



OHSU Molecular Diagnostic Center
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CYSTIC FIBROSIS **Molecular Diagnostic Center**

Cystic Fibrosis (CF) is the most common autosomal recessive genetic disease in the Caucasian population and appears in approximately one in every 2,500 newborns. In the Caucasian and Ashkenazi Jewish populations, about one in every 25 individuals carries one copy of a mutation in the CFTR gene at 7q31.2.

REASONS FOR REFERRAL

- Confirmation of clinical diagnosis
- Carrier identification in persons with a positive or negative family history
- Sperm & egg donors
- Abnormal fetal ultrasound
- Preconception and carrier screening

TESTING METHODOLOGY

Direct Mutation Analysis: Our laboratory uses the oligonucleotide ligation (OLA) methodology to test for the ACMG-recommended mutation panel for carrier screening which include:

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|---------|----------|----------------|-------------|-------------|----------|-------------|
| • ΔF508 | • R553X | • R117H | • 1078ΔT | • 1898+1G | • R1162X | • 711+1G>T |
| • ΔI507 | • G542X | • 621+1G→T | • R334W | • 2184delA | • 3659ΔC | • 2789+5G>A |
| • G551D | • N1303K | • I148T | • R347P | • 1717-1G→A | • W1282X | • A455E |
| • R560T | • G85E | • 3849+10kbC→T | • 3120+1G→A | | | |

Additional mutations tested include: **S549N, S549R, V520F, 3876delA, 394delTT, R347H, 3905insT, F508C(SNP)**, for a total of 33 mutations.

Reflex testing for 5T/7T/9T is performed for R117H carriers. Reflex testing for I506V and I507V is performed only when indicated.

Mutation Analysis

- 90% detection in the Caucasian/non-Ashkenazic population
- 97% detection in the Ashkenazic population
- 69% detection in the African-American population
- 57% detection in the Hispanic-American population

Test reporting follows the ACMG guidelines.

SPECIMEN REQUIREMENT

Blood: ACD (solution A or B):
Adult: 5 mL
Child: 5 mL
Infant: 2-3 mL

Requisition form must accompany specimen including ethnicity, clinical and family history information. Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact our laboratory.

TURNAROUND TIME: 2 weeks

CPT CODES: 83890, 83896x25, 83894, 83901, 83912

Cfinfo 5/04