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Hutchinson-Gilford Progeria Syndrome and Progeroid Laminopathies
Cell Lines Available

FIBROBLAST NON-CLASSIC MUTATIONS					
PSADFN485[5]	Proband	4 yrs 5 mos	Male	LMNA heterozygous c.412G>A; (p.E138K)	
PSADFN425[1, 5]	Proband	20 yrs 11 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	PSALBV295
PSADFN414[1, 2, 5]	Proband	12 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	PSADFN412
PSADFN412[1, 2, 5]	Proband	7 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	PSADFN414
PSADFN328[1, 2, 5] (Cells grow poorly)	Proband	12 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1822 G->A (p.Gly608Ser)	PSALBV296
PSADFN257[2, 5]	Proband	1 yr 10 mos	Male	LMNA Exon 10, homozygous c.1619 T>C (p.Met540Thr)	
PSADFN086[2, 5] (Cells grow poorly)	Proband	0 yrs 7 mos	Male	LMNA Exon 11, c.1968+1 G>A	PSALBV083

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[2]Histograms of mutational analysis sequenced by the PRF Cell and Tissue Bank available.
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FAMILY NON-CLASSIC FIBROBLAST CELL LINES					
Cell Line #	**Relation to Proband**	**Age at Donation**	**Gender**	**Mutational Analysis[2]**	**Other Lines From This Donor**
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[1]	Father of PSADFN392	49 yrs 1 mo	Male	Not performed	
PSMDFN393[1]	Mother of PSADFN392	44 yrs 8 mos	Female	Not performed	
PSADFN373[1]	Proband	5 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	PSALBV341
PSFDFN376[1]	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSFLBV344
PSMDFN375[1]	Mother of PSADFN373	32 yrs 9 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	PSMLBV343
PSADFN363[1, 2, 5]	Proband	8 mos	Male	LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn)	
PSFDFN365[1, 2, 5]	Father of PSADFN363	44 yrs 2 mos	Male	Not performed	
PSMDFN364[1, 2, 5]	Mother of PSADFN363	36 yrs 10 mos	Female	Not performed	

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FAMILY NON-CLASSIC FIBROBLAST CELL LINES CONTINUED					
Cell Line #	**Relation to Proband**	**Age at Donation**	**Gender**	**Mutational Analysis**	**Other Lines From This Donor**
PSMDFN346[2, 5]	Mother of PSADFN345 (proband line not available)	21 yrs 10 mos	Female	LMNA Exon 11, Negative	
PSADFN325[1, 2, 5]	Proband	6 yrs 9 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c. 1968+5G>C	
PSFDFN327[2, 5]	Father of HGADFN325	36 yrs 3 mos	Male	Not performed	
PSMDFN326[2, 5]	Mother of HGADFN325	36 yrs 10 mos	Female	Not performed	
PSADFN317[5]	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[1, 5]	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[1]	Mother of PSADFN317 & PSADFN318	36 yrs 8 mo	Female	Not performed	

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Cell Line #	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor
PSADFN386[1]	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. Journal of Medical Genetics 2017;54:212-216	
PSMDFN387[1]	Mother of PSADFN386	36 yrs 5 mos	Female	LMNA Exon 11, Negative	
PSFDFN388[1]	Father of PSADFN386	38 yrs 1 mo	Male	LMNA Exon 11, Negative	

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