

Cystic Fibrosis Reference Materials NOT Previously Characterized

Allele 1 ¹	Allele 2 ¹	Transformed	Cell type	Cell Bank	Cell line number	DNA number
F508del	W1282X	yes	Epithelial	ATCC	CRL-2777	
711 + 1G>T		?	CV	ECACC	DD1406	
F508del	G551D	no	Fibroblast	Coriell	GM00142	
G542X	H609R	yes	B-lymphocyte	Coriell	GM00504	
F508del	F508del	no	Fibroblast	Coriell	GM00770	
F508del	R347P	yes	B-lymphocyte	Coriell	GM00897	
621+1G-T	1154insTC	no	Fibroblast	Coriell	GM00999	
G542X	H609R	yes	B-lymphocyte	Coriell	GM01530	
R1162L		yes	B-lymphocyte	Coriell	GM01805	
F508del	V520F	no	Fibroblast	Coriell	GM02828	
F508del	W1282X	no	Fibroblast	Coriell	GM03401	
F508del	G551D	no	Fibroblast	Coriell	GM03465	
444delA	1812-1G>A	yes	B-lymphocyte	Coriell	GM04330	
F508del	G85E	no	Fibroblast	Coriell	GM04339	
F508del	W1282X	yes	B-lymphocyte	Coriell	GM04345	NA04345
F508del	F508del	yes	B-lymphocyte	Coriell	GM04540	NA04540
F508del	E92X	no	Fibroblast	Coriell	GM06966	
F508del	3849+10kbC>	yes	B-lymphocyte	Coriell	GM07381	NA07381
R553X		yes	B-lymphocyte	Coriell	GM07464	NA07464
F508del	R553X	yes	B-lymphocyte	Coriell	GM07469	NA07469
R553X		yes	B-lymphocyte	Coriell	GM07674	NA07674
566delA		yes	B-lymphocyte	Coriell	GM07829	NA07829
F508del	566delA	yes	B-lymphocyte	Coriell	GM07830	NA07830
M1101K		yes	B-lymphocyte	Coriell	GM07854	
M1101K	M1101K	yes	B-lymphocyte	Coriell	GM07860	NA07860
F508del	G551D	yes	B-lymphocyte	Coriell	GM08342	NA08342
F508del	R1162X	no	Fibroblast	Coriell	GM08889	
F508del	G551D	yes	B-lymphocyte	Coriell	GM11274	
F508del	G551D	yes	B-lymphocyte	Coriell	GM11286	NA11286
F508del	129G>C	yes	B-lymphocyte	Coriell	GM11279	NA11279
A455E	621+1G>T	yes	B-lymphocyte	Coriell	GM11290	NA11290
G542X		yes	B-lymphocyte	Coriell	GM11497	NA11497
G551D	R553X	yes	B-lymphocyte	Coriell	GM11761	NA11761
Q493X		yes	B-lymphocyte	Coriell	GM12926	
F508del	Δ exons 2,3	yes	B-lymphocyte	Coriell	GM18668	
F508del	1784, G>A	no	Fibroblast	Coriell	GM03465	
394delTT		yes	B-lymphocyte	Coriell	GM21080	
1778, G>A		yes	B-lymphocyte	Coriell	GM20932	
1778, G>A		yes	B-lymphocyte	Coriell	GM20837	
1779, T>G		yes	B-lymphocyte	Coriell	GM20915	NA20915
1172, G>A		yes	B-lymphocyte	Coriell	GM20834	
3876delA		yes	B-lymphocyte	Coriell	GM21069	
3905insT		yes	B-lymphocyte	Coriell	GM21572	

last updated 02/22/2008

¹ Allele names given here are colloquial. For proper CFTR mutation nomenclature, please refer to the table