Cystic Fibrosis Mutation Panel

Test ID: CFMPL [3070431]

Methodology: The CFMPL test uses targeted multiplexed PCR and Luminex tag sorting to identify the presence/absence of 39 pathogenic mutations in genomic DNA.

Genomic coordinates use the CFTR reference transcript NM_000492.3

Variant List:

Legacy Name	HGVS Nomenclature	Gene	Associated Phenotype
F508del	c.1521_1523del	*CFTR*	Cystic fibrosis
I507del	c.1519_1521del		
G542X	c.1624G>T		
G85E	c.254G>A		
621+1G>T	c.489+1G>T		
711+1G>T	c.579+1G>T		
N1303K	c.3909C>G		
R334W	c.1000C>T		
R347P	c.1040G>C		
A455E	c.1364C>A		
1717-1G>A	c.1585-1G>A		
R560T	c.1679G>C		
R553X	c.1657C>T		
G551D	c.1652G>A		
1898+1G>A	c.1766+1G>A		
2184delA	c.2052delA		
2789+5G>A	c.2657+5G>A		
3120+1G>A	c.2988+1G>A		
R1162X	c.3484C>T		
3659delC	c.3528delC		
3849+10kbC>T	c.3717+12191C>T		
W1282X	c.3846G>A		
R117H	c.350G>A		

1078delT	c.948delT		
394delTT	c.262_263delTT		
Y122X	c.366T>A		
R347H	c.1040G>A		
V520F	c.1558G>T		
A559T	c.1675G>A		
S549N	c.1646G>A		
S549R	c.1645A>C/T		
1898+5G>T	c.1766+5G>T		
2183AA>G	c.2051_2052delAAinsG		
2307insA	c.2175_2176insA		
Y1092X	c.3276C>A/G		
M1101K	c.3302T>A		
S1255X	c.3764C>A		
3876delA	c.3744delA		
3905insT	c.3773_3774insT		
5T[a]	c.1210-121T[5]		

a – The 5T allele is reported as ‘5T Present’ or ‘5T Absent’ only in specimens determined to carry the R117H variant.