

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

LYMPHOBLAST NON-CLASSIC MUTATIONS

Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
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

DF = Dermal Fibroblast
 LBV=Lymphoblast
 iPSC = Induced Pluripotent Stem Cell
 Cost of each DF & LBV cell line is \$40.25
 Cost of each Immortalized Fibroblast cell line is \$40.25
 Cost of each iPSC line is \$84.00

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)	