

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)

- | | | | |
|---|------------------------------------|---|--|
| <input type="checkbox"/> African-American | <input type="checkbox"/> Asian | <input type="checkbox"/> Hispanic | <input type="checkbox"/> Native American |
| <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Caucasian | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> 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 syndrome (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

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