

Using Genetic Testing for Diagnosis of Cystic Fibrosis (CF)

Indications:

- Clinical diagnosis of CF
- Meconium ileus in infants
- Positive finding in IRT newborn screening assay
- High risk of CF in fetus due to carrier status of parents
- Finding of echogenic bowel in fetus
- Family history of CF or know familial *CFTR* mutation
- No mutations or only one heterozygous *CFTR* mutation detected during targeted mutation analysis of patients with a clinical diagnosis of CF

Benefits:

- CFTR* sequencing
- Can confirm a clinical diagnosis of CF and identify the familial mutation(s)
 - Can confirm a positive IRT newborn screening test
 - May, in some cases, inform the prognosis based on the nature of the familial *CFTR* mutation(s)
 - Can inform genetic counseling by clarifying the carrier state in blood relatives of individuals affected with CF or known to carry a *CFTR* mutation

Background:

- Cystic Fibrosis (CF) is a recessively inherited multisystem disease affecting the respiratory, digestive, and male reproductive systems.^{1,2}
- Morbidity and mortality in CF are largely due to pulmonary complications resulting from chronic lower airway infection and inflammation.^{1,2}
- More than 95% of males with CF are infertile due to agenesis of Wolffian duct structures, and the great majority of CF patients suffer from pancreatic insufficiency.^{1,2}
- Early diagnosis of CF can help to prevent failure to thrive in infants and children through pancreatic enzyme replacement and chronic bacterial airway infection through antibiotic prophylaxis.^{1,2}
- Pancreatic sufficiency is often associated with a milder form of disease (non-classic CF) or, in males, isolated infertility without pulmonary or gastrointestinal manifestations (Congenital bilateral absence of the vas deferens, CBAVD).^{1,2}
- CF is caused by mutations in the *CFTR* gene.^{1,2}
- Certain *CFTR* mutations are known to be associated with non-classic CF or, in males, CBAVD.^{1,2}

References

1. Moskowitz SM, Chmiel JF, Stern DL, et al. Clinical practice and genetic counseling for cystic fibrosis and *CFTR*-related disorders. *Genet Med*. 2008;10:851-868.
2. Moskowitz SM, Chmiel JF, Stern DL, Cheng E, Cutting GR. (Updated March 26, 2001.) *CFTR*-related disorders. In: *GeneReviews* at GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2010. Available at <http://www.genetests.org>. Accessed February 21, 2010.

Ordering Information: Please see other side.

Ordering Information for Cystic Fibrosis (CF) Testing

Indications for Testing

- Clinical diagnosis of CF
- Meconium ileus in infants
- Positive finding in IRT newborn screening assay
- High risk of CF in fetus due to carrier status of parents
- Finding of echogenic bowel in fetus
- Family history of CF or know familial *CFTR* mutation
- No mutations or only one heterozygous *CFTR* mutation detected during targeted mutation analysis of patients with a clinical diagnosis of CF

Ordering Information

Gene(s)	Test Code
<i>CFTR</i>	160101

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.