Supplemental Table 1 Summary of whole body dual energy X-ray absorptiometry (DEXA) of Patient 4

	% Fat
Left arm	7.2
Right arm	7.5
Trunk	6.6
Left leg	4.7
Right leg	3.9
Subtotal	5.9
Head	18.3
Total	7.1

Supplemental Table 2 Laboratory data of the parents of Patient 4 with a heterozygous *PTRF* c.696697insC mutation.

	Father	Mother
Height/ Body weight	161cm/61kg	158cm/73kg
BMI	23.4	29.2
T-chol (140-199mg/dl)	262	ND
TG (<149mg/dl)	180	111
HDL-C (>40mg/dl)	55.1	71
LDL (<119mg/dl)	195	123
Fasting glucose (<99mg/dl)	113	ND
HbA1c (<5.1%)	ND	5.2
Others	Hypertension	Hypertension
Ultrasonography	Normal	ND

ND: not done

Supplemental Table 3 Primers used in this study

	Exon		Primers
PTRF	Ex 1-1	F	5' CTGCTTCTCTCCGGGTCTC 3'
		R	5' AGCTTGCTCACCGTATTGCT 3'
	Ex 1-2	F	5' CCGGCTCAGAAGAGCTGAT 3'
		R	5' GTCTCCCCACCCCAACTC 3'
	Ex 2-1	F	5' CCCTCTCTCCAGGATCGTC 3'
		R	5' AGCTTGTTCATGCGCTTCTC 3'
	Ex 2-2	F	5' CCGCGAGAACCTGGAGAA 3'
		R	5' ACAGCTCTAAGACTGGGAGTGG 3'
	mRNA	F	5’ GCTTCTCTCCGGGTCTCC 3’
		R	5’ACAGCTCTAAGACTGGGAGTGG 3’
CAV1	Ex 1	F	5' TCAGTTCCCTTAAAGCACAGC 3'
		R	5' GCAGTCGGGATATTTGGAGA 3'
	Ex 2	F	5' TGTTTTTCTTTTTGCATTTTTCC 3'
		R	5' GCTAACTGCCAGAGGAGAGC 3'
	Ex 3	F	5' TCTTTTCTTCTATTCTGTGCTCATGT 3'
		R	5' GAAATTGGCACCAGGAAAAT 3'
	mRNA	F	5’ CAGGGAAACCTCCTCACAGT 3’
		R	5’ GCAACTTGGAACTTGAAATTGG 3’
CAV2	mRNA	F	5’ AGTTCCTGACGGTGTTCCTG 3’
		R	5’ CGTCCTACGCTCGTACACAA 3’
CAV3	Ex 1	F	5' AGCGACCGCCGAGTTGG 3'

		R	5' CCGAGGCAGGCCTGCAGAGCC 3'
	Ex 2	F	5' GCTTCTGTGAGTTGAGGCTT 3'
		R	5' ATCATGGGGTATGGAGCAGT 3'
	mRNA	F	5’ GATGATGGCAGAAGAGCACA 3’
		R	5’ GTGGACAACAGACGGTAGCA 3’
LMNA	Ex 1	F	5' CCCAGATCCCGAGGTCCGAC 3'
		R	5' CCTCTCCACTCCCCGCCA 3'
	Ex 2	F	5' ATTGCAGGCATAGCAGCGC 3'
		R	5' GAGGGCCTAGGTAGAAGAG 3'
	Ex 3	F	5' TTCTTGTGTTCTGTGACCCCTT 3'
		R	5' CCCAAGTCTGTCATCACCCA 3'
	Ex 4	F	5' GTCCCTGGGTCTTGGCCTCC 3'
		R	5' GCCACCATCTGCCTGATCC 3'
	Ex 5	F	5' GCTGTAGCAGTGATGCCCAAC 3'
		R	5' CCAAAGCCCTGAGAAGTGAAG 3'
	Ex 6	F	5' CAGCTGTCTCCTACACCGACC 3'
		R	5' GGTCTAGTCAAGGCCAGTTGC 3'
	Ex 7	F	5' GAGGTGCTGGCAGTGTCCTC 3'
		R	5' CTTCTGTCTTGCCACTCTCTCC 3'
	Ex 8	F	5' ACCCAAGAGCCTGGGTGAGC 3'
		R	5' CACCCAAGGTCTCCCCAGAG 3'
	Ex 9	F	5' TTGGGTGTCGATGGGAGCGC 3'
		R	5' GCAGCTGCCTCCGATGTTGG 3'

