

DF=Dermal Fibroblast
LBV=Lymphoblast
iPSC = Induced Pluripotent Stem Cell
Cost of each DF & LBV cell line is \$150.00
Cost of each Immortalized Fibroblast cell line is \$150.00
Cost of each iPSC line is \$500.00

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

| FIBROBLASTS: PROGEROID LAMINOPATHIES (NOT PROGERIN-PRODUCING) | | | | | | | | |
|---|------------------------|--------------------|--------|---|--------------------------------|--|--|--|
| Cell Line # | Relation to Proband | Age at Donation | Gender | Mutational Analysis | Other Lines From This Donor | | | |
| PSADFN485 ^{1,7} | Proband | 4 yrs 5 mos | Male | LMNA heterozygous c.412G>A; (p.E138K) | | | | |
| PSADFN425 ^{1,7} | 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) | | | | |
| PSADFN257 ^{2, 5} | Proband | 1 yr 10 mos | Male | LMNA Exon 10, homozygous c.1619 T>C (p.Met540Thr) | | | | |
| PSADFN542 ⁶ | Proband | 75 yrs 2 mos | Male | LMNA Exon 11, heterozygous c.1930 C>T (p.Arg644Cys) | | | | |

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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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.

³Representative cultures from this cell line have tested negative for mycoplasma contamination at Rutgers University Cell and DNA Repository via real time PCR assay.

⁴Representative cultures from this cell line have tested negative for mycoplasma contamination at Ottawa Hospital Research Institute. Please note: mycoplasma testing is performed on random passages. As part of our on-going quality control, The Human Pluripotent Stem Cell Facility/Dr. William Stanford laboratory periodically tests the cultures for mycoplasma contamination using a PCR based approach (Detection of mycoplasma contaminations., Uphoff CC, Drexler HG., Methods Mol Biol. 2013;946:1-13. doi: 10.1007/978-1-62703-128-8_1. PMID:23179822).

⁶Mutational analysis was performed on fibroblasts only, not on DNA derived from blood

⁷Cell line has not been tested for the mutation(s). Mutational analysis is based on blood DNA.



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FIBROBLASTS: FAMILY SETS CONTAINING PROGEROID LAMINOPATHY FIBROBLAST CELL LINES (NOT PROGERIN-PRODUCING) **Other Lines** Relation to Age at Cell Line # Gender **Mutational Analysis** From This Donation **Proband** Donor ZMPste24 Exon 10, homozygous PSADFN3731,7 Proband 5 yrs 9 mos Male PSALBV341 c.1274T>C (p.Leu425Pro) ZMPste24 Exon 10, heterozygous Father of PSFDFN3761,7 32 yrs 6 mos Male PSFLBV344 PSADFN373 c.1274T>C (p.Leu425Pro) Mother of ZMPste24 Exon 10, heterozygous PSMDFN375^{1,7} 32 yrs 9 mos Female PSMLBV343 PSADFN373 c.1274T>C (p.Leu425Pro) Proband (& ZMPste24 Exon 6, heterozygous c.743C>T(p.Pro248Leu); Exon 10, PSADFN317⁷ sibling of 3 yr 9 mo Male PSADFN318) heterozygous c.1349G>A (p.Trp450Stop) Proband (& ZMPste24 Exon 6, heterozygous PSADFN3181, 7 c.743C>T(p.Pro248Leu); Exon 10, sibling of 5 mos Male PSADFN317) heterozygous c.1349G>A (p.Trp450Stop) Father of PSFDFN319 PSADFN317 & 39 yrs 0 mo Male Not performed PSADFN318 Mother of PSMDFN3201 PSADFN317 & 36 yrs 8 mo Not performed Female PSADFN318

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FIBROBLASTS: FAMILY SETS CONTAINING PROGEROID LAMINOPATHY FIBROBLAST CELL LINES (NOT PROGERIN-PRODUCING)

CONTINUED

| Cell Line # | Relation to Proband | Age at Donation | Gender | Mutational Analysis | Other Lines From This Donor |
|------------------------------|------------------------|--------------------|--------|---|-----------------------------------|
| PSADFN363 ^{1, 2, 5} | Proband | 8 mos | Male | LMNA Exon 6, heterozygous c.973G>A (p.Asp325Asn) | |
| PSFDFN365 ^{1, 2, 6} | Father of PSADFN363 | 44 yrs 2 mos | Male | LMNA Exon 6, negative | |
| PSMDFN364 ^{1, 2, 6} | Mother of PSADFN363 | 36 yrs 10 mos | Female | LMNA Exon 6, negative | |

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