

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

### FIBROBLASTS: PROGEROID LAMINOPATHIES (NOT PROGERIN-PRODUCING)

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN485 <sup>1,7</sup>	Proband	4 yrs 5 mos	Male	LMNA heterozygous c.412G>A; (p.E138K)	
PSADFN425 <sup>1,7</sup>	Proband	20 yrs 11 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	PSALBV295
PSADFN412 <sup>1,2,5</sup>	Proband	7 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	
PSADFN257 <sup>2,5</sup>	Proband	1 yr 10 mos	Male	LMNA Exon 10, homozygous c.1619 T>C (p.Met540Thr)	
PSADFN542 <sup>6</sup>	Proband	75 yrs 2 mos	Male	LMNA Exon 11, heterozygous c.1930 C>T ( p.Arg644Cys)	

<sup>1</sup>Representative 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).

<sup>2</sup>Histograms of mutational analysis sequenced by the PRF Cell and Tissue Bank available.

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

<sup>4</sup>Representative 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).

<sup>5</sup>Genetic 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.

<sup>6</sup>Mutational analysis was performed on fibroblasts only, not on DNA derived from blood

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



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

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<b>FIBROBLASTS: FAMILY SETS CONTAINING PROGEROID LAMINOPATHY            FIBROBLAST CELL LINES (NOT PROGERIN-PRODUCING)</b>					
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN373 <sup>1,7</sup>	Proband	5 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSALBV341
PSFDFN376 <sup>1,7</sup>	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFLBV344
PSMDFN375 <sup>1,7</sup>	Mother of PSADFN373	32 yrs 9 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMLBV343
PSADFN317 <sup>7</sup>	Proband (& sibling of PSADFN318)	3 yr 9 mo	Male	ZMPste24 Exon 6, heterozygous c.743C>T(p.Pro248Leu); Exon 10, heterozygous c.1349G>A (p.Trp450Stop)	
PSADFN318 <sup>1,7</sup>	Proband (& sibling of PSADFN317)	5 mos	Male	ZMPste24 Exon 6, heterozygous c.743C>T(p.Pro248Leu); Exon 10, heterozygous c.1349G>A (p.Trp450Stop)	
PSFDFN319	Father of PSADFN317 & PSADFN318	39 yrs 0 mo	Male	Not performed	
PSMDFN320 <sup>1</sup>	Mother of PSADFN317 & PSADFN318	36 yrs 8 mo	Female	Not performed	

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**FIBROBLASTS: FAMILY SETS CONTAINING PROGEROID LAMINOPATHY  
FIBROBLAST CELL LINES (NOT PROGERIN-PRODUCING)**

*CONTINUED*

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN363 <sup>1, 2, 5</sup>	Proband	8 mos	Male	LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn)	
PSDFDN365 <sup>1, 2, 6</sup>	Father of PSADFN363	44 yrs 2 mos	Male	LMNA Exon 6, negative	
PSMDFN364 <sup>1, 2, 6</sup>	Mother of PSADFN363	36 yrs 10 mos	Female	LMNA Exon 6, negative	

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