

FDA-cleared 139 variant panel

Variant cDNA name (ordered 5' to 3')	Variant Protein Name	Variant Legacy Name
c.1A>G	p.Met1Val	M1V
c.54-5940_273+10250del21kb	p.Ser18ArgfsX16	CFTRdele2,3
c.115C>T	p.Gln39X	Q39X
c.178G>T	p.Glu60X	E60X
c.200C>T	p.Pro67Leu	P67L
c.223C>T	p.Arg75X	R75X
c.254G>A	p.Gly85Glu	G85E
c.262_263delTT	p.Leu88IlefsX22	394delTT
c.273+1G>A		405+1G>A
c.274-1G>A		406-1G>A
c.274G>A	p.Glu92Lys	E92K
c.274G>T	p.Glu92X	E92X
c.292C>T	p.Gln98X	Q98X
c.325_327delTATinsG	p.Tyr109GlyfsX4	457TAT>G
c.328G>C	p.Asp110His	D110H
c.349C>T	p.Arg117Cys	R117C
c.350G>A	p.Arg117His	R117H
c.366T>A	p.Tyr122X	Y122X
c.442delA	p.Ile148LeufsX5	574delA
c.489+1G>T		621+1G>T
c.531delT	p.Ile177MetfsX12	663delT
c.532G>A	p.Gly178Arg	G178R
c.579+1G>T		711+1G>T
c.579+3A>G		711+3A>G
c.579+5G>A		711+5G>A
c.580-1G>T		712-1G>T
c.595C>T	p.His199Tyr	H199Y
c.613C>T	p.Pro205Ser	P205S
c.617T>G	p.Leu206Trp	L206W
c.658C>T	p.Gln220X	Q220X
c.720_741delAGGGAGAATGATGATGAAGTAC	p.Gly241GlufsX13	852del22
c.948delT	p.Phe316LeufsX12	1078delT
c.988G>T	p.Gly330X	G330X

c.1000C>T	p.Arg334Trp	R334W
c.1007T>A	p.Ile336Lys	I336K
c.1013C>T	p.Thr338Ile	T338I
c.1021T>C	p.Ser341Pro	S341P
c.1022_1023insTC	p.Phe342HisfsX28	1154insTC
c.1040G>A	p.Arg347His	R347H
c.1040G>C	p.Arg347Pro	R347P
c.1055G>A	p.Arg352Gln	R352Q
c.1081delT	p.Trp361GlyfsX8	1213delT
c.1116+1G>A		1248+1G>A
c.1127_1128insA	p.Gln378AlafsX4	1259insA
c.1202G>A	p.Trp401X	W401X (c.1202G>A)
c.1203G>A	p.Trp401X	W401X (c.1203G>A)
c.1209+1G>A		1341+1G>A
<i>c.1210-33_1210-6GT[TT]</i>		<i>PolyTG/PolyT</i>
c.1329_1330insAGAT	p.Ile444ArgfsX3	1461ins4
c.1364C>A	p.Ala455Glu	A455E
c.1393-1G>A		1525-1G>A
c.1397C>A	p.Ser466X	S466X (C>A)
c.1397C>G	p.Ser466X	S466X (C>G)
c.1400T>C	p.Leu467Pro	L467P
c.1418delG	p.Gly473GlufsX54	1548delG
c.1466C>A	p.Ser489X	S489X
c.1475C>T	p.Ser492Phe	S492F
c.1477C>T	p.Gln493X	Q493X
<i>c.1516A>G</i>	<i>p.Ile506Val</i>	<i>I506V</i>
<i>c.1519A>G</i>	<i>p.Ile507Val</i>	<i>I507V</i>
c.1519_1521delATC	p.Ile507del	I507del
c.1521_1523delCTT	p.Phe508del	F508del
<i>c.1523T>G</i>	<i>p.Phe508Cys</i>	<i>F508C</i>
c.1545_1546delTA	p.Tyr515X	1677delTA
c.1558G>T	p.Val520Phe	V520F
c.1573C>T	p.Gln525X	Q525X
c.1585-1G>A		1717-1G>A
c.1585-8G>A		1717-8G>A
c.1624G>T	p.Gly542X	G542X

