**FMR1-related Primary Ovarian Insufficiency (POI)**

**Test Code:** 1482  
**Department:** Molecular Genetics

**Test Synonyms:**  
FMR1-Related Disorders  
POI FMR1-related Primary Ovarian Insufficiency

**CPT Code(s):**  
83890  
83898  
83909

**Background:**  
FMR1-related primary ovarian insufficiency (POI) occurs in 20-25% of female FMR1 premutation carriers. POI is caused by a premutation, >54 trinucleotide (CGG) repeats, in the FMR1 gene. The hallmark clinical feature is cessation of menses before age 40 years. Genetic counseling is recommended.

**Reason for Referral**  
Testing is indicated for females with clinical features of early menopause, particularly those with a family history of Fragile X syndrome.

**Methodology:**  
PCR and capillary electrophoresis analysis is used to measure the number of CGG trinucleotide repeats in the 5’ untranslated region of the FMR1 gene at Xq27.3; additionally, CGG repeat size and methylation status is determined by Southern blot for large premutations (>CGG150) and for fully expanded alleles.

**Allele Types #CGG Repeats:**
- Normal: ~5 - ~44
- Intermediate: ~45 - ~54
- Premutation: ~55 – 200
- Full mutation (i.e. disease-causing): >200

**Specimen Requirements:**
- **Blood:** EDTA (purple-top) tube 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:**  
7-10 days
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**Shipment Sensitivity Requirements:**
Keep specimen cool during transit, but do not ship on dry ice. Contact Client Services for shipping kits and instructions. Please use the cold pack provided in the KDL shipping kit. Ship the specimen overnight express, using the FedEx priority overnight label provided. The specimen must arrive at the lab no more than 24 hours after collection.

**Additional Information:**

*Related tests in our laboratory:*
- Fragile X syndrome
- Fragile X-associated Tremor Ataxia syndrome (FXTAS)

Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact Client Services at (855) 535-1522.

**References:**
NCBI GeneReviews: *FMR1-Related Disorders* by Robert A. Saul and Jack C. Tarleton (2012)
http://www.ncbi.nlm.nih.gov/books/NBK1384/