

Ashkenazi Jewish Reference Materials Characterized by the GeT-RM

Disorder	Gene	Allele 1	Allele 2	Other Mutations (allele ID'd/#labs) ¹	Transformed	Cell Type	Cell Bank	Cell Line Number	DNA Number	Characterization methods ² (# labs)
Bloom	RECQL3	2281Del6/Ins7	2281Del6/Ins7		yes	B-lymphocyte	Coriell	GM03403	NA03403	TM-ASR (4), PCR/ASOH (2)
Bloom	RECQL3	2281Del6/Ins7			yes	B-lymphocyte	Coriell	GM04408	NA04408	TM-ASR (4), PCR/ASOH (2)
Canavan	ASPA	E285A	E285A		no	fibroblast	Coriell	GM04268	NA04268	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Canavan	ASPA	E285A			yes	B-lymphocyte	Coriell	GM17821	NA17821	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Canavan	ASPA	Y231X			yes	B-lymphocyte	Coriell	GM18929	NA18929	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Canavan	ASPA	A305E	G176D (0) ³		no	fibroblast	Coriell	GM00059	NA00059	TM-ASR (3), PCR/ASOH (1)
Fanconi group C	FANCC	IVS4+4A>T	IVS4+4A>T		no	fibroblast	Coriell	GM00449	NA00449	TM-ASR (4), PCR/ASOH (2)
Fanconi group C	FANCC	322delG			yes	B-lymphocyte	Coriell	GM12794	NA12794	TM-ASR (4), PCR/ASOH (2)
familial dysautonomia	IKBKAP	IVS20+6T>C			yes	B-lymphocyte	Coriell	GM05046	NA05046	TM-ASR (4), PCR/ASOH (1), RT-PCR (1)
familial dysautonomia	IKBKAP	IVS20+6T>C	IVS20+6T>C		yes	B-lymphocyte	Coriell	GM05106	NA05106	TM-ASR (4), PCR/ASOH (1), RT-PCR (1)
Gaucher	GBA	84GG	N370S		no	fibroblast	Coriell	GM00852	NA00852	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Gaucher	GBA	N370S	V394L (4/4)**		no	fibroblast	Coriell	GM01607	NA01607	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Gaucher	GBA	L444P			no	fibroblast	Coriell	GM04394	NA04394	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Gaucher	GBA	L444P	L444P		yes	B-lymphocyte	Coriell	GM08752	NA08752	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Gaucher	GBA	N370S	N370S		yes	B-lymphocyte	Coriell	GM10870	NA10870	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Gaucher	GBA	IVS2+1G>A			yes	B-lymphocyte	Coriell	GM01031	NA01031	TM-ASR (3), PCR/ASOH (1)
Glycogen Storage 1a	G6PC	Q347X	Q347X		yes	B-lymphocyte	Coriell	GM11215	NA11215	PCR/SNPE/CE (1), PCR/ASOH (1); RT-PCR(1)

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Glycogen Storage 1a	G6PC	R83C	Q347X		yes	B-lymphocyte	Coriell	GM11468	NA11468	PCR/SNPE/CE (1), PCR/ASOH (1); RT-PCR (1)
Glycogen Storage 1a	G6PC	R83C			yes	B-lymphocyte	Coriell	GM11470	NA11470	PCR/SNPE/CE (1), PCR/ASOH (1); RT-PCR (1)
Glycogen Storage 1a	G6PC	Q347X			yes	B-lymphocyte	Coriell	GM11471	NA11471	PCR/SNPE/CE (1), PCR/ASOH (1); RT-PCR (1)
Mucopolipidosis type IV	MCOLN1	IVS3-2A>G	IVS3-2A>G	Gaucher N370S (5/5)	no	fibroblast	Coriell	GM02528	NA02528	TM-ASR (4), RT-PCR (2)
Mucopolipidosis type IV	MCOLN1	IVS3-2A>G	Del6.4Kb		yes	B-lymphocyte	Coriell	GM02533	NA02533	TM-ASR (4), RT-PCR (2)
Niemann-Pick type A	SMPD1	L302P	L302P		no	fibroblast	Coriell	GM00112	NA00112	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Niemann-Pick type B	SMPD1	L302P			no	fibroblast	Coriell	GM03252	NA03252	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Niemann-Pick type A	SMPD1	fsP330			no	fibroblast	Coriell	GM13205	NA13205	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Niemann-Pick Type B	SMPD1	R496L	delR608 (5/5)**	Gaucher N370S (5/5)	no	fibroblast	Coriell	GM16193	NA16193	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Tay-Sachs	HEXA	G269S	IVS12+1G>C		yes	B-lymphocyte	Coriell	GM03461	NA03461	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Tay-Sachs	HEXA	G269S	1278_1279insTAT C		yes	B-lymphocyte	Coriell	GM03770	NA03770	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Tay-Sachs	HEXA	1278_1279insTAT C	1278_1279insTAT C		yes	B-lymphocyte	Coriell	GM11852	NA11852	TM-ASR (4), PCR/SNPE/CE (1), PCR/ASOH (1)
Tay-Sachs	HEXA	IVS9(+1)G>A			yes	B-lymphocyte	Coriell	GM09787	NA09787	TM-ASR (3), PCR/ASOH (1)
Tay-Sachs	HEXA	1278insTATC	R247W (3/4) ⁴	Gaucher N370S (3/3)	no	fibroblast	Coriell	GM04863	NA04863	TM-ASR (3), PCR/ASOH (1)

last updated: 02-26-2008

¹Not all labs assayed for all alleles. (Number of labs who identified allele/Number labs performing assay)

²Methods- **TM-ASR** - TM-Biosciences Tag-IT Mutation Detection Kit Ashkenazi Jewish Panel; **PCR/ASOH**- PCR followed by Allele-Specific Oligo hybridization and detection with chemiluminescent or radiolabeled probes; **PCR/SNPE/CE**- PCR followed by single nucleotide primer extension and capillary electrophoresis; **RT-PCR** - Real Time PCR

³Private mutation, not confirmed in this study

⁴One lab was unable to confirm this allele using the TM-ASR