

**The Progeria Research Foundation Cell and Tissue Bank
 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 |
| PSADFN412 ^{1,2,5} | Proband | 7 yrs 1 mo | Male | LMNA Exon 11, heterozygous c.1762T>C (p.C588R) | |
| 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 |

¹Representative 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).

²Histograms of mutational analysis sequenced by the PRF Cell and Tissue Bank available.

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⁵Genetic 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.

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| FAMILY NON-CLASSIC FIBROBLAST CELL LINES | | | | | |
|---|------------------------------------|------------------------|---------------|--|------------------------------------|
| Cell Line # | Relation to Proband | Age at Donation | Gender | Mutational Analysis² | 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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| 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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|------------------------|---------------------|-----------------|--------|---|-----------------------------|
| 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. <i>Journal of Medical Genetics</i> 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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