

Cystic Fibrosis Reference Materials Characterized by the GeT-RM

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 ^{2,3} (# labs)	Refs. ⁴
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	1 lab	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), E (1)	1
R347H	wt	yes	B-lymphocyte	Coriell	GM20737	NA20737	no	1 lab	A (1), B (1), C (1), D (1), E (1)	1
3876delA	wt	yes	B-lymphocyte	Coriell	GM20741	NA20741	no	1 lab	A (1), B (1), C (1), D (1), E (1)	1
S549N	wt	yes	B-lymphocyte	Coriell	GM20745	NA20745	no	1 lab	A (1), B (1), C (1), D (1), E (1)	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

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)

³ Assays indicated are designed to detect the expected allele(s). Other assays, not designed to detect these alleles were also performed, giving no false positive results.

⁴ References: 1) GeT-RM manuscript in preparation