

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

FIBROBLASTS: CLASSIC MUTATION									
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor				
HGADFN122 <sup>1, 2, 5</sup>	Proband	5 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV097				
HGADFN127 <sup>1, 2, 5</sup>	Proband	3 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV039				
HGADFN143 <sup>1, 2, 5</sup>	Proband	8 yrs 10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV011				
HGADFN155 <sup>1, 2, 5</sup>	Proband	1 yr 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)					
HGADFN164 <sup>1, 2, 5</sup>	Proband	4 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)					
HGADFN169 <sup>1, 2, 5</sup>	Proband	8 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)					
HGADFN178 <sup>1, 2, 5</sup>	Proband	6 yrs 11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV110				
HGADFN188 <sup>1, 2, 5</sup>	Proband	2 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV152				
HGADFN271 <sup>1, 2, 5</sup>	Proband	1 yr 3 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV152				

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

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

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



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

# FIBROBLASTS: FAMILY SETS CONTAINING A FIBROBLAST CELL LINE WITH THE CLASSIC HGPS MUTATION

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
HGADFN367 <sup>1, 2, 5</sup>	Proband	3 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGFDFN369 <sup>1, 2, 6</sup>	Father of HGADFN367	33 yrs 9 mos	Male	LMNA Exon 11, Negative				
HGMDFN368 <sup>1, 2, 6</sup>	Mother of HGADFN367	31 yrs 7 mos	Female	LMNA Exon 11, Negative				
HGADFN167 <sup>1, 2, 5</sup>	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV009 iPSC lines			
HGFDFN168 <sup>1, 2, 5</sup>	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFLBV021 HGFDFSV40T168 iPSC lines			
HGMDFN717 <sup>1, 2, 5</sup> (replaces HGMDFN090)	Mother of HGADFN167	53 yrs 3 mos	Female	LMNA Exon 11, Negative	HGMLBV010 HGMDFSV40T090 iPSC lines			
HGMDFN718 <sup>1, 2, 6</sup>	Mother of HGADFN496 (proband line no longer offered)	42 yrs 0 mos	Female	LMNA Exon 11, Negative				

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

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

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



## 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 <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				
PSADFN086 <sup>2, 5</sup> (Cells grow poorly)	Proband	0 yrs 7 mos	Male	LMNA Intron 11, heterozygous c.1968+1 G>A	PSALBV083				
PSADFN576 <sup>2, 5</sup>	Proband	34 yrs 11 mos	Female	LMNA Exon 11/Intron 11 junction, heterozygous c. 1968+5G>A					

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

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

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



PSMDFN387<sup>1</sup>

PSFDFN388<sup>1</sup>

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

#### FIBROBLASTS: FAMILY SETS CONTAINING A FIBROBLAST CELL LINE WITH NON-CLASSIC HGPS MUTATION (PROGERIN-PRODUCING) **Other Lines From This** Relation to Cell Line # Age at Donation Gender **Mutational Analysis** Proband Donor Proband LMNA Exon 11, heterozygous (mild PSADFN4891,7 3 yrs 2 mos Female c.1824C>T (p.Gly608Gly); and progeroid SMC3 c.562A>G, p.K188E phenotype) Father of PSFDFN7141,7 38 yrs 6 mos Male Negative HGADFN489 Mother of PSMDFN7131,7 35 yrs 11 mos Female Negative HGADFN489 Mosaic: DNA from Fibroblasts: c.1968 + 2T > CDNA from Blood: 4.7% c.1968+2T>C mutation, and 41.3% c.1968+2T>A mutation PSADFN3861 Proband 11 mos Female 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. Journal of Medical Genetics 2017;54:212-Mother of

