

Online-Only Supplemental Material

eTable 1: Characteristics of fibroblasts used.

eTable 2: Primer sequences and taqman MGB probes used for quantitative real time PCR.

eTable 1: Characteristics of fibroblasts used.

Sample ID	Gender	Age ^a	Biopsy source	ABCC6 genotype ^b		Genotype status
PXE patients						
P3M ^c	male	57	Neck	c.3421C>T (p.(Arg1141*))	c.3883-6G>A (SSM)	cht
P128M ^c	male	51	Neck	c.3769_3770insC (p.L1259fsX1277)	c.3769_3770insC (p.L1259fsX1277)	hm
P255F ^c	female	48	Arm	c.3421C>T (p.(Arg1141*))	c.2787+1G>T (SSM)	cht
Healthy controls						
M57A ^d (AG13145)	male	57	Arm	-	-	wt
M52A ^d (AG11482)	male	52	Arm	-	-	wt
F48A ^d (AG14284)	female	48	Arm	-	-	wt

Note. hm, homozygous; cht, compound heterozygous; ht, heterozygous; wt, wild type; SSM, splice site mutation

^a Age in years.

^b Nucleotide numbering refers to the cDNA sequence with the A of the ATG translation initiation start site as nucleotide +1 (GenBank accession number NM_001171.2).

^c Fibroblasts isolated from skin biopsies(1).

^d Fibroblasts purchased from Coriell Institute for Medical Research (Camden, USA).

eTable 2: Primer sequences and taqman MGB probes used for quantitative real time PCR.

Gene	Protein	5'-3' sequence	Reference ^a	Annealing temperature (°C)	Efficiency
ACTB <i>beta-Actin</i>	β-Actin	CGCGAGAAGATGACCC ATTGCCAATGGTGATGAC	NM_001101	59°C	2.0
GAPDH <i>glyceraldehyd-3-phosphat-dehydrogenase</i>	GAPDH	AGGTCGGAGTCAACGGAT TCCTGGAAGATGGTGATG	NM_002046	59°C	1.8
B2M <i>beta-2-microglobulin</i>	β2M	TGTGCTCGCGCTACTCTCTCTT CGGATGGATGAAACCCAGACA	NM_004048	59°C	2.0
NCL <i>nucleolin</i>	nucleolin	AAGGCACAGAACCGACTACG TCTCGCATCTCGCTCTTTCT	NM_005381.3	63°C	2.0
FNTB <i>Protein farnesyltransferase subunit beta</i>	FNTB	ATGCGCGTTGTTGCTTAACG TCTGCAAAACAAGCCTTGGTACA	NM_002028.3	66°C	2.0
ZMPSTE24 <i>Zinc metalloproteinase STE24</i>	ZMPSTE24	ACCACCGGAGTTAGGACAGA CCAGGGACTGAGTGATCTCAT	NM_005857.5	66°C	1.9
RPLPO <i>Large Ribosomal Protein</i>	Large Ribosomal Protein			60°C	1.9
GUSB <i>β-Glucuronidase</i>	β-Glucuronidase			60°C	1.8
LMNA^b <i>lamin A</i>	lamin A	CTTCTGCCTCCAGTGTCACG AGTTCTGGGGGCTCTGGGT probe: ACTCGCAGCTACCG	NM_170707.3	60°C	2.0
LMNA^b <i>lamin C</i>	lamin C	CAACTCCACTGGGGAAGAAGTG CGGCGGCTACCACTCAC probe: TGCGCAAGCTGGTG	NM_001282625.1	60°C	2.0
LMNA^b <i>progerin</i>	progerin	ACTGCAGCAGCTCGGGG TCTGGGGGCTCTGGGC probe: CGCTGAGTACAACCT	NM_001282626.1	60°C	2.0

^a Accession numbers from reference sequences taken from GenBank are shown.

^b primer and probe sequences were taken from Rodriguez *et al.* (2)

References

1. Hendig D, Langmann T, Kocken S, et al. Gene expression profiling of ABC transporters in dermal fibroblasts of pseudoxanthoma elasticum patients identifies new candidates involved in PXE pathogenesis. *Lab Invest*. 2008;88(12):1303-1315. doi:10.1038/labinvest.2008.96
2. Rodriguez S, Coppède F, Sagelius H, Eriksson M. Increased expression of the Hutchinson–Gilford progeria syndrome truncated lamin A transcript during cell aging. *Eur J Hum Genet*. 2009;17(7):928-937. doi:10.1038/ejhg.2008.270