

DF=Dermal Fibroblast
LBV=Lymphoblast
iPSC = Induced Pluripotent Stem Cell
Cost of each DF & LBV cell line is \$80.50
Cost of each Immortalized Fibroblast cell line is \$80.50
Cost of each iPSC line is \$500.00

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

LYMPHOBLASTS: NON-CLASSIC HGPS MUTATION (PROGERIN-PRODUCING) Relation to Age at **Other Lines From** Cell Line # Gender **Mutational Analysis Proband Donation** This Donor LMNA Exon 11, heterozygous PSALBV1993 Proband 11 yrs 3 mos Female c.1868 C>G (p.Thr623Ser) LMNA Exon 11, heterozygous PSALBV229³ Proband 5 yrs 9 mos Female c.1822G>A (p.Gly608Ser) LMNA Exon 11, heterozygous PSALBV296³ Proband 10 yrs 8 mos Female PSADFN328 c.1822G>A (p.Gly608Ser) LMNA Exon 11/Intron 11 junction, PSALBV3793 Proband 5 yrs 3 mos Male heterozygous c.1968+1G>A LMNA Exon 11, heterozygous PSALBV406³ Proband Male 8 mos c.1822G>A (p.Gly608Ser) LMNA Exon 11, heterozygous PSALBV427³ Proband 3 mos. Male c.1821G>A(p.V607V)

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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.

⁶Mutational analysis was performed on fibroblasts only, not on DNA derived from blood

⁷Cell line has not been tested for the mutation(s). Mutational analysis is based on blood DNA.



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LYMPHOBLASTS: FAMILY SET CONTAINING A LYMPHOBLAST CELL LINE WITH A NON-CLASSIC HGPS MUTATION (PROGERIN-PRODUCING)

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSALBV083 ³	Proband	6 mos	Male	LMNA Exon 11, heterozygous 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	

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