

Cystic Fibrosis Reference Materials Characterized by GeT-RM and Other Sources

Allele 1 ¹	Allele 2 ¹	Transformed	Cell type	Cell Bank	Cell line number	DNA number	on Coriell MUTCF-2 panel	DNA sequencing	Characterization Methods ² (# labs)	Refs. ³
M1101K	M1101K	yes	B-lymphocyte	Coriell	GM07857	NA07857	no	1 lab	A (1), B (1)	1
F508del	Y1092XC>A	yes	B-lymphocyte	Coriell	GM11285	NA11285	no	1 lab	A (1), B (1), C (1)	1
F508del	P574H	yes	B-lymphocyte	Coriell	GM11287	NA11287	no	2 labs	B (1)	1
F508del	G178R	yes	B-lymphocyte	Coriell	GM11288	NA11288	no	1 lab	A (1), B (1)	1
444delA	1812-1G>A	yes	B-lymphocyte	Coriell	GM11370	NA11370	no	1 lab	A (1), B (1), C (1)	1
R347H	wt	yes	B-lymphocyte	Coriell	GM20737	NA20737	no	1 lab	A (1), B (1), C (1), D (1), E	1
3876delA	wt	yes	B-lymphocyte	Coriell	GM20741	NA20741	no	1 lab	A (1), B (1), C (1), D (1), E	1
S549N	wt	yes	B-lymphocyte	Coriell	GM20745	NA20745	no	1 lab	A (1), B (1), C (1), D (1), E	1
3905insT	wt	yes	B-lymphocyte	Coriell	GM20836	NA20836	no	1 lab	A (1), B (1), C (1), D (1)	1
S549R	wt	yes	B-lymphocyte	Coriell	GM20925	NA20925	no	1 lab	A (1), B (1), C (1), D (1)	1
F508del	I507V	yes	B-lymphocyte	Coriell	GM21551	NA21551	no	1 lab	A (1), B (1), D (1)	1
R75X	wt	yes	B-lymphocyte	Coriell	GM20924	NA20924	no	1 lab	B (1)	1
E60X	F508del	yes	B-lymphocyte	Coriell	GM07732	NA07732	no	1 lab	B (1), C (1)	1
3120+G>A	621+1G>T	yes	B-lymphocyte	Coriell	GM07441	NA07441	yes	4 labs	J (1), K (1), L (2), M (1), Q	2, 3
F508del	R117H	yes	B-lymphocyte	Coriell	GM13591	NA13591	yes	4 labs	J (1), K (1), L (2), M (1), Q	2, 3
F508del	2184delA	yes	B-lymphocyte	Coriell	GM18799	NA18799	yes	1 lab		1, 2
F508del	1898+1G>A	yes	B-lymphocyte	Coriell	GM18800	NA18800	yes	1 lab		1, 2
ΔI507		yes	B-lymphocyte	Coriell	GM11277	NA11277	yes	1 lab		1, 2
F508del	F508del	yes	B-lymphocyte	Coriell	GM01531	NA01531	yes	1 lab		1, 2
G542X	G542X	yes	B-lymphocyte	Coriell	GM11496	NA11496	yes	1 lab		1, 2
F508del	R553X	yes	B-lymphocyte	Coriell	GM07552	NA07552	yes	1 lab		1, 2
G551D		yes	B-lymphocyte	Coriell	GM08338	NA08338	yes	1 lab		1, 2
2789+5G>A	2789+5G>A	yes	B-lymphocyte	Coriell	GM11859	NA11859	yes	1 lab		1, 2
W1282X		yes	B-lymphocyte	Coriell	GM11723	NA11723	yes	1 lab		1, 2
3849+10C>T	3849+10C>T	yes	B-lymphocyte	Coriell	GM11860	NA11860	yes	1 lab		1, 2
711+1G>T	621+1G>T	yes	B-lymphocyte	Coriell	GM11280	NA11280	yes	1 lab		1, 2
G85E	621+1G>T	yes	B-lymphocyte	Coriell	GM11282	NA11282	yes	1 lab		1, 2
R1162X		yes	B-lymphocyte	Coriell	GM12585	NA12585	yes	1 lab		1, 2
1717-1G>A		yes	B-lymphocyte	Coriell	GM12444	NA12444	yes	1 lab		1, 2
G1349D	N1303K	yes	B-lymphocyte	Coriell	GM11472	NA11472	yes	1 lab		1, 2
G551D	R347P	yes	B-lymphocyte	Coriell	GM12785	NA12785	yes	1 lab		1, 2
R334W	?	yes	B-lymphocyte	Coriell	GM12960	NA12960	yes	1 lab		1, 2
3659delC	F508del	yes	B-lymphocyte	Coriell	GM11275	NA11275	yes	1 lab		1, 2
F508del	621+1G>T	yes	B-lymphocyte	Coriell	GM11281	NA11281	yes	1 lab		1, 2
F508del	A455E	yes	B-lymphocyte	Coriell	GM11283	NA11283	yes	1 lab		1, 2
F508del	R560T	yes	B-lymphocyte	Coriell	GM11284	NA11284	yes	1 lab		1, 2
R553X		yes	B-lymphocyte	Coriell	GM07461		no	1 lab		1
F508del	F508del	yes	B-lymphocyte	Coriell	GM08345	NA08345	no	1 lab		1