Female

Male

LMNA Exon 11, Negative

LMNA Exon 11, Negative

36 yrs 5 mos

38 yrs 1 mo

PSADFN386 Father of

PSADFN386

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

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

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



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

# 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 <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, 6</sup>	Father of HGADFN325	36 yrs 3 mos	Male	LMNA Exon 11/Intron 11 junction, negative	
PSMDFN326 <sup>2, 5, 6</sup>	Mother of HGADFN325	36 yrs 10 mos	Female	LMNA Exon 11/Intron 11 junction, negative	
PSADFN392 <sup>2, 5</sup>	Proband	7 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1968+2T>C	
PSFDFN394 <sup>1, 6</sup>	Father of PSADFN392	49 yrs 1 mo	Male	LMNA Exon 11, negative <sup>6</sup>	
PSMDFN393 <sup>1, 6</sup>	Mother of PSADFN392	44 yrs 8 mos	Female	LMNA Exon 11, negative <sup>6</sup>	
PSMDFN346 <sup>2, 5</sup>	Mother of PSADFN345 (proband line not available)	21 yrs 10 mos	Female	LMNA Exon 11, Negative	

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

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

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



# 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)						

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

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

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



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

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

FIBROBLAST CELL LINES (NOT PROGERIN-PRODUCING)							
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			
PSADFN363 <sup>1, 2, 5</sup>	Proband	8 mos	Male	LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn)			
PSFDFN365 <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			

<sup>&</sup>lt;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).

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

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

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



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

IMMORTALIZED FIBROBLAST CELL LINES								
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
PSADFSV40T317 <sup>1,7</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)	PSADFN317			
HGFDFSV40T168 <sup>1, 2, 5</sup>	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021 iPSC lines			
HGMDFSV40T090 <sup>1, 2, 5</sup>	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010 iPSC lines			
HGFDFSV40T369 <sup>1, 2, 5</sup>	Father of HGADFN367	33 yrs 9 mos	Male	Negative	HGFDFN369			
PSFDFSV40T376 <sup>1,7</sup>	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFDFN376 PSFLBV344			
HGMDFSV40T368 <sup>1, 2, 5</sup>	Mother of HGADFN367	31 yrs 7 mos	Female	Negative	HGMDFN368			

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

<sup>&</sup>lt;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.

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

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



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

LYMPHOBLASTS: CLASSIC MUTATION								
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
HGALBV039 <sup>3</sup>	Proband	3 yrs 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN127			
HGALBV071 <sup>3</sup>	Proband	15 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV073 <sup>3</sup>	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 iPSC lines			
HGALBV110 <sup>3</sup>	Proband	5 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN178			
HGALBV113 <sup>3</sup>	Proband	12 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV145 <sup>3</sup>	Proband	13 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV146 <sup>3</sup>	Proband	9 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV150 <sup>3</sup>	Proband	6 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV152 <sup>3</sup>	Proband	11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN188			
HGALBV162 <sup>3</sup>	Proband	1 yr 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV172 <sup>3</sup>	Proband	7 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				

<sup>&</sup>lt;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).

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

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

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



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

LYMPHOBLASTS: CLASSIC MUTATION										
	CONTINUED									
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor					
HGALBV186 <sup>3</sup>	Proband	1 yr 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV234 <sup>3</sup>	Proband	10 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV236 <sup>3</sup>	Proband	6 yrs 1 mo	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV237 <sup>3</sup>	Proband	14 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV240 <sup>3</sup>	Proband	4 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV255 <sup>3</sup>	Proband	10 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV331 <sup>3</sup>	Proband	10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV338 <sup>3</sup>	Proband	3 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV351 <sup>3</sup>	Proband	4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV378 <sup>3</sup>	Proband	6 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						

<sup>&</sup>lt;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).

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

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

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



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

LYMPHOBLASTS: CLASSIC MUTATION										
	CONTINUED									
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor					
HGALBV389 <sup>3</sup>	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV395 <sup>3</sup>	Proband	1 yr 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV408 <sup>3</sup>	Proband	2 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV419 <sup>3</sup>	Proband	1 yr 11 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV424 <sup>3</sup>	Proband	15 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV437 <sup>3</sup>	Proband	4 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV439 <sup>3</sup>	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV451 <sup>3</sup>	Proband	7 yrs 7 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV452 <sup>3</sup>	Proband	1 yr 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV462 <sup>3</sup>	Proband	6 yr 2 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						
HGALBV471 <sup>3</sup>	Proband	7 yr 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)						

<sup>&</sup>lt;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).

