

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

### Cell Lines Available

<b>FIBROBLAST CLASSIC MUTATION</b>						
<b>Cell Line #</b>	<b>Clinically Affected</b>	<b>Relation to Proband</b>	<b>Age at Donation</b>	<b>Gender</b>	<b>Mutational Analysis</b>	<b>Other Lines From This Donor</b>
HGADFN003	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV073 iPSC lines
HGADFN122	Yes	Proband	5 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV097
HGADFN127	Yes	Proband	3 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV039
HGADFN143	Yes	Proband	8 yrs 10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV011
HGADFN155	Yes	Proband	1 yr 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGADFN164	Yes	Proband	4 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGADFN167	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV009 HGADFSV40T167 iPSC lines
HGFDFN168	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFLBV021 HGFDFSV40T168 iPSC lines
HGMDFN090	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMLBV010 HGMDFSV40T090 iPSC lines
HGADFN169	Yes	Proband	8 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGADFN178	Yes	Proband	6 yrs 11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV110
HGADFN188	Yes	Proband	2 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGALBV152
HGADFN271	Yes	Proband	1 yr 3 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGADFN367	Yes	Proband	3 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMDFN368	No	Mother of HGADFN367	31 yrs 7 mos	Female	Not Performed	
HGFDFN369	No	Father of HGADFN367	33 yrs 9 mos	Male	Not performed	

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<b>FIBROBLAST NON-CLASSIC MUTATIONS</b>						
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PSADFN086* *Cells grow poorly	Yes	Proband	0 yrs 7 mos	Male	LMNA Exon 11, c.1968+1 G>A	PSALBV083
PSADFN257	Yes	Proband	1 yr 10 mos	Male	LMNA Exon 10, homozygous c.1619 T>C (p.Met540Thr)	
PSADFN325	Yes	Proband	6 yrs 9 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c.1968+5G>C	
PSMDFN326	No	Mother of HGADFN325	36 yrs 10 mos	Female	Not performed	
PSFDFN327	No	Father of HGADFN325	36 yrs 3 mos	Male	Not performed	
PSADFN328* *Cells grow poorly	Yes	Proband	12 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1822 G->A (p.Gly608Ser)	PSALBV296
PSMDFN346	No	Mother of PSADFN345 (proband line not available)	21 yrs 10 mos	Female	LMNA Exon 11, Negative	
PSADFN363	Yes	Proband	8 mos	Male	LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn)	
PSMDFN364	No	Mother of PSADFN363	36 yrs 10 mos	Female	Not performed	
PSFDFN365	No	Father of PSADFN363	44 yrs 2 mos	Male	Not performed	
PSADFN317	Yes	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	Yes	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	No	Father of PSADFN317 & PSADFN318	39 yrs 0 mo	Male	Not performed	
PSMDFN320	No	Mother of PSADFN317 & PSADFN318	36 yrs 8 mo	Female	Not performed	

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  
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### FIBROBLAST NON-CLASSIC MUTATIONS CONTINUED

PSADFN373	Yes	Proband	5 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSALBV341
PSMDFN375	No	Mother of PSADFN373	32 yrs 9 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMLBV343
PSFDFN376	No	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFLBV344
PSADFN392	Yes	Proband	7 yrs 4 mos	Male	LMNA Exon 11, c.1968+2T>C	
PSMDFN393	No	Mother of PSADFN392	44 yrs 8 mos	Female	Not performed	
PSFDFN394	No	Father of PSADFN392	49 yrs 1 mo	Male	Not performed	
PSADFN412	Yes	Proband	7 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	PSADFN414
PSADFN414	Yes	Proband	12 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	PSADFN412
PSADFN425	Yes	Proband	20 yrs 11 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	PSALBV295