	Ex 10	F	5' ACCCTTCCCTGGCCCTGAC 3'
		R	5' CACCTGGGTTCCCTGTTCAAG 3'
	Ex 11	F	5' GGTCAGTCCCAGACTCGCC 3'
		R	5' CGCCTGCAGGATTTGGAAGAC 3'
	Ex 12	F	5' TGAGGGATGGGGGAGATGCT 3'
		R	5' GGGTGGGCATGAGGTGAGGA 3'
AGPAT2	Ex 1	F	5' GCAATAAGGGGCCTGAGC 3'
		R	5' AAAGTTAGGGAAGCGGAAGC 3'
	Ex 2	F	5' CTGTGTCTCCCGGTCTCCT 3'
		R	5' CCCACTCAAACCCCAGAAG 3'
	Ex 3	F	5' CAGGTAGCCAGGGAGAAGG 3'
		R	5' TGATGTGGGGGTCTTGTTTT 3'
	Ex 4	F	5' CAGCTGCTTTGCGAAGTG 3'
		R	5' GTGGTCACCTGCTGCCTTA 3'
	Ex 5	F	5' CTCCCCAACCACATGCAG 3'
		R	5' GGAAATGGGAACGATGAGG 3'
	Ex 6	F	5' TAGGGAGTCCAGGGGAAGAG 3'
		R	5' CCTCTGCCCATCCTCCAG 3'
BSCL2	Ex 2	F	5' CACAGGTATCAGCGTCTGGA 3'
		R	5' ACGCCAGCCCACCTATTT 3'
	Ex 3	F	5' CTCGTTCCTCAAAGCCAGTC 3'
		R	5' GAACTCCAAGGGCCTTACAA 3'
	Ex 4	F	5' GGGCAAAGAAGGTGTATGGA 3'

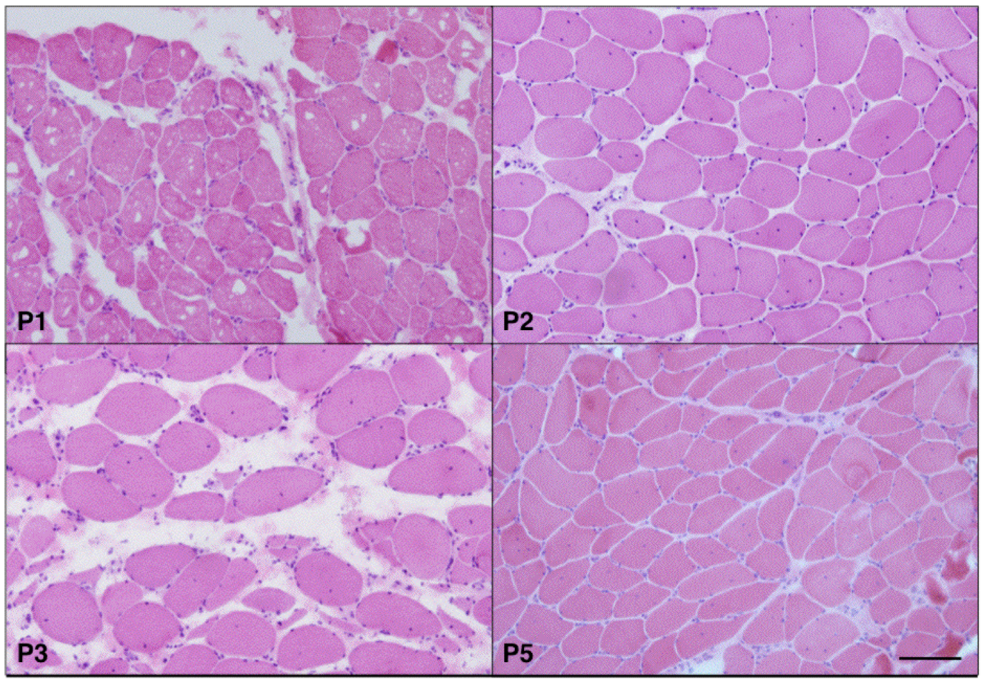
		R	5' AGGCCTTTCTCAAGTCTTCCT 3'
	Ex 5	F	5' GGCTAAGAGGGAGGTGGAAT 3'
		R	5' CCCCTACCCATTCTGATCCT 3'
	Ex 6	F	5' GATCTGGCATGCACCTGTC 3'
		R	5' CAGGTAGAATCCTGGGCTAATG 3'
	Ex 7	F	5' TCAGATGAGGCGGGTAAGAG 3'
		R	5' GACCCTCTTGGTGGAAGGTT 3'
	Ex 8	F	5' TGGTGCTCTCTGGTAACTGC 3'
		R	5' CCCTGACCACCCACAAAG 3'
	Ex 9	F	5' GCATGAGGGTCCTGATTGTT 3'
		R	5' CGGTGATACCCTAAGCCTCA 3'
	Ex 10	F	5' ACCGACTGAGACAAGGGTCA 3'
		R	5' TGAAGGAGAAAGCCAAGGAG 3'
	Ex 11	F	5' GCCACCACACTCTCTCCTTC 3'
		R	5' CCAGGAAAGGGAAAAACAAA 3'
	Ex 12	F	5' GGCCCCCGGTTAATCTAATA 3'
		R	5' GAAAAACGAGGGGAGAGGAG 3'
PPARG	Ex 1	F	5' AACGGATTGATCTTTTGCTAGA 3'
		R	5' CAAACACAACCTGGAAGACAAA 3'
	Ex 2	F	5' CGCCCAGATGAGATTACTTTG 3'
		R	5' TGAGAGATGAGTCCAATTCTAGTCC 3'
	Ex 3	F	5' CCCTGTTGCCTTTTTAGGAC 3'
		R	5' GGGGTTCTGCTGAAATGAAA 3'