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

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

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



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

# LYMPHOBLASTS: FAMILY SETS CONTAINING A LYMPHOBLAST CELL LINE WITH THE CLASSIC HGPS MUTATION

WITH THE CLASSIC HOLD WOLATION								
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
HGALBV009 <sup>3</sup>	Proband	5 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 iPSC lines			
HGFLBV021 <sup>3</sup>	Father of HGALBV009	37 yrs 0 mos	Male	LMNA Exon 11, Negative	HGFDFN168 iPSC lines			
HGMLBV010 <sup>3</sup>	Mother of HGALBV009	36 yrs 11 mos	Female	LMNA Exon 11, Negative	HGMDFN090 iPSC lines			
HGALBV011 <sup>3</sup>	Proband	6 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN143			
HGFLBV031 <sup>3</sup>	Father of HGALBV011	36 yrs 5 mos	Male	LMNA Exon 11, Negative				
HGMLBV023 <sup>3</sup>	Mother of HGALBV011	33 yrs 8 mos	Female	LMNA Exon 11, Negative				
HGALBV016 <sup>3</sup>	Proband	15 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGMLBV017 <sup>3</sup>	Mother of HGALBV016	40 yrs	Female	Not performed				
HGSLBV019 <sup>3</sup>	Sibling of HGALBV016	19 yrs 9 mos	Male	Not performed				
HGALBV040 <sup>3</sup>	Proband	12 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGFLBV042 <sup>3</sup>	Father of HGALBV040	38 yrs	Male	Not performed				

<sup>&</sup>lt;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).

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

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

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



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

# LYMPHOBLASTS: FAMILY SETS CONTAINING A LYMPHOBLAST CELL LINE WITH THE CLASSIC HGPS MUTATION

#### **CONTINUED**

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
HGMLBV041 <sup>3</sup>	Mother of HGALBV040	43 yrs	Female	Not performed	
HGALBV055 <sup>3</sup>	Proband	7 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV065 <sup>3</sup>	Mother of HGALBV055	26 yrs 0 mo	Female	LMNA Exon 11, Negative	
HGALBV057 <sup>3</sup>	Proband	4 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGFLBV067 <sup>3</sup>	Father of HGALBV057	40 yrs 8 mos	Male	LMNA Exon 11, Negative	
HGMLBV066 <sup>3</sup>	Mother of HGALBV057	28 yrs 2 mos	Female	LMNA Exon 11, Negative	
HGALBV074 <sup>3</sup>	Proband	10 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV076 <sup>3</sup>	Mother of HGALBV074	42 yrs 8 mos	Female	Not performed	
HGSLBV075 <sup>3</sup>	Sibling of HGALBV074	12 yrs 6 mos	Female	Not performed	
HGSLBV077 <sup>3</sup>	Sibling of HGALBV074	16 yrs 10 mos	Male	Not performed	

<sup>&</sup>lt;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).

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

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

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



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

# LYMPHOBLASTS: FAMILY SETS CONTAINING A LYMPHOBLAST CELL LINE WITH THE CLASSIC HGPS MUTATION

#### **CONTINUED**

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
HGALBV097 <sup>3</sup>	Proband	2 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN122
HGFLBV099 <sup>3</sup>	Father of HGALBV097	51 yrs 6 mos	Male	Not performed	
HGMLBV098 <sup>3</sup>	Mother of HGALBV097	41 yrs 6 mos	Female	Not performed	
HGALBV132 <sup>3</sup>	Proband	1 yr 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV133 <sup>3</sup>	Mother of HGALBV132	24 yrs 7 mos	Female	LMNA Exon 11, Negative	
HGFLBV134 <sup>3</sup>	Father of HGALBV132	27 yrs 5 mos	Male	LMNA Exon 11, Negative	
HGALBV314 <sup>3</sup>	Proband	2 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGSLBV353 <sup>3</sup>	Sibling of HGALBV314	7 mos	Female	LMNA Exon 11, Negative	
HGSLBV359 <sup>3</sup>	Sibling of HGALBV314	7 mos	Male	LMNA Exon 11, Negative	

<sup>&</sup>lt;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).

