

HOTLINE: Effective May 16, 2022

3000258

**Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation  
(Temporary Delay as of 01/21/2021 - no referral available)**

CF FX SMA

**Specimen Required:** Collect: Lavender (K<sub>2</sub>EDTA). Also acceptable: Pink (K<sub>2</sub>EDTA).

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

Storage/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.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

**Interpretive Data:**

**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.

**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; methylation analysis will be added. Additional charges apply.

**HOTLINE NOTE:** **There is a clinically significant charting name change associated with this test.**

Change the charting name for component 2013677, CFystic Fibrosis, Expanded 165 Variants Panel, Interp from CFystic Fibrosis, Expanded 165 Variants Panel, Interp to **CF Expanded Variant Panel Interp.**