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<b>IMMORTALIZED FIBROBLAST CELL LINES</b>						
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PSADFSV40T317	Yes	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
HGMDFSV40T090	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010 iPSC lines
HGFDFSV40T168	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021 iPSC lines
HGMDFSV40T368	No	Mother of HGADFN367	31 yrs 7 mos	Female	Not Performed	HGMDFN368
HGFDFSV40T369	No	Father of HGADFN367	33 yrs 9 mos	Male	Not performed	HGFDFN369
PSFDFSV40T376	No	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFDFN376 PSFLBV344



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<b>LYMPHOBLAST CLASSIC MUTATION</b>						
<b>Cell Line #</b>	<b>Clinically Affected</b>	<b>Relation to Proband</b>	<b>Age at Donation</b>	<b>Gender</b>	<b>Mutational Analysis</b>	<b>Other Lines From This Donor</b>
HGALBV009	Yes	Proband	5 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 iPSC lines
HGMLBV010	No	Mother of HGALBV009	36 yrs 11 mos	Female	LMNA Exon 11, Negative	HGMDFN090 iPSC lines
HGFLBV021	No	Father of HGALBV009	37 yrs 0 mos	Male	LMNA Exon 11, Negative	HGFDFN168 iPSC lines
HGALBV011	Yes	Proband	6 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN143
HGMLBV023	No	Mother of HGALBV011	33 yrs 8 mos	Female	LMNA Exon 11, Negative	
HGFLBV031	No	Father of HGALBV011	36 yrs 5 mos	Male	LMNA Exon 11, Negative	
HGALBV016	Yes	Proband	15 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV017	No	Mother of HGALBV016	40 yrs	Female	Not performed	
HGSLBV019	No	Sibling of HGALBV016	19 yrs 9 mos	Male	Not performed	
HGALBV039	Yes	Proband	3 yrs 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN127
HGALBV040	Yes	Proband	12 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV041	No	Mother of HGALBV040	43 yrs	Female	Not performed	
HGFLBV042	No	Father of HGALBV040	38 yrs	Male	Not performed	
HGALBV055	Yes	Proband	7 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV065	No	Mother of HGALBV055	26 yrs 0 mo	Female	LMNA Exon 11, Negative	
HGALBV057	Yes	Proband	4 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV066	No	Mother of HGALBV057	28 yrs 2 mos	Female	LMNA Exon 11, Negative	
HGFLBV067	No	Father of HGALBV057	40 yrs 8 mos	Male	LMNA Exon 11, Negative	
HGALBV071	Yes	Proband	15 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	

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### LYMPHOBLAST CLASSIC MUTATION CONTINUED

HGALBV073	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 iPSC lines
HGALBV074	Yes	Proband	10 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV076	No	Mother of HGALBV074	42 yrs 8 mos	Female	Not performed	
HGSLBV075	No	Sibling of HGALBV074	12 yrs 6 mos	Female	Not performed	
HGSLBV077	No	Sibling of HGALBV074	16 yrs 10 mos	Male	Not performed	
HGALBV097	Yes	Proband	2 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN122
HGMLBV098	No	Mother of HGALBV097	41 yrs 6 mos	Female	Not Performed	
HGFLBV099	No	Father of HGALBV097	51 yrs 6 mos	Male	Not Performed	
HGALBV110	Yes	Proband	5 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN178
HGALBV113	Yes	Proband	12 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV132	Yes	Proband	1 yr 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGMLBV133	No	Mother of HGALBV132	24 yrs 7 mos	Female	LMNA Exon 11, Negative	
HGFLBV134	No	Father of HGALBV132	27 yrs 5 mos	Male	LMNA Exon 11, Negative	
HGALBV145	Yes	Proband	13 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV146	Yes	Proband	9 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV150	Yes	Proband	6 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV152	Yes	Proband	11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN188
HGALBV162	Yes	Proband	1 yr 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV172	Yes	Proband	7 yrs 9 mos	Female	LMNA Exon 11, c.1824C>T (p.Gly608Gly)	
HGALBV186	Yes	Proband	1 yr 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV236	Yes	Proband	6 yrs 1 mo	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV237	Yes	Proband	14 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	