c.1645A>C	p.Ser549Arg	S549R (c.1645A>C)
c.1646G>A	p.Ser549Asn	S549N
c.1647T>G	p.Ser549Arg	S549R (c.1647T>G)
c.1652G>A	p.Gly551Asp	G551D
c.1654C>T	p.Gln552X	Q552X
c.1657C>T	p.Arg553X	R553X
c.1675G>A	p.Ala559Thr	A559T
c.1679+1.6kbA>G		1811+1.6kbA>G
c.1679G>A	p.Arg560Lys	R560K
c.1679G>C	p.Arg560Thr	R560T
c.1680-1G>A		1812-1G>A
c.1753G>T	p.Glu585X	E585X
c.1766+1G>A		1898+1G>A
c.1766+3A>G		1898+3A>G
c.2012delT	p.Leu671X	2143delT
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA>G
c.2052_2053insA	p.Gln685ThrfsX4	2184insA
c.2052delA	p.Lys684AsnfsX38	2184delA
c.2125C>T	p.Arg709X	R709X
c.2128A>T	p.Lys710X	K710X
c.2175_2176insA	p.Glu726ArgfsX4	2307insA
c.2195T>G	p.Leu732X	L732X
c.2215delG	p.Val739TyrfsX16	2347delG
c.2290C>T	p.Arg764X	R764X
c.2453delT	p.Leu818TrpfsX3	2585delT
c.2464G>T	p.Glu822X	E822X
c.2490+1G>A		2622+1G>A
c.2491G>T	p.Glu831X	E831X
c.2537G>A	p.Trp846X	W846X
c.2551C>T	p.Arg851X	R851X
c.2583delT	p.Phe861LeufsX3	2711delT
c.2657+5G>A		2789+5G>A
c.2668C>T	p.Gln890X	Q890X
c.2780T>C	p.Leu927Pro	L927P
c.2834C>T	p.Ser945Leu	S945L
c.2875delG	p.Ala959HisfsX9	3007delG

c.2908G>C	p.Gly970Arg	G970R
c.2988+1G>A		3120+1G>A
c.2988G>A		3120G>A
c.2989-1G>A		3121-1G>A
c.3140-26A>G		3272-26A>G
c.3194T>C	p.Leu1065Pro	L1065P
c.3196C>T	p.Arg1066Cys	R1066C
c.3197G>A	p.Arg1066His	R1066H
c.3230T>C	p.Leu1077Pro	L1077P
c.3266G>A	p.Trp1089X	W1089X
c.3276C>A	p.Tyr1092X	Y1092X (C>A)
c.3276C>G	p.Tyr1092X	Y1092X (C>G)
c.3302T>A	p.Met1101Lys	M1101K
c.3310G>T	p.Glu1104X	E1104X
c.3472C>T	p.Arg1158X	R1158X
c.3484C>T	p.Arg1162X	R1162X
c.3528delC	p.Lys1177SerfsX15	3659delC
c.3587C>G	p.Ser1196X	S1196X
c.3611G>A	p.Trp1204X	W1204X (c.3611G>A)
c.3612G>A	p.Trp1204X	W1204X (c.3612G>A)
c.3659delC	p.Thr1220LysfsX8	3791delC
c.3717+12191C>T		3849+10kbC>T
c.3731G>A	p.Gly1244Glu	G1244E
c.3744delA	p.Lys1250ArgfsX9	3876delA
c.3752G>A	p.Ser1251Asn	S1251N
c.3773_3774insT	p.Leu1258PhefsX7	3905insT
c.3846G>A	p.Trp1282X	W1282X
c.3873+1G>A		4005+1G>A
c.3884_3885insT	p.Ser1297PhefsX5	4016insT
c.3909C>G	p.Asn1303Lys	N1303K
c.3937C>T	p.Gln1313X	Q1313X
c.3964-78_4242+577del		CFTRdele22,23
c.4077_4080delTGTTinsAA		4209TGTT>AA
c.4251delA	p.Glu1418ArgfsX14	4382delA

Italics indicate that these variants are conditionally reported.