

### **Cell Lines Available**

FIBROBLAST CLASSIC MUTATION								
Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor		
*Line is no longer available due to low stock; if line is needed for research with matching iPSC lines, please email Susan Campbell	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV073 & iPSC lines		
HGADFN122	Yes	Proband	5 yrs `0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV097		
HGADFN127	Yes	Proband	3 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV039		
HGADFN143	Yes	Proband	8 yrs 10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV011		
HGADFN155	Yes	Proband	1 yr 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGADFN164	Yes	Proband	4 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGADFN167	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV009 & iPSC lines		
HGFDFN168	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	LBV HGFLBV021 & iPSC lines		
HGMDFN090	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	LBV HGMLBV010 & iPSC lines		
HGADFN169	Yes	Proband	8 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGADFN178	Yes	Proband	6 yrs 11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV110		
HGADFN188	Yes	Proband	2 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	LBV HGALBV152		
HGADFN271	Yes	Proband	1 yr 3 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGADFN367	Yes	Proband	3 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGMDFN368	No	Mother of HGADFN367	31 yrs 7 mos	Female	Not Performed			
HGFDFN369	No	Father of HGADFN367	33 yrs 9 mos	Male	Not performed			

Version Date: 3/6/15 Page **1** of **10** 

DF = Dermal Fibroblast LBV=Lymphoblast iPSC = Induced Pluripotent Stem Cell Cost of each DF & LBV cell line is \$40.25 Cost of each iPSC line is \$84.00

FIBROBLAST CLASSIC MUTATIONS CONTINUED								
HGADFN370	Yes	Proband	10 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGMDFN371	No	Mother of HGADFN370	45 yrs 7 mos	Female	Not performed			
HGFDFN372	No	Father of HGADFN370	44 yrs 6 mos	Male	Not performed			

Version Date: 3/6/15 Page **2** of **10** 



### **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	LBV 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)	LBV 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				
PSADNF317	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)				

Version Date: 3/6/15 Page **3** of **10** 

	FIBROBLAST NON-CLASSIC MUTATIONS CONTINUED								
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				
PSADFN373	Yes	Proband	5 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	LBV PSALBV341			
PSMDFN375	No	Mother of PSADFN373	32 yrs 9 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	LBV PSMLBV343			
PSFDFN376	No	Father of PSADFN373	32 yrs 6 mos	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	LBV 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)	DF PSADFN414			
PSADFN414	Yes	Proband	12 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1762T>C (p.C588R)	DF PSADFN412			
PSADFN425	Yes	Proband	20 yrs 11 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T	LBV PSALBV295			

Version Date: 3/6/15 Page **4** of **10** 



### **Cell Lines Available**

LYMPHOBLAST CLASSIC MUTATION									
Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
HGALBV009	Yes	Proband	5 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN167 & iPSC lines			
HGMLBV010	No	Mother of HGALBV009	36 yrs 11 mos	Female	LMNA Exon 11, Negative	DF HGMDFN090 & iPSC lines			
HGFLBV021	No	Father of HGALBV009	37 yrs 0 mos	Male	LMNA Exon 11, Negative	DF HGFDFN168 & iPSC lines			
HGALBV011	Yes	Proband	6 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN143			
HGMLBV023	No	Mother of HGALBV011	33 yrs 8 mos	Female	LMNA Exon 11, Negative				
HGFLBV031	No	Father of HGALBV011	36 yrs 5 mos	Male	LMNA Exon 11, Negative				
HGALBV016	Yes	Proband	15 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGMLBV017	No	Mother of HGALBV016	40 yrs	Female	Not performed				
HGSLBV019	No	Sibling of HGALBV016	19 yrs 9 mos	Male	Not performed				
HGALBV039	Yes	Proband	3 yrs 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN127			
HGALBV040	Yes	Proband	12 yrs 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGMLBV041	No	Mother of HGALBV040	43 yrs	Female	Not performed				
HGFLBV042	No	Father of HGALBV040	38 yrs	Male	Not performed				
HGALBV055	Yes	Proband	7 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGMLBV065	No	Mother of HGALBV055	26 yrs 0 mo	Female	LMNA Exon 11, Negative				
HGALBV057	Yes	Proband	4 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGMLBV066	No	Mother of HGALBV057	28 yrs 2 mos	Female	LMNA Exon 11, Negative				
HGFLBV067	No	Father of HGALBV057	40 yrs 8 mos	Male	LMNA Exon 11, Negative				
HGALBV071	Yes	Proband	15 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				

