FraX E Mental Retardation Syndrome (FRAXE)

Test Code: 1490

Test Synonyms:
FRXE Syndrome

CPT Code(s):
83890
83892
83894 x 2
83896 x 2
82897 x 2
83898

Background:
FRAX E mental retardation is a mild form of mental retardation associated with the expansion of a CCG trinucleotide repeat located in the \( FMR2 \) gene. Expansion >200 of the CCG is associated with abnormal methylation and resulting repression of the \( FMR2 \) gene.

<table>
<thead>
<tr>
<th>Phenotype (males)*</th>
<th>CTG Repeats</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>≤30</td>
</tr>
<tr>
<td>Indeterminate</td>
<td>31-60</td>
</tr>
<tr>
<td>Premutation</td>
<td>61-200</td>
</tr>
<tr>
<td>Affected</td>
<td>&gt;200</td>
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*Females with one \( FMR2 \) gene GCC repeat in the expanded size range are carriers of FRAX E Mental Retardation and are at risk to be affected by this disorder.

Reason for Referral:
- Confirmation of a clinical diagnosis of FRAXE syndrome
- Carrier testing in adult females with a family history of FRAXE syndrome
- Prenatal diagnosis

Methodology:
PCR analysis to measure the trinucleotide repeat number in Exon 1 of the Frax E gene at Xq 28. The CCG repeat is amplified by PCR and analyzed by denaturing polyacrylamide gel electrophoresis. The size of the PCR product is determined by comparison to molecular weight standards run on the same gel. For very large CCG repeats sizing is also done by Southern blot analysis using the restriction enzyme \( H_{ind} III \). The restriction fragment containing the GCC repeat is detected using an FMR2 specific probe. Sizing of the GCC containing restriction fragment is done by comparison to molecular weight standards run on the same gel.

Specimen Requirements:
- **Blood:** EDTA (purple-top) or ACD (yellow-top) tube
  - Adult: 6.0 mL
  - Child: 3.0 mL
A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information, including ethnicity, clinical history, and family history.

Test Performed (Days):
Weekly

Turn Around Time:
14 – 21 Days

Shipment Sensitivity Requirements:
Package and ship specimen to remain cold, but not frozen. Ship via overnight express, using the FedEx priority overnight label provided. Contact Client Services for shipping kits and instructions at (855) 535-1522.

Additional Information:
Testing is diagnostic for normal, carrier, and disease states.

References: