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Cystic Fibrosis (CFTR Gene) Mutation Analysis

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| Test Ordering Code: | 1031 |
| CPT Codes: | 83891, 83900, 83901x13, 83914x33, 83909, 83912 |
| Specimen: | Whole blood anticoagulated with EDTA, 1mL minimum. Maintain specimen at 4° C after collection and ship at room temperature. |
| Turnaround Time: | 5-7 business days |
| Clinical Significance: | Cystic Fibrosis (CF) is the most common autosomal recessive inherited disorder affecting children and young adults which is characterized by pulmonary disease, deficient exocrine pancreatic function, and complications due to the presence of abnormally thick mucus in many organs including the small intestine, liver, and reproductive tract. |
| Indications for Testing: | Carrier Screening Diagnosis of Cystic Fibrosis |
| Methodology: | This test uses multiplex PCR to amplify 15 segments of the CFTR gene that contain the 32 mutation sites being analyzed. The presence or absence of the mutation is determined by an oligo ligation assay, where the ligation products are then analyzed by electrophoresis. Analyzed mutations include: $\Delta F508$, $\Delta I507$, V520F, 1717-1G→A, G542X, G551D, R553X, R560T, S549R, S549N, 3849+10kbC→T, R117H, 711+1G→T, 1078 ΔT , R347P, R347H, R334W, A455E, R1162X, 3659 ΔC , W1282X, 3905insT, N1303K, G85E, 394 ΔTT , 621+1G→T, 2183AA→G, 1898+1G→A, 2184 ΔA , 2789+5G→A, 3120+1G→A, 3876 ΔA . Reflex testing for 5T/7T/9T is performed when R117H/C is detected and reflex testing is also carried out for I506V, I507V when either $\Delta I507$ or $\Delta F508$ mutation present. |
| Reporting of Results: | Negative: The patient does not have any of the mutations tested. Positive: A list of found mutations |
| Intended Use: | This test only detects the 33 core mutations and two reflex test mutations listed in the methodology section, and not other mutations in the CFTR gene. |
| References: | Preconception and Prenatal Carrier Screening for Cystic Fibrosis. Clinical and Laboratory Guidelines. The American College of Obstetricians and Gynecologists. Oct. 2001 |