One Visit. One Blood Draw. Many Answers.

Carrier screening can be performed on only two tubes of blood. The chart below depicts which specimens are acceptable for analysis for each common genetic disease.

Detailed specimen requirements for CF, SMA, and fragile X syndrome are readily available at www.labcorp.com.

Specimen Options

Test	CF*plus* Testing	Spinal Muscular Atrophy Testing	Fragile X Syndrome Testing
Blood	✔	✔	✔
Mouthwash	✔		

To learn more about CF, SMA and fragile X syndrome testing, please visit:

www.mytestingoptions.com
www.labcorp.com

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Prenatal Genetic Screening Visit

CF, SMA and Fragile X Syndrome

Integrated GENETICS
LabCorp Specialty Testing Group

Your Partner for Genetic Testing

One visit can lead to many answers with Integrated Genetics' carrier screening for CF, SMA and fragile X syndrome.

Prenatal Genetic Screening Visit Testing Includes:

CF, SMA and Fragile X Syndrome

One visit. Many answers.

Carrier Testing

	Cystic Fibrosis (CF) The most common inherited disease of children and young adults.	**Spinal Muscular Atrophy (SMA)** The most common inherited cause of early childhood death.	**Fragile X Syndrome** The most common inherited cause of mental retardation.
Carrier Frequency	Ranges from 1 in 24 to 1 in 94, varies by ethnicity[1]	Ranges from 1 in 47 to 1 in 72 in the U.S., varies by ethnicity[7]	1 in 260 females[9]
Incidence	1 in 3,500[2]	1 in 11,000[7]	1 in 4,000 males, 1 in 8,000 females[10]
Inheritance	Autosomal recessive	Autosomal recessive	X-linked
Clinical Characteristics	■ Disorder of mucus production ■ CF causes the body to produce abnormally thick mucus, leading to life threatening lung infections, digestion problems, diarrhea, poor growth and infertility ■ Symptoms of the disease range from mild to severe ■ CF does not affect intelligence	■ Progressive degeneration of lower motor neurons ■ Muscle weakness and, in the most common type, respiratory failure by age two ■ Muscles responsible for crawling, walking, swallowing and head and neck control are the most severely affected ■ Variability of severity and age of onset ■ SMA does not affect intelligence	■ Mild learning disabilities to severe mental retardation ■ Approximately one-third of all children diagnosed with fragile X syndrome also have autism and hyperactivity ■ Almost all males with full mutations have developmental delay/mental retardation ■ ~50% of females with a full mutation have IQs in the borderline or mentally retarded range; of the remaining 50%, half have learning disabilities
Typical Lifespan	37 years[1]	Less than 2 years for the most common type.[8]	Normal
Test Detection Rate	**Carrier detection rate with CF*plus*® 97 mutation panel:**[3-6] Caucasian: 93% Ashkenazi Jewish: 97% African American: 81% Hispanic: 78% *The Cystic Fibrosis Profile, DNA Analysis (32 mutation analysis) is also available.*	**Carrier detection rate:**[7] Caucasian: 95% Ashkenazi Jewish: 91% African American: 71% Hispanic: 90% Asian: 93% Asian Indian: 90%	**Carrier detection rate:** 99%[11]
Recommended Follow-Up for a Positive Result	Test partner	Test partner	Offer genetic counseling and prenatal diagnosis
Timing of Testing	■ Preconception ■ First and second trimester of pregnancy ■ Need carrier testing only once	■ Preconception ■ First and second trimester of pregnancy ■ Need carrier testing only once	■ Preconception ■ First and second trimester of pregnancy ■ Need carrier testing only once

Please Note: This chart includes testing that pertains to all racial and ethnic groups. Additional ethnic-specific tests (e.g., Ashkenazi Jewish carrier testing and hemoglobinopathy screening) should be ordered as appropriate.
Carrier frequencies and incidences are approximate.