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F508del	Q493X	yes	B-lymphocyte	Coriell	GM11278	NA11278	no	1 lab		1
V520F		yes	B-lymphocyte	Coriell	GM12961	NA12961	no	1 lab		1
F508C		yes	B-lymphocyte	Coriell	GM13033	NA13033	no	1 lab		1
G85E	D1152H	yes	B-lymphocyte	Coriell	GM13423	NA13423	no	1 lab		1
1078delT		yes	B-lymphocyte	Coriell		CD00003	no		F (1), N (1), Q (1)	3
1898+1G>A		yes	B-lymphocyte	Coriell		CD00004	no		F (1), N (1), Q (1)	3
1898+1G>A		yes	B-lymphocyte	Coriell		CD00005	no		F (1), N (1), Q (1)	3
1898+1G>A		yes	B-lymphocyte	Coriell		CD00006	no		F (1), N (1), Q (1)	3
1898+1G>A		yes	B-lymphocyte	Coriell		CD00007	no		F (1), N (1), Q (1)	3
2184delA		yes	B-lymphocyte	Coriell		CD00008	no		F (1), N (1), Q (1)	3
394delTT		yes	B-lymphocyte	Coriell		CD00009	no		F (1), N (1), Q (1)	3
I148T		yes	B-lymphocyte	Coriell		CD00010	no		F (1), N (1), Q (1)	3
I148T		yes	B-lymphocyte	Coriell		CD00012	no		F (1), N (1), Q (1)	3
S1235R		yes	B-lymphocyte	Coriell		CD00013	no	6 labs	I (1), O (1), P (2)	3

last updated 03/11/2008

¹ Allele names given here are colloquial. For proper CFTR mutation nomenclature, please refer to the table titled "CFTR Mutation HGVS Nomenclature" on the GeT-RM website

² Methods: A) CFTR 70+6 (Luminex), B) In-House Developed Assay, C) Invader® Assay (Third Wave Technologies), D) OLA V3.0 (Abbott/Celera), E) Signature® CF Expand (Asuragen), F) INNO-LiPA reverse hybridization (Innogenetics), G) Mass Spectrometry, H) Allele Specific Amplification Assay with gel electrophoresis, I) Restriction Fragment Length Polymorphism, J) Restriction Fragment Length Polymorphism + sequencing, K) Heteroduplex Analysis, L) Restriction Fragment Length Polymorphism + Heteroduplex Analysis + sequencing, M) eSensor® chip (Motorola Life Sciences) + sequencing, N) 87 mutation Heteroduplex Assay (Genzyme Genetics), O) Heteroduplex Analysis + sequencing, P) Restriction Fragment Length Polymorphism + Heteroduplex Analysis + Oligonucleotide Ligation Assay, Q) Oligonucleotide Ligation Assay (Applied Biosystems)

³ References: 1) GeT-RM manuscript in preparation; 2) Coriell's ACMG23 panel (MUTCF) panel (ccr.coriell.org/Sections/Search/Panel_Detail.aspx?Ref=MUTCF-2&PgId=202); 3) SH Bernacki et al., Clin Chem 51(11):2013-2024 (2005)