Cystic Fibrosis, DNA Analysis

**TEST NAME**
- **Cystic Fibrosis, DNA Analysis**

**METHODOLOGY**
- Multiplex PCR

**LOINC CODE**
- 38404-0

**SPECIMEN REQUIREMENTS**

<table>
<thead>
<tr>
<th>Specimen</th>
<th>Specimen Volume (min)</th>
<th>Specimen Type</th>
<th>Specimen Container</th>
<th>Transport Environment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Preferred</td>
<td>5 mL (3 mL)</td>
<td>Whole Blood</td>
<td>EDTA (Lavender Top)</td>
<td>Room Temperature</td>
</tr>
</tbody>
</table>

**Instructions**
- To receive a complete personalized report based on results, patient demographics and clinical scenario, please completely fill out the Cystic Fibrosis Patient Information Form and include with specimen. This form can be downloaded from the following link: [Cystic Fibrosis Patient Information Form](#)

- Stability: Room Temperature 8 Days, Refrigerated 8 days, Frozen Not Acceptable. Do not centrifuge.

- Specimen cannot be shared with other testing for risk of DNA contamination.

**GENERAL INFORMATION**

**Testing Schedule**
- Wednesday

**Expected TAT**
- Within 14 days

**Clinical Use**
- This is a qualitative genotyping test that provides information intended to be used for carrier testing in adults of reproductive age, as an aid in newborn screening, and in confirmatory diagnostic testing in newborns and children. This test is not indicated for use in fetal diagnostic or pre-implantation testing. This test is not intended for stand-alone diagnostic purposes. Personalized reports include risk assessment, concise genotype results, and clinical relevance. Further assessment is recommended when appropriate. Genetic Counseling is available through Access Genetics.

- Method: Genomic DNA is evaluated using the Luminex xTAG Cystic Fibrosis 60 kit, an FDA-approved device employing a multiplex polymerase chain reaction (PCR) using oligonucleotide primers specific for regions of the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. The resultant data is analyzed for 60 mutations and variants including the 23 mutations recommended by the American College of Medical Genetics and American College of Obstetricians and Gynecologists (ACMG/ACOG) for CF carrier testing. Reflex analysis is performed as recommended for intron 8-5T/7T/9T, I506V, I507V, and F508C variants. Results are interpreted by Board Certified Molecular Geneticists.

**CPT Code(s)**
- 81220; If reflex performed add 81224

**Lab Section**
- Reference Lab