

## CYSTIC FIBROSIS CARRIER SCREENING

List of the 60 CFTR gene mutations and 4 variants (benign polymorphisms) included in the test panel:

ΔF508*	1717-1G>A*	W1282X*	2307insA
ΔI507*	R560T*	1078delT	Y1092X
G542X*	R553X*	394delTT	M1101K
G85E*	G551D*	Y122X	S1255X
R117H*	1898+1G>A*	R347H	3876delA
621+1G>T*	2184delA*	V520F	3905insT
711+1G>T*	2789+5G>A*	A559T	<i>5T/7T/9T</i>
N1303K*	3120+1G>A*	S549N	<i>F508C</i>
R334W*	R1162X*	S549R	<i>I507V</i>
R347P*	3659delC*	1898+5G>T	<i>I506V</i>
A455E*	3849+10kbC>T*	2183AA>G	S1196X
CFTR dele2, 3	L206W	1677delTA	3199del6
E60X	935delA	2055del9>A	R1066C
R75X	G330X	2143delT	W1089X
406-1G>A	Q493X	K710X	D1152H
G178R	Q890X	R1158X	3791delC

\*asterisk denotes ACMG/ACOG panel  
benign polymorphisms variants *italicized*