

The Progeria Research Foundation Cell and Tissue Bank
Hutchinson-Gilford Progeria Syndrome and Progeroid Laminopathies
Cell Lines Available

LYMPHOBLAST NON-CLASSIC MUTATION					
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSALBV199 ³	Proband	11 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1868 C>G (p.Thr623Ser)	
PSALBV229 ³	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	
PSALBV296 ³	Proband	10 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	PSADFN328
PSALBV379 ³	Proband	5 yrs 3 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c.1968+1G>A	
PSALBV406 ³	Proband	8 mos	Male	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	
PSALBV427 ³	Proband	3 mos.	Male	LMNA Exon 11, heterozygous c.1821G>A(p.V607V)	
PSALBV432 ³	Proband	4 yrs 5 mos	Male	LMNA Exon 9, homozygous c.1580G>T, (p.A527L)	

¹Representative cultures from this cell line have tested negative for mycoplasma contamination at The PRF Cell and Tissue Bank. Please note: mycoplasma testing is performed on random passages. As part of our on-going quality control, The PRF Cell and Tissue Bank periodically tests the cultures for mycoplasma contamination using R&D Systems Mycoplasma Detection Kit (catalog # CUL001B).

²Histograms of mutational analysis sequenced by the PRF Cell and Tissue Bank available.

³Representative cultures from this cell line have tested negative for mycoplasma contamination at Rutgers University Cell and DNA Repository via real time PCR assay.

⁴Representative cultures from this cell line have tested negative for mycoplasma contamination at Ottawa Hospital Research Institute. Please note: mycoplasma testing is performed on random passages. As part of our on-going quality control, The Human Pluripotent Stem Cell Facility/Dr. William Stanford laboratory periodically tests the cultures for mycoplasma contamination using a PCR based approach (Detection of mycoplasma contaminations., Uphoff CC, Drexler HG., Methods Mol Biol. 2013;946:1-13. doi: 10.1007/978-1-62703-128-8_1. PMID:23179822).

⁵Genetic sequencing on blood DNA agrees with fibroblast DNA unless otherwise noted. Blood sequencing performed for the PRF Diagnostics Program or outside facility. Please contact the PRF Cell and Tissue Bank coordinator for additional details.

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LYMPHOBLAST NON-CLASSIC FAMILY CELL LINES					
PSALBV083 ³	Proband	6 mos	Male	LMNA Exon 11, c.1968+1 G>A	PSADFN086
PSFLBV084 ³	Father of PSALBV083	31 yrs 9 mos	Male	Not performed	
PSMLBV085 ³	Mother of PSALBV083	32 yrs 2 mos	Female	Not performed	
PSALBV245 ³	Proband, sibling of PSALBV339 (2 deceased siblings were homozygous for same mutation)	4 mos	Male	LMNA Exon 9, homozygous c.1579C>T(p.Arg527Cys)	
PSALBV339 ³	Proband, Sibling of PSALBV245 (2 deceased siblings were homozygous for same mutation)	1 mos	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSMLBV238 ³	Mother of PSALBV245 & PSALBV339	~24 yrs	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSFLBV239 ³	Father of PSALBV245 & PSALBV339	~25 yrs	Male	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSALBV295 ³	Proband	17 yrs 3 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6, 1158-44 C>T	PSADFN425
PSMLBV360 ³	Mother of PSALBV295	46 yrs 1 mos	Female	LMNA Exon 1, Negative Intron 6, heterozygous c.1158-44 C>T	
PSFLBV361 ³	Father of PSALBV295	49 yrs 8 mos	Male	LMNA Exon 1 & Intron 6, Negative	

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LYMPHOBLAST NON-CLASSIC FAMILY CELL LINES CONTINUED

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSALBV341 ³	Proband (sibling of PSALBV342)	5 yrs 2 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSADFN373
PSALBV342 ³	Proband (sibling of PSALBV341)	2 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	
PSMLBV343 ³	Mother of PSALBV341 & PSALBV342	32 yrs 2 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMDFN375
PSFLBV344 ³	Father of PSALBV341 & PSALBV342	32 yrs	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFDFN376

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