

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

**LYMPHOBLAST NON-CLASSIC MUTATION**

| Cell Line #            | Relation to Proband | Age at Donation | Gender | Mutational Analysis                                       | Other Lines From This Donor |
|------------------------|---------------------|-----------------|--------|---|-----------------------------|
| PSALBV199 <sup>3</sup> | Proband             | 11 yrs 3 mos    | Female | LMNA Exon 11, heterozygous c.1868 C>G (p.Thr623Ser)       |                             |
| PSALBV229 <sup>3</sup> | Proband             | 5 yrs 9 mos     | Female | LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)        |                             |
| PSALBV296 <sup>3</sup> | Proband             | 10 yrs 8 mos    | Female | LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)        | PSADFN328                   |
| PSALBV379 <sup>3</sup> | Proband             | 5 yrs 3 mos     | Male   | LMNA Exon 11/Intron 11 junction, heterozygous c.1968+1G>A |                             |
| PSALBV406 <sup>3</sup> | Proband             | 8 mos           | Male   | LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)        |                             |
| PSALBV427 <sup>3</sup> | Proband             | 3 mos.          | Male   | LMNA Exon 11, heterozygous c.1821G>A(p.V607V)             |                             |
| PSALBV432 <sup>3</sup> | Proband             | 4 yrs 5 mos     | Male   | LMNA Exon 9, homozygous c.1580G>T, (p.A527L)              |                             |

<sup>1</sup>Representative 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).

<sup>2</sup>Histograms of mutational analysis sequenced by the PRF Cell and Tissue Bank available.

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

<sup>4</sup>Representative 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).

<sup>5</sup>Genetic 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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| LYMPHOBLAST NON-CLASSIC FAMILY CELL LINES |   |              |        |   |           |
|---|---|--------------|--------|---|-----------|
| PSALBV083 <sup>3</sup>                    | Proband   | 6 mos        | Male   | LMNA Exon 11, c.1968+1 G>A  | PSADFN086 |
| PSFLBV084 <sup>3</sup>                    | Father of PSALBV083   | 31 yrs 9 mos | Male   | Not performed   |           |
| PSMLBV085 <sup>3</sup>                    | Mother of PSALBV083   | 32 yrs 2 mos | Female | Not performed   |           |
| PSALBV245 <sup>3</sup>                    | 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 <sup>3</sup>                    | Proband, Sibling of PSALBV245 (2 deceased siblings were homozygous for same mutation) | 1 mos        | Female | LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)                          |           |
| PSMLBV238 <sup>3</sup>                    | Mother of PSALBV245 & PSALBV339   | ~24 yrs      | Female | LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)                          |           |
| PSFLBV239 <sup>3</sup>                    | Father of PSALBV245 & PSALBV339   | ~25 yrs      | Male   | LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)                          |           |
| PSALBV295 <sup>3</sup>                    | Proband   | 17 yrs 3 mos | Male   | LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys)<br>Intron 6, 1158-44 C>T | PSADFN425 |
| PSMLBV360 <sup>3</sup>                    | Mother of PSALBV295   | 46 yrs 1 mos | Female | LMNA Exon 1, Negative<br>Intron 6, heterozygous c.1158-44 C>T             |           |
| PSFLBV361 <sup>3</sup>                    | Father of PSALBV295   | 49 yrs 8 mos | Male   | LMNA Exon 1 & Intron 6, Negative  |           |

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**LYMPHOBLAST NON-CLASSIC FAMILY CELL LINES CONTINUED**

| Cell Line #            | Relation to Proband             | Age at Donation | Gender | Mutational Analysis                                    | Other Lines From This Donor |
|------------------------|---------------------------------|-----------------|--------|--|-----------------------------|
| PSALBV341 <sup>3</sup> | Proband (sibling of PSALBV342)  | 5 yrs 2 mos     | Male   | ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)   | PSADFN373                   |
| PSALBV342 <sup>3</sup> | Proband (sibling of PSALBV341)  | 2 yrs 9 mos     | Male   | ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)   |                             |
| PSMLBV343 <sup>3</sup> | Mother of PSALBV341 & PSALBV342 | 32 yrs 2 mos    | Female | ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro) | PSMDFN375                   |
| PSFLBV344 <sup>3</sup> | Father of PSALBV341 & PSALBV342 | 32 yrs          | Male   | ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro) | PSFDFN376                   |

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