CFTR TaqMan Genotyping Assays

Applied Biosystems™ TaqMan™ Genotyping Assays offer a research menu of 198 cystic fibrosis transmembrane conductance regulator (CFTR) variants. See below for a list of available mutations to customize an Applied Biosystems™ TaqMan™ OpenArray™ Genotyping Plate today.

Common mutations*					
1078delT	2184delA	621+1G>T	G542X	Q890X	R347P
1677delTA	2789+5G>A	711+1G>T	G551D	R1066C	R553X
1717-1G>A	3120+1G>A	711+5G>A	G85E	R1158X	R560T
1811+1.6kbA>G	3659delC	A455E	I507del	R1162X	R75X
1898+1G>A	3849+10kbC>T	A559T	L206W	R117C	S1251N
1898+5G>T	3905insT	D1152H	M1101K	R117H	S549N
2143delT	394delTT	E60X	N1303K	R334W	V520F
2183AA>G	406-1G>A	F508del	Q552X	R347H	W1282X
Regional mutations*					
European					
1154insTC	2869insG	D110H	I336K	Q220X	S492F
1525-1G>A	4005+1G>A	E585X	K710X	Q493X	S945L
1812-1G>A	405+1G>A	E822X	L1065P	R1066H	T338I
2043delG	457TAT>G	E92K	L1077P	R1070Q	W846X(c.2537G>A)
2184insA	574delA	G1244E	L558S	R352Q	W846X(c.2538G>A)
2711delT	852del22	G178R	P67L	S1196X	Y122X
African-American		**Hispanic**		**Middle Eastern**	
2307insA	G330X	1288insTA	663delT	1548delG	
3791delC	G480C	2055del9>A	935delA	I1234V	
405+3A>C	S1255X	3171delC	H199Y	Q359K/T360K	
444delA		3199del6	P205S	Y569D	
		3876delA	Q98R		
Additional mutations					
1213delT	2622+1G>A	c.1486T>G	G551S	Q39X	S466X(c.1397C>A)
1248+1G>A	2789+2insA	c.4028delG	G622D	Q525X	S466X(c.1397C>G)
1259insA	2942insT	C524X	G970R	Q98X	S489X
1341+1G>A	3007delG	CFTRdele2.3	I506V	R1070W	S549R(c.1645A>C)
1461ins4	3120G>A	CFTRdele22.23	I507V	R1283M	S549R(c.1647T>G)
1471delA	3121-1G>A	D579G	L227R	R170H	T351S
1717-8G>A	3272-26A>G	D614G	L467P	R334L	W1089X
1833delT	3667del4	D836Y	L732X	R352W	W1204X(c.3611G>A)
1898+3A>G	3821delT	delF311	L927P	R560G	W1204X(c.3612G>A)
1924del7	4209TGTT>AA	E1104X	L967S	R560K	W401X(c.1202G>A)
1949del84	4382delA	E1371X	M1V	R709X	W401X(c.1203G>A)
2105-2117 del13insAGAAA	5T	E831X	P1013H	R764X	Y1092X(c.3276C>A)
2108delA	711+3A>G	E92X	P574H	R851X	Y1092X(c.3276C>G)
2183delAA	712-1G>T	F508C	P5L	S1255P	
2347delG	936delTA	G1069R	Q1238X	S341P	
2585delT	9T	G1349D	Q1313X	S364P	

*Demographics data source: Castellani C et al. (2008) Consensus on the use and interpretation of cystic fibrosis mutation analysis clinical practice. *J Cyst Fibros* 7:179–196.

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