CYSTIC FIBROSIS (CF) TESTING

ordering recommendations

Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants (2013661)	**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
Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants, Fetal (2013662) 0050608—Maternal sample is recommended to exclude maternal cell contamination.	**Tests cultured 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
Cystic Fibrosis (*CFTR*) Sequencing (0051110)	**Detects 97% 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.
Cystic Fibrosis (*CFTR*), 165 Variants with Reflex to Sequencing (2013663)	**Tests for 165 pathogenic variants. If two *CFTR* variants are not identified, sequencing is performed to detect 97% 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 test • Patients with symptoms of a *CFTR*-related disorder
Cystic Fibrosis *(CFTR)* Sequencing with Reflex to Deletion/Duplication (0051640)	**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%.** 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*), 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%.** Order for patients with a clinical diagnosis of CF as a cost-effective method for determining the causative variants present.

ARUP LABORATORIES

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