

Ashkenazi Jewish Reference Materials NOT Previously Characterized

Disorder	Gene	Allele 1	Allele 2	Transformed	Cell type	Cell Bank	Cell line Number	DNA Number	Verified
Bloom	RECQL3	2281del6ins7	2281del6ins7	yes	B-lymphocyte	Coriell	GM09960	NA09960	no
Bloom	RECQL3	1784C>A	1784C>A	yes	B-lymphocyte	Coriell	GM16375		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	yes	B-lymphocyte	Coriell	GM16377		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM00811		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM01492		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM02085		no
Bloom	RECQL3	2996delA	?	no	fibroblast	Coriell	GM02520		no
Bloom	RECQL3	1610insA	2328C>T	no	fibroblast	Coriell	GM02548		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM02932		no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM03402		no
Bloom	RECQL3	2281del6ins7	1bpdel3233	no	fibroblast	Coriell	GM03498		no
Bloom	RECQL3	1610insA	1610insA	no	fibroblast	Coriell	GM05289	NA05289	no
Bloom	RECQL3	2281del6ins7	2281del6ins7	no	fibroblast	Coriell	GM16859		no
Bloom	RECQL3	2695C>T	2695C>T	no	fibroblast	Coriell	GM16860		no
Bloom	RECQL3	2923delC	2923delC	no	fibroblast	Coriell	GM16881		no
Bloom	RECQL3	3261delT	2281del6ins7	no	fibroblast	Coriell	GM16882		no
Bloom	RECQL3	2051A>G	IVS5-2A>G	no	fibroblast	Coriell	GM16883		no
Bloom	RECQL3	2098C>T	?	no	fibroblast	Coriell	GM16886		no
Bloom	RECQL3	1933C>T	1933C>T	no	fibroblast	Coriell	GM16890		no
Bloom	RECQL3	1933C>T	1933C>T	no	fibroblast	Coriell	GM16891		no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C		yes	B-lymphocyte	Coriell	GM05107	NA05107	no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C		yes	B-lymphocyte	Coriell	GM05108	NA05108	no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C		yes	B-lymphocyte	Coriell	GM16768		no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C		yes	B-lymphocyte	Coriell	GM16769		no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C	2507(+ 6)T>C	yes	B-lymphocyte	Coriell	GM16770		no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C	2507(+ 6)T>C	yes	B-lymphocyte	Coriell	GM16771		no
F. Dysautonomia	IKBKAP	2507(+ 6)T>C	2507(+ 6)T>C	yes	B-lymphocyte	Coriell	GM16772		no
Gaucher type I	GBA	N370S	N370S	yes	B-lymphocyte	Coriell	GM10871		no
Gaucher type I	GBA	N370S	N370S	yes	B-lymphocyte	Coriell	GM10873	NA10873	no
Gaucher type I	GBA	N370S	N370S	yes	B-lymphocyte	Coriell	GM10874	NA10874	no
Gaucher type I	GBA	N370S	1-BP INS, 84GG	no	fibroblast	Coriell	GM00372		no
Gaucher type I	GBA	L444P	L444P	no	fibroblast	Coriell	GM07968		no
Gaucher type I	GBA	L444P	L444P	no	fibroblast	Coriell	GM10915		no
Gaucher type I	GBA	N370S		no	fibroblast	Coriell	GM02528	NA02528	no

Disorder	Gene	Allele 1	Allele 2	Transformed	Cell type	Cell Bank	Cell line Number	DNA Number	Verified
Gaucher type II	GBA	L444P	L444P (& 2 bp	no	fibroblast	Coriell	GM00877		no
Gaucher type II	GBA	6482C, 6468C,	wt	no	fibroblast	Coriell	GM00878		no
Gaucher type II	GBA	P415R	L444P	no	fibroblast	Coriell	GM01260		no
Gaucher type II	GBA	G325R	C342G	no	fibroblast	Coriell	GM02627		no
Gaucher type II	GBA	L444P	L444P	no	fibroblast	Coriell	GM08760		no
Glycogen Storage 1a	G6PC	Q347X	Q347X	yes	B-Lymphocyte	Coriell	GM11416	NA11416	no
Niemann-Pick Type A	SMPD1	1-BP del, P330fs		no	fibroblast	Coriell	GM00559		no
Niemann-Pick Type A	SMPD1	R496L	1-BP del, P330fs	no	fibroblast	Coriell	GM16194		no
Niemann-Pick Type A	SMPD1	L302P	L302P	no	fibroblast	Coriell	GM16195		no
Tay-Sachs	HEXA	G269S	IVS12(+1)G>C	yes	B-lymphocyte	Coriell	GM03441	NA03441	no
Tay-Sachs	HEXA	G269S	1278insTATC	yes	B-lymphocyte	Coriell	GM03575	NA03575	no
Tay-Sachs	HEXA	1278insTATC		no	fibroblast	Coriell	GM00515		no
Tay-Sachs	HEXA	W420C	?	no	fibroblast	Coriell	GM00527		no
Tay-Sachs	HEXA	Q482K	Q482K	no	fibroblast	Coriell	GM01110		no
Tay-Sachs	HEXA	1278insTATC	splice site mut	no	fibroblast	Coriell	GM02968		no
Tay-Sachs	HEXA	1278insTATC	wt	no	fibroblast	Coriell	GM03052		no
Tay-Sachs	HEXA	1278insTATC	1278insTATC	no	fibroblast	Coriell	GM11853		no

last updated: 02-26-2008