Fragile X Syndrome

Description:

Fragile X syndrome is the most common inherited cause of mental retardation. It affects predominantly males but females may show autistic features, learning disabilities, or mental retardation. Increases in the number of CGG repeats in the Fragile X gene (*FMR1*) result in decreasing functionality of the FMR1 protein. Repeat size greater than 200 is associated with Fragile X syndrome. Females with repeat sizes of 55 up to 200 are considered pre-mutation carriers, as they are at risk for passing on an expanded Fragile X gene with more than 200 repeats. Premutation carriers in both males and females have been described with learning difficulties, behavioral problems, and even mental retardation. Older males with premutations may develop neurologic symptoms such as tremors and ataxia (abnormal gait) called the Fragile X Tremor Ataxia Syndrome (FXTAS). FXTAS is a late-onset, progressive disorder affecting cognition and behavior. Females with premutations are at some risk for FXTAS-type symptoms but are at a higher than expected risk for developing premature ovarian failure.

Indications:

- Mental retardation or developmental delay of unknown etiology predominantly in males
- Family history of Fragile X syndrome or nonspecific mental retardation.
- 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 2mLs of whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity. Direct amniotic fluid (20ml), chorionic villi, or products of conception (POC) 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 Friday.

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