FRAXE Syndrome: FEMALE CCG Repeats Only

Test Code: MFEXF  
Turnaround time: 3 weeks  
CPT Codes: 81243 x1

Condition Description

In patients who have the cytogenetic changes of fragile X syndrome but lack the molecular changes characteristic of that disorder (i.e., are FMR1-mutation negative), a second site of fragility, symbolized FRAXE, has been demonstrated to be expanded. Data suggests that an etiologic relationship may exist between FRAXE and nonspecific X-linked intellectual disability. Loss of expression of the gene AFF2, also known as FMR2, has been correlated with FRAXE expansion. Sequence analysis and deletion/duplication analysis are available to identify mutations in the AFF2 gene.

For patients with suspected FRAXE syndrome, repeat expansion testing and methylation analysis in males and repeat expansion testing only in females are recommended as the first step in mutation identification. For patients in whom mutations are not identified, full gene sequencing and deletion/duplication analysis are also available.

Click here for the OMIM summary on this condition.

Genes

AFF2, FMR2

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of FRAXE syndrome
- Carrier testing in adult females with a family history of FRAXE syndrome

Methodology

PCR and Southern blot analysis are used to size normal and abnormal CCG expansions.

Detection

All cases of FRAXE syndrome caused by CCG expansion of the AFF2 gene will be detected by this assay. Rare cases of FRAXE syndrome caused by mutation of the AFF2 gene will not be detected by this assay.

Reference Range


Specimen Requirements

Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

Related Tests

- Full gene sequencing and deletion/duplication analysis of the AFF2 gene is available for those individuals in whom repeat analysis and methylation testing is negative.
- Testing for fragile X syndrome at the FRAXA site is available by repeat expansion/methylation analysis, FMR1 sequencing, and FMR1 deletion/duplication analysis.
- X-Linked Intellectual Disability Panel
- Prenatal testing is available to adult females who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.

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