

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

**FIBROBLAST NON-CLASSIC MUTATIONS**

PSADFN485 <sup>5</sup>	Proband	4 yrs 5 mos	Male	LMNA heterozygous c.412G>A; (p.E138K)	
PSADFN425 <sup>1,5</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)	
PSADFN328 <sup>1,2,5</sup> (Cells grow poorly)	Proband	12 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1822 G->A (p.Gly608Ser)	PSALBV296
PSADFN257 <sup>2,5</sup>	Proband	1 yr 10 mos	Male	LMNA Exon 10, homozygous c.1619 T>C (p.Met540Thr)	
PSADFN086 <sup>2,5</sup> (Cells grow poorly)	Proband	0 yrs 7 mos	Male	LMNA Exon 11, c.1968+1 G>A	PSALBV083
PSADFN542	Proband	75 yrs 2 mos	Male	LMNA heterozygous Exon 11, 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.

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<b>FAMILY NON-CLASSIC FIBROBLAST CELL LINES</b>					
<b>Cell Line #</b>	<b>Relation to Proband</b>	<b>Age at Donation</b>	<b>Gender</b>	<b>Mutational Analysis<sup>2</sup></b>	<b>Other Lines From This Donor</b>
PSADFN489	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	
PSFDFN714	Father of HGADFN489	38 yrs 6 mos	Male	Negative	
PSMDFN713	Mother of HGADFN489	35 yrs 11 mos	Female	Negative	
PSADFN392	Proband	7 yrs 4 mos	Male	LMNA Exon 11, c.1968+2T>C	
PSFDFN394 <sup>1</sup>	Father of PSADFN392	49 yrs 1 mo	Male	Not performed	
PSMDFN393 <sup>1</sup>	Mother of PSADFN392	44 yrs 8 mos	Female	Not performed	
PSADFN373 <sup>1</sup>	Proband	5 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSALBV341
PSFDFN376 <sup>1</sup>	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFLBV344
PSMDFN375 <sup>1</sup>	Mother of PSADFN373	32 yrs 9 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMLBV343
PSADFN363 <sup>1, 2, 5</sup>	Proband	8 mos	Male	LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn)	
PSFDFN365 <sup>1, 2, 5</sup>	Father of PSADFN363	44 yrs 2 mos	Male	Not performed	
PSMDFN364 <sup>1, 2, 5</sup>	Mother of PSADFN363	36 yrs 10 mos	Female	Not performed	

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Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSMDFN346 <sup>2,5</sup>	Mother of PSADFN345 (proband line not available)	21 yrs 10 mos	Female	LMNA Exon 11, Negative	
PSADFN325 <sup>1,2,5</sup>	Proband	6 yrs 9 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c. 1968+5G>C	
PSFDFN327 <sup>2,5</sup>	Father of HGADFN325	36 yrs 3 mos	Male	Not performed	
PSMDFN326 <sup>2,5</sup>	Mother of HGADFN325	36 yrs 10 mos	Female	Not performed	
PSADFN317 <sup>5</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>1,5</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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PSADFN386 <sup>1</sup>	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 <sup>1</sup>	Mother of PSADFN386	36 yrs 5 mos	Female	LMNA Exon 11, Negative	
PSFDFN388 <sup>1</sup>	Father of PSADFN386	38 yrs 1 mo	Male	LMNA Exon 11, Negative	

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