

# Fragile X Syndrome

## Description:

Fragile X syndrome is the most common inherited cause of intellectual disability. It affects predominantly males but affected females may show mild intellectual disability. Large increases in the number of CGG repeats (greater than 200) in the *FMR1* gene result in decreased expression of the FMRP protein and is associated with Fragile X syndrome. Females with between 55 to 200 repeats are considered to be premutation carriers, as they are at risk for passing on an expanded number of CGG repeats in the following generation. Premutation carriers in both males and females have been described with behavioral problems and mild intellectual disability. Males carrying a premutation may develop late-onset neurologic symptoms such as tremors and ataxia (abnormal gait) known as the Fragile X Tremor Ataxia Syndrome (FXTAS). Female premutation carriers are at some risk for FXTAS-type symptoms, but are at a higher than expected risk for developing premature ovarian failure.

## Indications:

- Intellectual disability or developmental delay of unknown etiology predominantly in males.
- Family history of Fragile X syndrome or nonspecific intellectual disability.
- Features of autism in either males or females.
- Women with premature menopause.
- Tremor or ataxia in males over 50 with known family history of Fragile X.

## Specimen:

At least 2 mLs of whole blood in lavender top (EDTA) tube or 2 cytobrushes. Label with patient's name, birth date, and date of collection. Direct amniotic fluid (20ml), chorionic villi, or products of conception (POC)

(10-200mg sterile tissue) acceptable for *FMR1* repeat size analysis only.

## Testing Methodology:

DNA extracted from peripheral white blood cells is analyzed by the polymerase chain reaction (PCR). The PCR fragment is designed to flank the area of repeats and effectively alters the size of the PCR product correspondingly to the repeat size. The DNA of premutation males and full mutation males and females is also subjected to allele-specific methylation PCR (mPCR) followed by capillary electrophoresis to determine methylation status.

## Sensitivity:

Full mutations of the Fragile X gene (*FMR1*) are associated with Fragile X syndrome in >99% of cases.

## Turn-Around Time:

14 days

## CPT Codes:

- 81243, 81244

Please call 1-866-450-4198 for current pricing, insurance preauthorization or with any billing questions.

## Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for the clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

## Shipping Instructions:

Please enclose test requisition with sample. **All information must be completed before sample can be processed.**

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

## Ship to:

Genetics and Genomics Diagnostic Laboratory  
3333 Burnet Avenue NRB 1042  
Cincinnati, OH 45229  
513-636-4474

## References:

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