MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: July 20, 2022
Effective Date: August 23, 2022

Cystic Fibrosis and Spinal Muscular Atrophy Carrier Screen Panel, Varies

Test ID: CFSMN

Useful for:

- Reproductive risk refinement via carrier screening for individuals in the general population for cystic fibrosis and spinal muscular atrophy.
- Reproductive risk refinement via carrier screening for individuals with a family history of cystic fibrosis and/or spinal muscular atrophy when familial variants are not available
- This test is **not useful for** clinical diagnosis of an affected individual.

Genetics Information:

This test includes targeted testing to evaluate over 500 genes including the 23 cystic fibrosis transmembrane conductance regulator (*CFTR*) variants recommended by the American College of Medical Genetics and Genomics as well as targeted testing of survival motor neuron 1 (*SMN1*) and *SMN2*.

Methods:

Targeted Genotyping Array

Reference Values:

An interpretive report will be provided

Ordering Guidance:

- This test is specifically for carrier screening purposes and is not intended for diagnostic purposes. For diagnostic testing, order CFMP / Cystic Fibrosis, *CFTR* Gene, Variant Panel, Varies.
- If the reproductive partner is also having this test performed, call the lab for a revised risk assessment.
- Targeted testing for familial variants (also called site-specific or known mutation testing) is available for all genes on this panel under FMTT / Familial Mutation, Targeted Testing, Varies. Call 800-533-1710 to obtain more information about this testing option.

Necessary Information:

If there is a family history of cystic fibrosis (CF) or spinal muscular atrophy (SMA), the known genetic variant in the family should be supplied for best interpretation of results.

Specimen Requirements:

Specimen Type:	Whole blood
Patient Preparation:	A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. **Do not** aliquot.
Additional Information:	To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.
Minimum Volume:	1 mL

Note:

Specimen preferred to arrive within 96 hours of collection**.**

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Cautions:

- A negative result does not eliminate the risk of carrier status for any of the included conditions, due to the possibility that the patient carries a variant that is not interrogated with this assay or the rare chance of a false-negative result for a tested variant. For tested variants, the negative predictive value of this screen is greater than 98%. The patient's residual risk to be a carrier after a negative screen is dependent on ethnic background and family history.
- A positive control was not available for all variants targeted on this panel. For more information regarding availability of a positive control for each variant see Targeted Variants Detected by Focused Carrier Screening Tests. The negative predictive value of these targets is unknown.
- Rare variants (ie, polymorphisms) exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(4) This assay was designed to specifically target known pathogenic or likely pathogenic variants. In rare cases, DNA variants of undetermined significance may be identified. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

- Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.
- An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects deidentified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

CPT Code:
81220
81329
81222

Day(s) Performed: Thursday & Sunday

Report Available: 14 to 42 days

Questions

Contact Michelle Raths, Laboratory Technologist Resource Coordinator at 800-533-1710.