**To:** Dartmouth Health providers involved in genetic testing for cystic fibrosis (CF)

**From:** Wahab A. Khan, PhD, FACMG

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**Date:** October 31, 2023

**RE:** TruSight Cystic Fibrosis testing by next generation sequencing (NGS)

Effective immediately, the CGAT (Clinical Genomics and Advanced Technology) Laboratory will be analyzing additional DNA sequence variants in the *CFTR* gene that are not included in our current analysis for cystic fibrosis (CF) testing. These additional CF-causing variants are included to align with a recent update to the minimum set recommended for testing by the American College of Medical Genetics and Genomics (see PMID: 37310422 position statement). Our new test will analyze a total of 175 sequence variants in the *CFTR* gene, including the minimum set of 100 variants now recommended.

This updated test for carrier screening or diagnosis of CF will be ordered and reported in the same way as previous cystic fibrosis testing by NGS. Specimen collection details are provided below:

* Specimen Type: Whole blood
* Collection tube: Lavender top EDTA vacutainer
* Volume: 3 mL (minimum: 1 mL)
* Storage/Stability: Refrigerated @ 4oC for 5 days
* Transport: Refrigerated
* Maximum turnaround time: 12 days
* eD-H order code: LAB3390

**For additional information, please contact the CGAT-Molecular lab 603-650-8257 or email CGAT\_NGS\_Group@hitchcock.org with questions or concerns.**