	Ex 4	F	5' GCTGCTTCCATGTGTCATAAA 3'
		R	5' GGTCTGGCAGCTATAATGAGAA 3'
	Ex 5	F	5' GGAGGGCTTCTACTGTGTGG 3'
		R	5' GTGTGCATTTGTAGCGCAGT 3'
	Ex 6	F	5' GTTTTCTGAACCTGGGATGG 3'
		R	5' ACCATCATCCCACCCTCTTT 3'
	Ex 7	F	5' CGTGCAAAACACTGTACCAAG 3'
		R	5' TGGAAGAAGGGAAATGTTGG 3'
AKT2	Ex 2	F	5' CAGACTGTGCCCTGTCCAC 3'
		R	5' TGGCTCCTCAGGCTGGTA 3'
	Ex 3	F	5' CCTGGGACATCACTCAACCT 3'
		R	5' TCCTGTGAGCACCAGAACAC 3'
	Ex 4	F	5' TAGCAGGGCTATCCCCTCAG 3'
		R	5' CCCAGGGAAAATCTCTCCAG 3'
	Ex 5	F	5' TCAGCTGTTCGACTCTGAGG 3'
		R	5' ATGGAAACCAAGGAGAGCAG 3'
	Ex 6	F	5' TGGCAACAGTGTCTTTTGGT 3'
		R	5' CTCAGGGTCAGGCTCCAG 3'
	Ex 7	F	5' CTGCATTTATGGCTGGGAAT 3'
		R	5' TCTCTGAGCTCTGTCCAAAGG 3'
	Ex 8	F	5' AGGGTGTGTGTGTGCAGAAT 3'
		R	5' TAAAGAAGGAGGCCCCAGAG 3'
	Ex 9	F	5' GGAAGGTGAGGGCAGGTG 3'

