



DF=Dermal Fibroblast
 LBV=Lymphoblast
 iPSC = Induced Pluripotent Stem Cell
 Cost of each DF & LBV cell line is \$150.00
 Cost of each Immortalized Fibroblast cell line is \$150.00
 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

FIBROBLASTS: NON-CLASSIC HGPS MUTATIONS (PROGERIN-PRODUCING)

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN328 ^{1, 2, 5} (Cells grow poorly)	Proband	12 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1822 G->A (p.Gly608Ser)	PSALBV296
PSADFN086 ^{2, 5} (Cells grow poorly)	Proband	0 yrs 7 mos	Male	LMNA Intron 11, heterozygous c.1968+1 G>A	PSALBV083
PSADFN576 ^{2, 5}	Proband	34 yrs 11 mos	Female	LMNA Exon 11/Intron 11 junction, heterozygous c. 1968+5G>A	

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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FIBROBLASTS: FAMILY SETS CONTAINING A FIBROBLAST CELL LINE WITH NON-CLASSIC HGPS MUTATION (PROGERIN-PRODUCING)					
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN489 ^{1,7}	Proband (mild progeroid phenotype)	3 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly); and SMC3 c.562A>G, p.K188E	
PSDFDN714 ^{1,7}	Father of HGADFN489	38 yrs 6 mos	Male	Negative	
PSMDFN713 ^{1,7}	Mother of HGADFN489	35 yrs 11 mos	Female	Negative	
PSADFN386 ¹	Proband	11 mos	Female	Mosaic: DNA from Fibroblasts: c.1968 +2T>C DNA from Blood: 4.7% c.1968+2T>C mutation, and 41.3% c.1968+2T>A mutation See reference: Bar DZ, Arlt MF, Brazier JF, et al. A novel somatic mutation achieves partial rescue in a child with Hutchinson-Gilford progeria syndrome. <i>Journal of Medical Genetics</i> 2017;54:212-216	
PSMDFN387 ¹	Mother of PSADFN386	36 yrs 5 mos	Female	LMNA Exon 11, Negative	
PSDFDN388 ¹	Father of PSADFN386	38 yrs 1 mo	Male	LMNA Exon 11, Negative	

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FIBROBLASTS: FAMILY SETS CONTAINING A FIBROBLAST CELL LINE WITH NON-CLASSIC HGPS MUTATION (PROGERIN-PRODUCING)

CONTINUED

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN325 ^{1, 2, 5}	Proband	6 yrs 9 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c.1968+5G>C	
PSDFDN327 ^{2, 5, 6}	Father of HGADFN325	36 yrs 3 mos	Male	LMNA Exon 11/Intron 11 junction, negative	
PSMDFN326 ^{2, 5, 6}	Mother of HGADFN325	36 yrs 10 mos	Female	LMNA Exon 11/Intron 11 junction, negative	
PSADFN392 ^{2, 5}	Proband	7 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1968+2T>C	
PSDFDN394 ^{1, 6}	Father of PSADFN392	49 yrs 1 mo	Male	LMNA Exon 11, negative ⁶	
PSMDFN393 ^{1, 6}	Mother of PSADFN392	44 yrs 8 mos	Female	LMNA Exon 11, negative ⁶	
PSMDFN346 ^{2, 5}	Mother of PSADFN345 (proband line not available)	21 yrs 10 mos	Female	LMNA Exon 11, Negative	

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