

Cystic Fibrosis: *CFTR* Common Mutation Panel

Test Code: CF

Turnaround time: 2 weeks

CPT Codes: 81220 x1

Condition Description

Cystic fibrosis (CF) is a chronic genetic condition involving multiple organ systems. Classical CF primarily involves the respiratory and digestive systems, and may have a range of clinical severity. Pulmonary symptoms often include lower airway inflammation, chronic cough, chronic sinusitis, and recurrent infections. Digestive symptoms often include meconium ileus, pancreatic insufficiency resulting in malabsorption and/or failure to thrive, diabetes mellitus, and hepatobiliary disease. Congenital bilateral absence of the vas deferens (CBAVD) is seen in men without pulmonary or digestive symptoms of CF, and results in azoospermia [1]. CBAVD is a significant cause of male infertility.

CF is caused by mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. Individuals with mutations in the *CFTR* gene may also present with milder or atypical symptoms such as pancreatitis or chronic sinusitis.

The incidence of CF is approximately 1 in 2500 live births among Caucasians and is inherited in an autosomal recessive pattern. The carrier frequency is estimated to be approximately 1 in 25 in the Caucasian population, 1 in 24 in the Ashkenazi Jewish population, 1 in 61 in the African American population, 1 in 58 in the Hispanic population, and 1 in 94 in the Asian population [2].

The current recommendation from the American College of Obstetrics and Gynecologists [2,3] and the American College of Medical Genetics Subcommittee on Cystic Fibrosis [4] is that screening for cystic fibrosis should be offered to all patients, regardless of ethnicity.

This panel tests for the 39 most common *CFTR* mutations (listed below), including the core panel of 23 mutations for cystic fibrosis as recommended by the American College of Medical Genetics in 2004 [4].

<i>DeltaF508</i>	<i>Delta507</i>	<i>R117H</i>	<i>W1282X</i>	<i>3120+1G>A</i>	<i>G85E</i>	<i>Y122X</i>	<i>R334W</i>
<i>R347P</i>	<i>R347H</i>	<i>A455E</i>	<i>V520F</i>	<i>G542X</i>	<i>S549R</i>	<i>S549N</i>	<i>A559T</i>
<i>G551D</i>	<i>R553X</i>	<i>R560T</i>	<i>Y1092X</i>	<i>M1101K</i>	<i>R1162X</i>	<i>S1255X</i>	<i>N1303K</i>
<i>394delIT</i>	<i>621+1G>T</i>	<i>711+1G>T</i>	<i>1078delT</i>	<i>1717-1G>A</i>	<i>3876delA</i>	<i>1898+1G>A</i>	<i>1898+5G>T</i>
<i>2183AA>G</i>	<i>2184delA</i>	<i>3905insT</i>	<i>2307insA</i>	<i>2789+5G>A</i>	<i>3659delC</i>	<i>3849+10kbC>T</i>	

Reported results describe the presence or absence of mutations tested and, if appropriate, provides calculated residual carrier risk, and risks to future pregnancies.

[Click here](#) for the GeneReviews summary on this condition.

Visit www.ThinkGenetic.com for patient-friendly information on [cystic fibrosis](#).

References:

- Xu et al. Cystic fibrosis transmembrane conductance regulator is vital to sperm fertilizing capacity and male fertility (2007). PNAS 104(23):9816-21.
- ACOG Committee Opinion. Update on Carrier Screening for Cystic Fibrosis (2011). Obstet Gynecol Apr; 117(4): 1029-31.
- ACOG Committee Opinion. Update on Carrier Screening for Cystic Fibrosis (2005). Obstet Gynecol 106:1465-8.
- Watson, M. et al. Cystic Fibrosis population carrier screening: 2004 revision of the American College of Medical Genetics mutation panel (2004). Genet Med 6(5):387-91.

Genes

[CFTR](#)

Indications

Testing is indicated for:

- Individuals with a diagnosis of CF, or atypical presentations of CF (chronic pancreatitis, sinusitis).
- Males with congenital bilateral absence of the vas deferens (CBAVD).
- Carrier screening for individuals of Caucasian or Ashkenazi Jewish background
- Family members of an affected individual at risk to be carriers of CF.

Methodology

This panel of 39 mutations in the *CFTR* gene is tested for by an allele-specific primer extension assay and includes the recommended ACMG panel of 23 common mutations.

Specimen Requirements

Submit only 1 of the following specimen types

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Related Tests

- Sequencing and deletion/duplication analysis of the *CFTR* gene is available for individuals who test negative with the common mutation panel, when other mutations are suspected.
- *CFTR* poly T analysis is performed as a reflex test when an R117H mutation is detected or when evaluating males with CBAVD.
- Ashkenazi Jewish Carrier Panel is available to screen for the panel of 9 autosomal recessive conditions common in individuals of Ashkenazi Jewish background.
- Prenatal testing is available to couples who are confirmed carriers of gene mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.