

CFTR Variants Detected by Retired CF Assays

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(**Bold** = ACMG recommended 23 pathogenic variants)

Prior to Nov 1991 (1 variant)	Nov 1991-Aug 1994 (10 variants)	Aug 1994-Dec 1996 (11 variants)	Jan 1997-Jan 2001 (12 variants)	Feb 2001-Nov 2002 (20 variants)	Nov 2002-Nov 2007 (29 variants)	Nov 2007-Jul 2010 (24 variants)	Jul 2010-Oct 2016 (39 variants)
F508del	F508del	F508del	F508del	F508del	F508del	F508del	F508del
	I507del	I507del	NOT IN PANEL	I507del	I507del	I507del	I507del
	G542X	G542X	G542X	G542X	G542X	G542X	G542X
	S549N	S549N	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	S549N
	S549I	S549I	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL
	S549R	S549R	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL	S549R
	G551D	G551D	G551D	G551D	G551D	G551D	G551D
	R553X	R553X	R553X	R553X	R553X	R553X	R553X
	R560T	R560T	NOT IN PANEL	NOT IN PANEL	R560T	R560T	R560T
	N1303K	N1303K	N1303K	N1303K	N1303K	N1303K	N1303K
		W1282X	W1282X	W1282X	W1282X	W1282X	W1282X
			R117H	R117H	R117H	R117H	R117H
			621+1G>T	621+1G->T	621+1G>T	621+1G>T	621+1G>T
			R334W	R334W	R334W	R334W	R334W
			1717-1G>A	1717-1G->A	1717-1G>A	1717-1G>A	1717-1G>A
			R1162X	R1162X	R1162X	R1162X	R1162X
			3849+10kbC>T	3849+10kbC->T	3849+10kbC>T	3849+10kbC>T	3849+10kbC>T
			E60X	E60X	NOT IN PANEL	E60X	E60X
			1078delT	1078delT	1078delT	1078delT	1078delT
			R347P	R347P	R347P	R347P	R347P
			A455E	A455E	A455E	A455E	A455E
			2183AA->G	2183AA>G	NOT IN PANEL	2183AA>G	2183AA>G
			3659delC	3659delC	3659delC	3659delC	3659delC
			S1251N	S1251N	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL
				G85E	G85E	G85E	G85E
				I148T	NOT IN PANEL	NOT IN PANEL	NOT IN PANEL
				711+1G>T	711+1G>T	711+1G>T	711+1G>T
				1898+1G>A	1898+1G>A	1898+1G>A	1898+1G>A
				2184delA	2184delA	2184delA	2184delA
				2789+5G>A	2789+5G>A	2789+5G>A	2789+5G>A
				3120+1G>A	3120+1G>A	3120+1G>A	3120+1G>A
				D1152H	NOT IN PANEL	D1152H	D1152H
						Q493X	Q493X
						3905insT	3905insT
						Y1092X	Y1092X
						V520F	V520F
						R347H	R347H
						D1270N	D1270N
						3876delA	3876delA
						394delTT	394delTT
						3849+4A>G	3849+4A>G
						Y122X	Y122X