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

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

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



# 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 PSALBV296<sup>3</sup> **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 PSALBV406<sup>3</sup> Proband Male 8 mos c.1822G>A (p.Gly608Ser) LMNA Exon 11, heterozygous PSALBV427<sup>3</sup> Proband 3 mos. Male c.1821G>A(p.V607V)

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

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

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



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

# 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 <sup>3</sup>	Proband	6 mos	Male	LMNA Exon 11, heterozygous c.1968+1 G>A	PSADFN086
PSFLBV084 <sup>3</sup>	Father of PSALBV083	31 yrs 9 mos	Male	Not performed	
PSMLBV085 <sup>3</sup>	Mother of PSALBV083	32 yrs 2 mos	Female	Not performed	

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

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

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



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

LYMPHOBLASTS: PROGEROID LAMINOPATHY MUTATION						
(NOT PROGERIN-PRODUCING)						
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor	
PSALBV432 <sup>3</sup>	Proband	4 yrs 5 mos	Male	LMNA Exon 9, homozygous c.1580G>T, (p.R527L)		

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

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

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



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

# LYMPHOBLASTS: FAMILY SETS CONTAINING LYMPHOBLAST CELL LINES WITH PRODEROID LAMINOPATHY (NON-PROGERIN PRODUCING)

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSALBV245 <sup>3</sup>	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 <sup>3</sup>	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 <sup>3</sup>	Mother of PSALBV245 & PSALBV339	~24 yrs	Female	LMNA Exon 9, heterozygous c.1579C>T, (p.Arg527Cys)	
PSFLBV239 <sup>3</sup>	Father of PSALBV245 & PSALBV339	~25 yrs	Male	LMNA Exon 9, heterozygous c.1579C>T, (p.Arg527Cys)	

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

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

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



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

# LYMPHOBLASTS: FAMILY SETS CONTAINING LYMPHOBLAST CELL LINES WITH PRODEROID LAMINOPATHY (NON-PROGERIN PRODUCING)

#### **CONTINUED**

Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSALBV295 <sup>3</sup>	Proband	17 yrs 3 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	PSADFN425
PSMLBV360 <sup>3</sup>	Mother of PSALBV295	46 yrs 1 mos	Female	LMNA Exon 1, Negative Intron 6, heterozygous c.1158-44 C>T	
PSFLBV361 <sup>3</sup>	Father of PSALBV295	49 yrs 8 mos	Male	LMNA Exon 1 & Intron 6, Negative	
PSALBV341 <sup>3</sup>	Proband (sibling of PSALBV342)	5 yrs 2 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSADFN373
PSALBV342 <sup>3</sup>	Proband (sibling of PSALBV341)	2 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	
PSMLBV343 <sup>3</sup>	Mother of PSALBV341 & PSALBV342	32 yrs 2 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMDFN375
PSFLBV344 <sup>3</sup>	Father of PSALBV341 & PSALBV342	32 yrs	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFDFN376

<sup>&</sup>lt;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).

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

<sup>&</sup>lt;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>&</sup>lt;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).

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

INDUCED PLURIPOTENT STEM CELL LINES					
Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
HGADFN003 iPS1B <sup>4</sup>	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN003 iPS1C <sup>4</sup>	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN003 iPS1D <sup>4</sup>	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN167 iPS1J <sup>4</sup>	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 HGALBV009
HGADFN167 iPS1Q <sup>4</sup>	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 HGALBV009
HGMDFN090 iPS1B <sup>4</sup>	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010
HGMDFN090 iPS1C <sup>4</sup>	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010
HGFDFN168 iPS1D2 <sup>4</sup>	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021
HGFDFN168 iPS1P <sup>4</sup>	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021

<sup>&</sup>lt;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).

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