UAB MEDICAL GENOMICS LABORATORY

Fragile X Syndrome Testing (FRX)

Ordering Information

Acceptable specimen types:

- Fresh blood sample (3-6 ml EDTA; no time limitations associated with receipt)

Turnaround time:

15 working days

Price, CPT codes, and Z code:

$280 or $560 if Tier 2 Southern blot is needed (USD – institutional/self-pay);

CPT: 81243, with reflex to 81404 if needed

Z code: ZB6A7

Candidates for this test:

Patients with mental retardation, developmental delay, autistic features, or a family history of Fragile X

Specimen shipping and handling:

- Please find acceptable specimen type above.
- All submitted specimens must be sent at room temperature. DO NOT ship on ice.
- Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping company's diagnostic envelope).

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www.uab.edu/medicine/genetics/medical-genomics-laboratory

- To request a sample collection kit, please visit the website or email medgenomics@uabmc.edu to complete the specimen request form.
- Please contact the MGL (via email at medgenomics@uabmc.edu, or via phone at 205-934-5562) prior to sample shipment and provide us with the date of shipment and tracking number of the package so that we can better ensure receipt of the samples.

Required forms:

- Test Requisition Form
- Form for Customs (for international shipments)

Note: Detailed and accurate completion of this document is necessary for reporting purposes. The Medical Genomics Laboratory issues its clinical reports based on the demographic data provided by the referring institution on the lab requisition form. It is the responsibility of the referring institution to provide accurate information. If an amended report is necessary due to inaccurate or illegible documentation, additional reports will be drafted with charge.

Requests for testing may not be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information
- DNA samples must be extracted in a CLIA or equivalent certified lab

For more information, test requisition forms, or sample collection and mailing kits, please call: 205-934-5562.

Disorder Background

Fragile X syndrome is one of the most common genetic causes of mental retardation in males with an estimated incidence of 1:4000-6250 in males. Males affected with Fragile X typically have moderate mental retardation, and can present with characteristic physical features such as a long face, prominent ears and macroorchidism. Approximately 50% of females with full mutations are affected. Their level of developmental impairment is, on average, less severe than that seen in males. Fragile X is usually caused by a trinucleotide expansion in the *FMR1* gene on the X chromosome. Normal individuals have <40 CGG repeats. Individuals with pre-mutation alleles have 59-230 repeats whereas individuals with intermediate sized alleles ("gray zone" alleles) have 41-58 repeats, broadly defined. Individuals with a full mutation have > 230- > 2000 repeats.

Test Description

Polymerase chain reaction analysis is used a first tier test to define the repeat size. If a homozygous female, premutation, or full mutation is identified or suspected, a second tier of testing consists of *Eco* RI and *Eag* I restriction digest followed by Southern blot analysis using the DNA probe, StB12.3 to determine methylation status of the *FMR1* gene and approximate number of trinucleotide repeats. Polymerase chain reaction analysis is used to further define the repeat number more precisely.

REFERENCES available on website.