

TEST CHANGE

Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation

3000258, CF FX SMA

Specimen Requirements:

Patient Preparation:

Collect: Lavender (K2EDTA). Also acceptable: Pink (K2EDTA).

Specimen Preparation: Transport 5 mL whole blood. (Min: 3 mL)

Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin, yellow (ACD solution A), or lithium heparin tubes. Frozen specimens in glass collection tubes.

Remarks:

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: unacceptable

Methodology: Matrix-Assisted Laser Desorption Ionization-Time of Flight (MALDI-TOF) Mass Spectrometry / Polymerase Chain Reaction (PCR) / Capillary Electrophoresis / Multiplex Ligation-Dependent Probe Amplification (MLPA)

Note: Fragile X: If a CGG repeat of 100 or greater is detected by PCR and capillary electrophoresis; methylation analysis will be added. Additional charges apply.

CPT Codes: 81220; 81329; 81243; if reflexed, add 81244

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

Refer to report.

Reference Interval:

[Refer to](#) [By](#) report

HOTLINE NOTE: There is a prompt change associated with this test. Refer to the Hotline Test Mix for interface build information.