

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~~^{1 week}; Refrigerated: 1 ~~week~~^{month}; Frozen: ~~unacceptable~~^{1 month}

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~~)

Performed: Sun-Sat

Reported: 4-14 days

Note: Cystic Fibrosis (CF): The Cystic Fibrosis (CFTR) Expanded Variant Panel includes the 23 pathogenic CF variants recommended by the American College of Medical Genetics for population carrier screening as well as many others. Fragile X: If a CGG repeat of 100 or greater is detected by PCR and ~~capillary electrophoresis~~^{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.~~^{Refer to report.}

~~This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was~~

~~performed in a CLIA-certified laboratory and is intended for clinical purposes.~~

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

By report
