

Cystic Fibrosis 139-Variant Assay

The assay tests for: 134 CF-causing variants; one ACMG recommended panel variant (R117H, classified as a Mutation of Varying Clinical Consequence, MVCC, by CFTR2); one conditionally reported modifying variant (PolyTG/PolyT); and three conditionally reported benign variants (I506V, I507V, F508C)¹⁶; for a total of 139 reported variants.

The 134 CF-causing variants correspond to 129 CF-causing variants in the CFTR2 database. The CFTR2 database includes five CF-causing variants for which the same protein level change can arise from two distinct nucleotide changes [e.g., S466X(C>A) and S466X(C>G)]. These five variants are listed according to the amino acid codon in the CFTR2 database (e.g., S466X) while the assay reports each individual variant [e.g., S466X(C>A) and S466X(C>G)]. The list of 139 variants reported by the Cystic Fibrosis 139-Variant Assay is provided below.

Cystic Fibrosis 139-Variant Assay Summary of Variants

M1V**	T338I**	Q552X	3121-1G>A**
CFTR dele2,3	1154insTC	R553X	3272-26A>G
Q39X**	S341P**	A559T	L1065P**
E60X	R347H	R560T	R1066C
P67L	R347P	R560K**	R1066H
R75X	R352Q	1811+1.6kb A>G**	L1077P**
G85E	1213delT**	1812-1 G>A	W1089X
394delTT	1248+1G>A**	E585X**	Y1092X(C>A)
405+1 G>A**	1259insA**	1898+1G>A	Y1092X(C>G)**
406-1G>A	W401X (c.1202G>A)**	1898+3A>G**	M1101K
E92X	W401X (c.1203G>A)**	2143delT	E1104X**
E92K**	1341+1G>A**	R709X	R1158X
Q98X**	<i>PolyTG/PolyT</i>	K710X	R1162X
457TAT>G**	1461ins4**	2183delAA>G	3659delC
D110H	A455E	2184insA	S1196X
R117C	1525-1G>A**	2184delA	W1204X (c.3611G>A)**
R117H	S466X (C>A)**	2307insA	W1204X (c.3612G>A)**
Y122X	S466X (C>G)	L732X**	3791delC
574delA**	L467P**	2347delG**	3849+10kbC>T
621+1G>T	1548delG†	R764X	G1244E**
663delT	S489X**	2585delT**	3876delA
G178R	S492F**	E822X**	S1251N
711+1G>T	Q493X	2622+1G>A**	3905insT
711+3A>G**	I507del	E831X**	W1282X
711+5 G>A**	F508del	W846X	4005+1G>A**
712-1 G>T**	1677delTA	R851X**	N1303K
H199Y**	V520F	2711delT**	4016insT**
P205S	Q525X**†	2789+5G>A	Q1313X**
L206W	1717-8G>A	Q890X	4209TGTT>AA**
Q220X**	1717-1G>A	L927P**	CFTRdele22,23
852del22**	G542X	S945L**	4382delA**
1078delT	S549R (c.1645A>C)	3007delG**	<i>I506V</i>
G330X	S549R (c.1647T>G)	G970R**	<i>I507V</i>
R334W	S549N	3120G>A	<i>F508C</i>
I336K	G551D	3120+1G>A	