



# CYSTIC FIBROSIS (CF) TESTING

ordering recommendations

## Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants (2013661)

## Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants, Fetal (2013662)

0050608—Maternal sample is recommended to rule out maternal cell contamination.

## Cystic Fibrosis (*CFTR*) Sequencing (0051110)

## Cystic Fibrosis (*CFTR*), 165 Variants with Reflex to Sequencing (2013663)

## Cystic Fibrosis (*CFTR*) Sequencing with Reflex to Deletion/Duplication (0051640)

### Tests for 165 pathogenic variants.

Order for the following indications:

- Carrier testing in a healthy patient
- Patient has symptoms of CF
- Patient has family history of CF

### Tests amniocytes for 165 pathogenic variants.

Order for fetal samples with the following indications:

- Both parents have *CFTR* variants which are known to be included on the panel
- Echogenic or cystic dilatation of the fetal bowel

### Detects 97 percent of the more than 2,000 described *CFTR* gene variants.

Order for patients with symptoms of classic CF or a *CFTR*-related disorder without two variants identified by the CF 165 pathogenic variants test.

### Tests for 165 pathogenic variants. If two *CFTR* variants are not identified, sequencing is performed to detect 97 percent of the more than 2,000 described *CFTR* variants.

Order for patients with the following indications if not previously tested using the CF 165 pathogenic variants test:

- Patients with symptoms of classic CF
- Patients with a positive sweat chloride
- Patients with symptoms of a *CFTR*-related disorder

### Tests for more than 2,000 *CFTR* gene variants. If two pathogenic *CFTR* variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.

Order for patients with symptoms of classic CF or a *CFTR*-related disorder without two variants identified by the CF 165 pathogenic variants test.



**Cystic Fibrosis (CFTR)  
Deletion/Duplication  
(0051642)**

**Tests for large deletions or duplications in the CFTR gene, accounting for 1–2 percent of variants.**

Order for the following patients:

- Symptoms of classic CF or a CFTR-related disorder without two identified pathogenic variants following CFTR sequencing.
- To determine carrier status in individuals who have a family member with a large CFTR gene deletion.

**Cystic Fibrosis (CFTR), 165  
Variants with Reflex to  
Sequencing and Reflex to  
Deletion/Duplication  
(2013664)**

**Tests for 165 pathogenic CFTR pathogenic variants. If two pathogenic CFTR variants are not identified, sequencing is performed. If two CFTR variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.**

Order for patients with a clinical diagnosis of CF as a cost-effective method for determining the causative variants present.



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