

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

Cell Lines Available

FIBROBLAST NON-CLASSIC MUTATIONS						
Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
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	

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