Fragile X Syndrome

This test is used to determine fragile X gene status in males and females with mild to severe mental retardation, a family history of fragile X syndrome or suspected X-linked mental retardation, or confirmation of diagnosis in patients diagnosed by cytogenetic methods. This test is also used to determine fragile X gene status in males and females with symptoms suggestive of Fragile X Associated Tremor Ataxia Syndrome and in females with premature ovarian insufficiency.

Testing Method and Background

The FMR1 AmplideX PCR (Asuragen) provides the ability to assess the status of the CGG-repeat expansion in the promoter region of the FMR1 gene. Genomic DNA is amplified using the polymerase chain reaction (PCR) and are analyzed through capillary electrophoresis technology. Any samples showing an expansion in the FMR1 gene greater than 50 repeats are reflexed to methylation analysis.

FMR1-related disorders include fragile X syndrome, fragile X-associated tremor/ataxia syndrome (FXTAS) and fragile X-associated premature ovarian insufficiency (FXPOI). Fragile X syndrome occurs in individuals with an FMR1 full mutation and is characterized by moderate mental retardation in affected males and mild mental retardation in affected females. Behavioral abnormalities, sometimes including autism spectrum disorder, are common. FXTAS develops in 40% of males over the age of 50 and 8% of females over the age of 40, who have an FMR1 premutation. FXTAS is characterized by late-onset, progressive cerebellar ataxia and intention tremor. FXPOI occurs in approximately 20% of females who have an FMR1 premutation.

Highlights of Fragile X Syndrome Testing

Targeted Region

FMR1: CGG-repeat expansion in the promotor region

- Any samples showing an expansion in the FMR1 gene greater than 50 repeats are reflexed to methylation analysis.

Ordering Information

Get started (non-HFHS): Print a Genetic Hereditary Disorder requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "Fragile X Syndrome" (DNA2100005)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Extracted DNA - from a CLIA-certified Laboratory

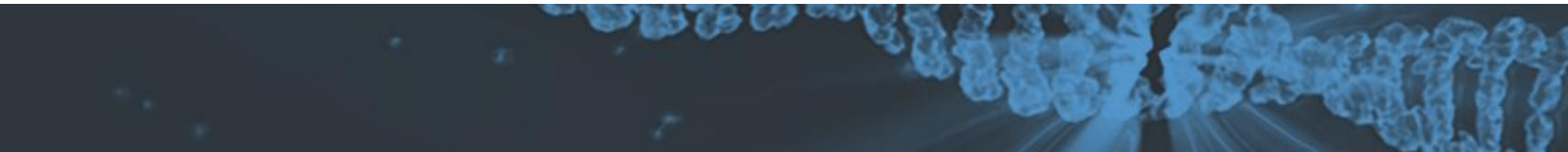
Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-7 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81243, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

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