

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** Fragile X Repeat Analysis
- **Test Code:** 2318

Related Diseases & Genes:

- Fragile X syndrome (OMIM #: 300624) || Gene: *FMR1* (OMIM #: 309550)
- Fragile X tremor/ataxia syndrome (OMIM #: 300623)
- Premature ovarian failure 1 (OMIM #: 311360)

Synonyms:

- FRAGILE X MENTAL RETARDATION PROTEIN; FMRP
- FMR1 Gene CGG Repeat Increase

Test Details:

- **Methodology:** Fragment Analysis; Southern Blot
- **Performed:** Tuesday
- **Reported:** 4 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Neurogenetics

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For yesterday, today and *tomorrows...*

