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.
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 ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Reason for Referral:

☐ Carrier screen: Patient has no symptoms or family history of cystic fibrosis (CF), fragile X syndrome (FXS), or spinal muscular atrophy (SMA).

☐ Family history is positive for ☐ CF ☐ FXS ☐ SMA

Relationship of affected relative to patient: _____

The relative is: ☐ a healthy carrier ☐ affected

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing)

or describe the test results: _____

☐ Known carrier: Patient is a known healthy carrier of (has had previous testing): ☐ CF ☐ FXS ☐ SMA

Test result of patient: _____

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

Is reproductive partner symptomatic or a known healthy carrier? ☐ No ☐ Yes (specify: ☐ CF ☐ FXS ☐ SMA) ☐ NA

Partner is: ☐ a healthy carrier ☐ affected/symptomatic

Has DNA testing been performed for the partner? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test result: _____

Does reproductive partner have a family history of CF, FXS, or SMA? ☐ No ☐ Yes (specify: ☐ CF ☐ FXS ☐ SMA) ☐ NA

Relationship of affected relative to partner: _____

Relative is: ☐ a healthy carrier ☐ affected/symptomatic

Has DNA testing been performed for the relative? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test results: _____

Check the test you intend to order.

Panel including carrier testing for 3 disorders—CF, FXS, and SMA:

☐ **3000258 Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation:** Carrier screening for CF, FXS and SMA.

Panel components as individual tests:

☐ **2013661 Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants:** Carrier screening for CF.
☐ **2009033 Fragile X (*FMR1*) with Reflex to Methylation Analysis:** Carrier screening for FXS.
☐ **2013436 Spinal Muscular Atrophy (SMA) Copy Number Analysis:** Carrier screening for SMA.

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

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member.

See aruplab.com/genetics for additional carrier screening options
(e.g., Ashkenazi Jewish screening, hemoglobinopathy screening, or expanded carrier screening).

Master Label

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