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HGALBV240	Yes	Proband	4 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV255	Yes	Proband	10 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV304	Yes	Proband	12 yrs 9 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV314	Yes	Proband	2 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGSLBV353	No	Sibling of HGALBV314	7 mos	Female	LMNA Exon 11, Negative	
HGSLBV359	No	Sibling of HGALBV314	7 mos	Male	LMNA Exon 11, Negative	
HGALBV331	Yes	Proband	10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV338	Yes	Proband	3 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV351	Yes	Proband	4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV378	Yes	Proband	6 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV389	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV234	Yes	Proband	10 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV395	Yes	Proband	1 yr 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV408	Yes	Proband	2 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV419	Yes	Proband	1 yr 11 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV424	Yes	Proband	15 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV437	Yes	Proband	4 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV439	Yes	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV451	Yes	Proband	7 yrs 7 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV452	Yes	Proband	1 yr 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV462	Yes	Proband	6 yr 2 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	
HGALBV471	Yes	Proband	7 yr 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	



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<b>Cell Line #</b>	<b>Clinically Affected</b>	<b>Relation to Proband</b>	<b>Age at Donation</b>	<b>Gender</b>	<b>Mutational Analysis</b>	<b>Other Lines From This Donor</b>
PSALBV083	Yes	Proband	6 mos	Male	LMNA Exon 11, c.1968+1 G>A	PSADFN086
PSFLBV084	No	Father of PSALBV083	31 yrs 9 mos	Male	Not Performed	
PSMLBV085	No	Mother of PSALBV083	32 yrs 2 mos	Female	Not Performed	
PSALBV199	Yes	Proband	11 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1868 C>G (p.Thr623Ser)	
PSMLBV238	No	Mother of PSALBV245 & PSALBV339	~24 yrs	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSFLBV239	No	Father of PSALBV245 & PSALBV339	~25 yrs	Male	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSALBV245	Yes	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	No	Sibling of PSALBV245(2 deceased siblings were homozygous for same mutation)	1 mos	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)	
PSALBV229	Yes	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	
PSALBV295	Yes	Proband	17 yrs 3 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	PSADFN425
PSMLBV360	No	Mother of PSALBV295	46 yrs 1 mos	Female	LMNA Exon 1, Negative Intron 6, heterozygous c.1158-44 C>T	
PSFLBV361	No	Father of PSALBV295	49 yrs 8 mos	Male	LMNA Exon 1 & Intron 6, Negative	
PSALBV296	Yes	Proband	10 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	PSADFN328



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### LYMPHOBLAST NON-CLASSIC MUTATIONS CONTINUED

PSALBV379	Yes	Proband	5 yrs 3 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c.1968+1G>A	
PSALBV341	Yes	Proband (and sibling of PSALBV342)	5 yrs 2 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSADFN373
PSALBV342	Yes	Proband (and sibling of PSALBV341)	2 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	
PSMLBV343	No	Mother of PSALBV341 & PSALBV342	32 yrs 2 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMDFN375
PSFLBV344	No	Father of PSALBV341 & PSALBV342	32 yrs	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFDFN376
PSALBV406	Yes	Proband	8 mos	Male	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)	
PSALBV427	Yes	Proband	3 mos.	Male	LMNA Exon 11, heterozygous c.1821G>A(p.V607V)	
PSALBV432	Yes	Proband	4 yrs 5 mos	Male	LMNA Exon 9, homozygous c.1580G>T, (p.A527L)	



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<b>INDUCED PLURIPOTENT STEM CELLS</b>						
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HGADFN003 iPS1B	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN003 iPS1C	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN003 iPS1D	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN003 HGALBV073
HGADFN167 iPS1J	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 HGALBV009
HGADFN167 iPS1Q	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	HGADFN167 HGALBV009
HGMDFN090 iPS1B	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010
HGMDFN090 iPS1C	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	HGMDFN090 HGMLBV010
HGFDFN168 iPS1D2	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021
HGFDFN168 iPS1P	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	HGFDFN168 HGFLBV021