



One-Day Cystic Fibrosis DNA Test

Mutations Detected (33):

ACMG/ACOG Recommended Mutation Panel (25):

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

Plus 8 additional mutations:

3905insT	2183AA→G
S1251N	E60X
Q552X	2143delT
394delTT	711+5G→A

- 5T/7T/9T typing is performed on a reflex basis. If desired, this typing can be performed on any sample at no additional cost.
- 3199del6 typing is performed on a reflex basis on all I148T positive patients.
- With our test methodology, the ΔF508 probe is specific and will not give a false positive result with the polymorphisms F508C, I506V, or I507V.

Detection Rates:

<u>Ethnic Group</u>	<u>Detection Rate</u>
European Caucasian	90%
Ashkenazi Jewish	97%
African American	69%
Hispanic	72%
Asian	Not determined

Sample Requirements:

- Cheek cells or 5 ml of blood in an EDTA (lavender) tube (collection kits provided at no charge)
- Send at room temperature; shipping provided

Turnaround Time:

- Cheek cells and blood: 1 business day
- Prenatal sample: 3 business days (direct or cultured)

Prenatal Testing:

- Provided on both direct and cultured cells, amnio or CVS
- Maternal cell contamination studies performed at no charge when cystic fibrosis DNA testing is being performed on a prenatal sample and a maternal blood cell sample is provided

Please call Kimball Genetics for more information.

