To: Medical Staff, House Staff, Patient Care Centers, and Outpatient Clinics

From: Daniela del Gaudio, PhD, FACMG, Clinical Molecular Genetics Laboratory Director
Soma Das, PhD, FACMG, Clinical Molecular Genetics Laboratory Director

Date: 7/1/2025

Subject: Carrier Screening for Spinal Muscular Atrophy, Cystic Fibrosis and Fragile X Syndrome; Diagnostic Testing for Cystic Fibrosis and Fragile X Syndrome

---

Effective Date: 7/1/2025

Laboratory Section: Molecular Diagnostics

Description/Summary:

Effective July 1, 2025, UCM Molecular Diagnostic Laboratory will start offering in-house testing for the following tests.

Spinal muscular atrophy (SMA), cystic fibrosis, and Fragile X Syndrome carrier testing: These tests are recommended for individuals planning a pregnancy or currently pregnant, especially those with relevant family history, ethnicity-based risk factors, or as part of a reproductive risk assessment.

Diagnostic cystic fibrosis testing is appropriate for individuals suspected to have cystic fibrosis. Diagnostic Fragile X testing is appropriate for individuals with neurodevelopmental disorders, intellectual disability, or autism spectrum disorder.

The in-house tests will replace the current tests that are sent to Mayo and LabCorp (Inheritest CF/SMA Panel only).

	Carrier Testing			**Diagnostic Testing**	
Test Name	Spinal Muscular Atrophy (SMA) Carrier Testing, Deletion/Duplication analysis	Cystic Fibrosis (CFTR) Carrier Screening	Fragile X (FMR1) Carrier Testing	Cystic Fibrosis (CFTR) Diagnostic Testing	Fragile X (FMR1) Diagnostic Testing
Test Code	LABMDSMA	LABMDCFCAR	LABMDFRAGXCAR	LABMDCFTRMUT	LABMDFRAGX
CPT Code(s)	81329	81220	81243	81222, 81223	81243, 81244*
Turnaround Time	10 days	14 days	10 days	28 days	10 days
Specimen Requirements	3mL whole blood, EDTA	3mL whole blood, EDTA	3mL whole blood, EDTA	3mL whole blood, EDTA	3mL whole blood, EDTA
Methodology	MLPA^	Next-generation sequencing	PCR and fragment analysis	Next-generation sequencing	PCR and fragment analysis

* CPT code 81224 is only billed if the methylation portion of the assay is performed
^ Multiplex ligation-dependent probe amplification

Questions: If there are any questions regarding the change, test requirements or ordering, please contact:

Daniela del Gaudio, Molecular Genetics Laboratory Director, ddelgaudio@bsd.uchicago.edu
Kelly Arndt, Clinical Genomics Laboratory Manager, Kelly.Arndt@uchicagomedicine.org