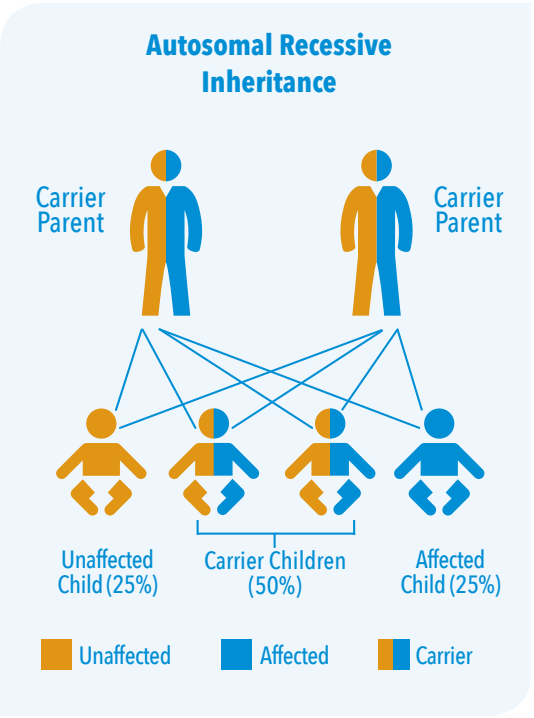
PREMIER GENOMICS

CYSTIC FIBROSIS GENETIC CARRIER SCREENING

1 in 38 Americans are symptomless carriers of cystic fibrosis–an inherited, life-threatening condition affecting the lungs, pancreas, and other organs.[1,2]

Cystic fibrosis (CF) is the most common life-threatening autosomal recessive condition in the non-Hispanic white population. It is a progressive, multisystem disease that primarily affects the pulmonary, pancreatic, and gastrointestinal systems. CF is caused by mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. When both parents are carriers of a mutation in this gene, they have a 1 in 4 chance of having a child with CF.[2]

Characteristics of Cystic Fibrosis[2]	
	• Coughing • Wheezing • Breathing difficulty • Persistent lung infections
	• Loose stools • Abdominal pain • Slow growth • Failure to thrive
	• Male infertility
	• Individuals with CF have a life expectancy of approximately 37 years, and the cause of death is typically lung damage • 15% of individuals with CF have a mild form of the disease and live an average of 56 years
Interventions, such as antibiotics for lung infections, may improve outcomes.[3]	



ACOG and ACMG recommend offering CF carrier screening to all patients, regardless of ethnicity.[2,4]

Screening results inform patients of their carrier status and can help guide decision-making for patients who are pregnant, planning a pregnancy, or considering fertility options.

32- and 97-mutation panels are available that detect the most common variations of the *CFTR* gene. Each panel screens for the 23 most common mutations recommended by both the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG), as well as additional multiethnic mutations.

		32 Mutations		97 Mutations	
Ethnicity	**Carrier frequency[1]**	**Detection rate**	**Residual risk**	**Detection rate**	**Residual risk**
Non-Hispanic Caucasian	1 in 25	90%	1 in 240	93%	1 in 343
Jewish (Ashkenazi)	1 in 24	97%	1 in 800	97%	1 in 767
Hispanic	1 in 58	73%	1 in 167	78%	1 in 260
African American	1 in 61	69%	1 in 207	81%	1 in 316
Asian	1 in 94	55%	1 in 183	55%	1 in 183

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CYSTIC FIBROSIS GENETIC TESTING PROCESS

THE PROCESS	The DNA collection procedure is simple and requires only an in-office buccal (cheek) swab with our sample collection kit. UPS will pick up your patient samples and deliver them directly to our lab (labels and instructions are included in each kit). Dedicated account services are always available to answer questions and manage requests.
THE TURN-AROUND TIME	7 to 10 days
THE REPORT	Upon completion of DNA extraction and evaluation at our advanced laboratory facility, a comprehensive report is generated and uploaded to a secure portal with dedicated physician log-in and downloading capabilities. Physician-to-physician consultation is also available with our Medical Director or our Genetic Counselor. Monograph is available upon request.

CONTACT YOUR REPRESENTATIVE OR REACH US DIRECTLY AT OUR EMAIL ADDRESS BELOW TO OBTAIN SAMPLE TEST REPORTS AND DNA SAMPLE COLLECTION KITS FOR THIS OR ANY OF OUR OTHER ADVANCED GENETIC TESTING PANELS.

Jewish Genetic Diseases Carrier Screening	*BRCA1/2* Genetic Testing
Fragile X Syndrome Genetic Screening	Hereditary Colon Cancer/Lynch Syndrome Genetic Testing
Cystic Fibrosis Genetic Carrier Screening	Pharmacogenomics Testing

ABOUT PREMIER GENOMICS

Premier Genomics is committed to advancing the field of personalized genetic medicine by offering cutting-edge genetic screening services to help practitioners and their patients in pursuit of tailored treatment and optimized, personalized health care. We work together with patients and their insurance providers to help ensure that access to these important genetic tests does not cause patients financial hardship.

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CLIA-accredited, CAP-certified clinical laboratory

References: 1. Rohlfs EM, Zhou Z, Heim RA, et al. Cystic fibrosis carrier testing in an ethnically diverse US population. *Clin Chem.* 2011;57(6):841-848. **2.** American Congress (formerly College) of Obstetricians and Gynecologists (ACOG) Committee on Genetics. ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis. *Obstet Gynecol.* 2011;117(4):1028-1031. **3.** Cutting GR. Cystic fibrosis genetics: from molecular understanding to clinical application. *Nat Rev Genet.* 2015;16(1):45-56. **4.** Watson MS, Cutting GR, Desnick RJ, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel [published corrections appear in *Genet Med.* 2004;6(6):548 and *Genet Med.* 2005;7(4):286]. *Genet Med.* 2004;6(5):387-391.