Version Date: 3/6/15 Page **5** of **10** 

LYMPHOBLAST CLASSIC MUTATION CONTINUED								
HGALBV073	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN003 & iPSC lines		
HGALBV074	Yes	Proband	10 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGMLBV076	No	Mother of HGALBV074	42 yrs 8 mos	Female	Not performed			
HGSLBV075	No	Sibling of HGALBV074	12 yrs 6 mos	Female	Not performed			
HGSLBV077	No	Sibling of HGALBV074	16 yrs 10 mos	Male	Not performed			
HGALBV097	Yes	Proband	2 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN122		
HGMLBV098	No	Mother of HGALBV097	41 yrs 6 mos	Female	Not Performed			
HGFLBV099	No	Father of HGALBV097	51 yrs 6 mos	Male	Not Performed			
HGALBV110	Yes	Proband	5 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN178		
HGALBV113	Yes	Proband	12 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV132	Yes	Proband	1 yr 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGMLBV133	No	Mother of HGALBV132	24 yrs 7 mos	Female	LMNA Exon 11, Negative			
HGFLBV134	No	Father of HGALBV132	27 yrs 5 mos	Male	LMNA Exon 11, Negative			
HGALBV145	Yes	Proband	13 yrs 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV146	Yes	Proband	9 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV150	Yes	Proband	6 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV152	Yes	Proband	11 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN188		
HGALBV162	Yes	Proband	1 yr 6 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV172	Yes	Proband	7 yrs 9 mos	Female	LMNA Exon 11, c.1824C>T (p.Gly608Gly)			
HGALBV186	Yes	Proband	1 yr 0 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV236	Yes	Proband	6 yrs 1 mo	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			
HGALBV237	Yes	Proband	14 yrs 1 mo	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)			

Version Date: 3/6/15 Page **6** of **10** 

	LYMPHOBLAST CLASSIC MUTATION CONTINUED								
HGALBV240	Yes	Proband	4 yrs 2 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV255	Yes	Proband	10 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV304	Yes	Proband	12 yrs 9 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV314	Yes	Proband	2 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGSLBV353	No	Sibling of HGALBV314	7 mos	Female	LMNA Exon 11, Negative				
HGSLBV359	No	Sibling of HGALBV314	7 mos	Male	LMNA Exon 11, Negative				
HGALBV331	Yes	Proband	10 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV338	Yes	Proband	3 yrs 4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV351	Yes	Proband	4 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV378	Yes	Proband	6 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV389	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV234	Yes	Proband	10 yrs 7 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV395	Yes	Proband	1 yr 6 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV408	Yes	Proband	2 yrs 8 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV419	Yes	Proband	1 yr 11 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV424	Yes	Proband	15 yrs 5 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV437	Yes	Proband	4 yrs 4 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				
HGALBV439	Yes	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)				

