
- Any male or female with autism or autistic-like characteristics
- Individuals with a family history of fragile X syndrome or mental retardation of unknown cause
- Women with premature menopause
- Individuals with a previous fragile X cytogenetic test result that was positive or inconclusive

In the obstetric setting:

- Pregnant women or their spouses with a family history of fragile X syndrome or mental retardation of unknown cause
- Fetuses of a mother known to be a fragile X carrier

Methodology:

Both Southern blot and polymerase chain reaction (PCR) analysis are performed on every sample. The combination of these two approaches yields the most accurate results and the most detailed information.

Results Include the Following:

- Presence/absence of the mutation
- Mutation category (premutation, full mutation, or mosaicism)
- CGG repeat number
- Methylation status of full mutations
- Genetic interpretation, recommendations, and education

Our Fragile X DNA Test Service Provides:

- Diagnostic testing
- Carrier identification
- Prenatal testing
- Genetic consultation by board-certified geneticists

Specimen Requirements:

- 10 ml blood in an EDTA (lavender top) tube, room temperature
- For prenatal testing: direct or cultured amniocytes; direct or cultured CVS

Turnaround Time:

- 7-11 days

Please call Kimball Genetics for more information.

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