ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform molecular genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CYSTIC FIBROSIS (CF), FRAGILE X SYNDROME (FXS), AND SPINAL MUSCULAR ATROPHY (SMA) CARRIER SCREENING

Patient Name _____ **Date of Birth** _____ **Sex** ☐ **F** ☐ **M**

Physician _____ **Physician Phone** _____

Practice Specialty _____ **Physician Fax** _____

Genetic Counselor _____ **Counselor Phone** _____

Patient's Ethnicity (check all that apply)

☐ African-American
☐ Hispanic
☐ Ashkenazi Jewish
☐ Middle Eastern
☐ Asian
☐ Native American
☐ Caucasian
☐ Other _____

Reason for Testing (choose one of the reasons below):

☐ Routine carrier screening; healthy patient with no family history of cystic fibrosis (CF), fragile X syndrome (FXS), or spinal muscular atrophy (SMA)

☐ Family history; healthy patient with a family history of CF, FXS, or SMA:
Name of condition in family _____ Relationship of affected relative to patient: _____
Is relative healthy or affected with disease? _____ Test result of relative: _____

☐ Known carrier; patient is a known healthy carrier of CF, FXS, and/or SMA (has had previous testing):
If yes, which condition? _____ Test result of patient: _____

☐ Other: (e.g., abnormal ultrasound finding). Please describe: _____

Is reproductive partner symptomatic or a known healthy carrier of CF, FXS, or SMA? ☐ Yes ☐ No ☐ NA
If yes: Which condition? _____ Is partner healthy or symptomatic? _____
Please provide test result of partner _____

Does reproductive partner have a family history of CF, FXS, or SMA? ☐ Yes ☐ No ☐ NA
If yes: Which condition? _____ Relationship of affected relative to partner: _____
Is relative healthy or affected with disease? _____ Test result of relative: _____

Check **the test you intend to order:**

Panel that includes testing for carrier status for 3 disorders—CF, FXS, and SMA:

☐ **3000258** **Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation**—carrier screening for cystic fibrosis (CF), fragile X sy (FXS), and spinal muscular atrophy (SMA).

Panel components available as individual tests:

☐ **2013661** **Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants**—carrier screening for cystic fibrosis (CF)
☐ **2009033** **Fragile X (*FMR1*) with Reflex to Methylation Analysis**—carrier screening for fragile X syndrome (FXS)
☐ **2013436** **Spinal Muscular Atrophy (SMA) Copy Number Analysis**—carrier screening for spinal muscular atrophy (SMA)

Targeted testing for known CF mutation (laboratory report from family member REQUIRED)

☐ **2001961** **Familial Mutation, Targeted Sequencing**—targeted testing for a known familial sequence mutation

Additional carrier screening options (Ashkenazi Jewish screening, hemoglobinopathy screening, or expanded carrier screening) are available.

See **www.aruplab.com/genetics** for additional options.

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141

Master Label