

Quarterly HOTLINE: Effective May 21, 2018

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**New Test**      **3000258**      **Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation**      **CF FX SMA**

Available Now



Additional Technical Information



Patient History for Genetic Carrier Screen, CF, FXS, SMA

**Methodology:** Polymerase Chain Reaction/Fluorescence Monitoring, Polymerase Chain Reaction/Capillary Electrophoresis, Multiplex Ligation-dependent Probe Amplification

**Performed:** Sun-Sat

**Reported:** 4-14 days

**Specimen Required:** Collect: Lavender (EDTA). Also acceptable: Pink (K<sub>2</sub>EDTA) or Yellow (ACD Solution A).

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

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

**Reference Interval:** By report

**Interpretive Data:** See Compliance Statement C: [www.aruplab.com/CS](http://www.aruplab.com/CS)

**Note:** Cystic Fibrosis: The 165-variant test includes the 23 pathogenic CF variants recommended by the American College of Medical Genetics for population carrier screening.

Fragile X: If a CGG repeat of 55 or greater is detected by PCR and Capillary Electrophoresis; methylation analysis will be added. Additional charges apply.

**CPT Code(s):** 81220, 81401, 81243; if reflexed add 81244

New York DOH Approved.

**HOTLINE NOTE:** Refer to the Test Mix Addendum for interface build information.