Version Date: 3/6/15 Page **7** of **10** 



### **Cell Lines Available**

	LYMPHOBLAST NON-CLASSIC MUTATIONS								
Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
PSALBV083	Yes	Proband	6 mos	Male	LMNA Exon 11, c.1968+1 G>A	DF PSADFN086			
PSFLBV084	No	Father of PSALBV083	31 yrs 9 mos	Male	Not Performed				
PSMLBV085	No	Mother of PSALBV083	32 yrs 2 mos	Female	Not Performed				
PSALBV199	Yes	Proband	11 yrs 3 mos	Female	LMNA Exon 11, heterozygous c.1868 C>G (p.Thr623Ser)				
PSMLBV238	No	Mother of PSALBV245 & PSALBV339	~24 yrs	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)				
PSFLBV239	No	Father of PSALBV245 & PSALBV339	~25 yrs	Male	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)				
PSALBV245	Yes	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	No	Sibling of PSALBV245(2 deceased siblings were homozygous for same mutation)	1 mo	Female	LMNA Exon 9, heterozygous c.1579C>T(p.Arg527Cys)				
PSALBV229	Yes	Proband	5 yrs 9 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)				
PSALBV295	Yes	Proband	17 yrs 3 mos	Male	LMNA Exon 1, heterozygous c.331G>A (p.Glu111Lys) Intron 6,1158-44 C>T				
PSMLBV360	No	Mother of PSALBV295	46 yrs 1 mo	Female	LMNA Exon 1, Negative Intron 6, heterozygous c.1158-44 C>T				
PSFLBV361	No	Father of PSALBV295	49 yrs 8 mos	Male	LMNA Exon 1 & Intron 6, Negative				
PSALBV296	Yes	Proband	10 yrs 8 mos	Female	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)				

Version Date: 3/6/15 Page **8** of **10** 

LYMPHOBLAST NON-CLASSIC MUTATIONS CONTINUED								
PSALBV379	Yes	Proband	5 yrs 3 mos	Male	LMNA Exon 11/Intron 11 junction, heterozygous c.1968+1G>A			
PSALBV341	Yes	Proband (and sibling of PSALBV342)	5 yrs 2 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)	DF PSADFN373		
PSALBV342	Yes	Proband (and sibling of PSALBV341)	2 yrs 9 mos	Male	ZMPste24 Exon 10, homozygous c.1274T>C (p.Leu425Pro)			
PSMLBV343	No	Mother of PSALBV341 & PSALBV342	32 yrs 2 mos	Female	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	DF PSMDFN375		
PSFLBV344	No	Father of PSALBV341 & PSALBV342	32 yrs	Male	ZMPste24 Exon 10, heterozygous c.1274T>C (p.Leu425Pro)	DF PSFDFN376		
PSALBV406	Yes	Proband	8 mos	Male	LMNA Exon 11, heterozygous c.1822G>A (p.Gly608Ser)			
PSALBV427	Yes	Proband	3 mos.	Male	LMNA Exon 11, heterozygous c.1821G>A(p.V607V)			
PSALBV432	Yes	Proband	4 yrs 5 mos	Male	LMNA Exon 9, homozygous c.1580G>T, (p.A527L)			

Version Date: 3/6/15 Page **9** of **10** 



### **Cell Lines Available**

	INDUCED PLURIPOTENT STEM CELLS								
Cell Line #	Clinically Affected	Relation to Proband	Age at Donation	Gender	Mutational Analysis	Other Lines From This Donor			
HGADFN003 iPS1B	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN003; LBV HGALBV073			
HGADFN003 iPS1C	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN003; LBV HGALBV073			
HGADFN003 iPS1D	Yes	Proband	2 yrs 0 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN003; LBV HGALBV073			
HGADFN167 iPS1J	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN167; LBV HGALBV009			
HGADFN167 iPS1Q	Yes	Proband	8 yrs 5 mos	Male	LMNA Exon 11, heterozygous c.1824C>T (p.Gly608Gly)	DF HGADFN167; LBV HGALBV009			
HGMDFN090 iPS1B	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	DF HGMDFN090; LBV HGMLBV010			
HGMDFN090 iPS1C	No	Mother of HGADFN167	37 yrs 10 mos	Female	LMNA Exon 11, Negative	DF HGMDFN090; LBV HGMLBV010			
HGFDFN168 iPS1D2	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	DF HGFDFN168; LBV HGFLBV021			
HGFDFN168 iPS1P	No	Father of HGADFN167	40 yrs 5 mos	Male	LMNA Exon 11, Negative	DF HGFDFN168; LBV HGFLBV021			

Version Date: 3/6/15 Page **10** of **10**