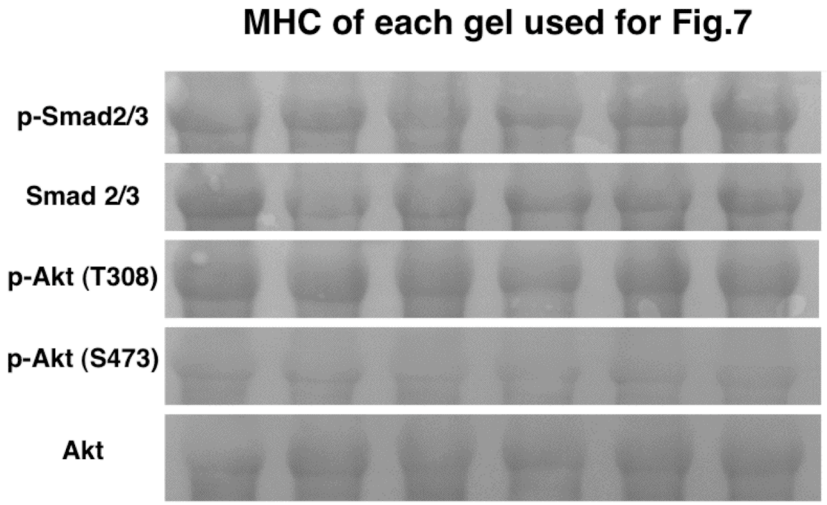
		R	5' AGTGTGAGTCCCATGTGGTG 3'
	Ex 10	F	5' TGGGAGCATGCACCTGAG 3'
		R	5' GGCTGGAAACACACAGGTCT 3'
	Ex 11	F	5' CTGAGGGCAGGACTGTGATG 3'
		R	5' GACACACTGCGACCCTACAA 3'
	Ex 12-13	F	5' ATACAGGGCCTGCCTGCT 3'
		R	5' GCCCTCCTTGAGAAGTGAGTT 3'
	Ex 14	F	5' CCCTCTCCTGACTGGTCCTC 3'
		R	5' GTGGGGACACAAACCAAAAA 3'
ZMPSTE24	Ex 1	F	5' AAGGGACGAGTGTCGGTGT 3'
		R	5' TCGAAGCCAAGGCTACTCC 3'
	Ex 2	F	5' TGGCAAGCTATAAACCATTCG 3'
		R	5' TGAAAATGAAAACAACCAGACA 3'
	Ex 3	F	5' TGTCCTTTCTTTCTTTATACCATGC 3'
		R	5' TTAGTGGAAAGCCTGCCAAG 3'
	Ex 4	F	5' CTTGTTGATTTGTTTGCCAGT 3'
		R	5' CAGGACAAAAGCACAGAAGTTTT 3'
	Ex 5	F	5' GCAGAACCAGTTTCTCAGTTTC 3'
		R	5' TCTCACCAAGGAACTTTTGC 3'
	Ex 6	F	5' CCTGGGAATACCAGAGCAAG 3'
		R	5' TCAAATAAAACAAACCACTTGGA 3'
	Ex 7	F	5' CTCCAAAGGACCCCAAACTT 3'
		R	5' CACAGGAACTGTGTATAAAAACTAAGG 3'

	Ex 8	F	5' AGGGCTATTACTGGGTTAAAAGA 3'
		R	5' CCTCTCATGCCTGCCATAGT 3'
	Ex 9	F	5' CCCATAGTGAAATCAGCTTGGTA 3'
		R	5' TTGAAGCAGGCAAGAGCATA 3'
	Ex 10	F	5' TCCTCTTATCCCAAGCCAAA 3'
		R	5' TCAAGAGCTGGAACATGCTG 3'
MSTN	mRNA	F	5’ TGATGTCCAGAGGGATGACA 3’
		R	5’ GAGTCTCGACGGGTCTCAAA 3’
GAPDH	mRNA	F	5’ GGTAAAGTGGATATTGTTGCCATCAATG 3’
		R	5’ GGAGGGATCTCGCTCCTGGAAGATGGTG 3’

Supplemental figures



Supplemental Figure 1 Hematoxylin and eosin staining of biopsied skeletal muscle from Patient 1 (P1), Patient 2 (P2), Patient 3 (P3), and Patient 5 (P5) shows variable dystrophic changes.



Supplemental Figure 2 CBB staining of each gel for immunoblotting analysis for phosphorylation of Smad and Akt (Figure 7) shows similar protein amounts of myosin heavy chain (MHC).