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Appendix Figure Legends

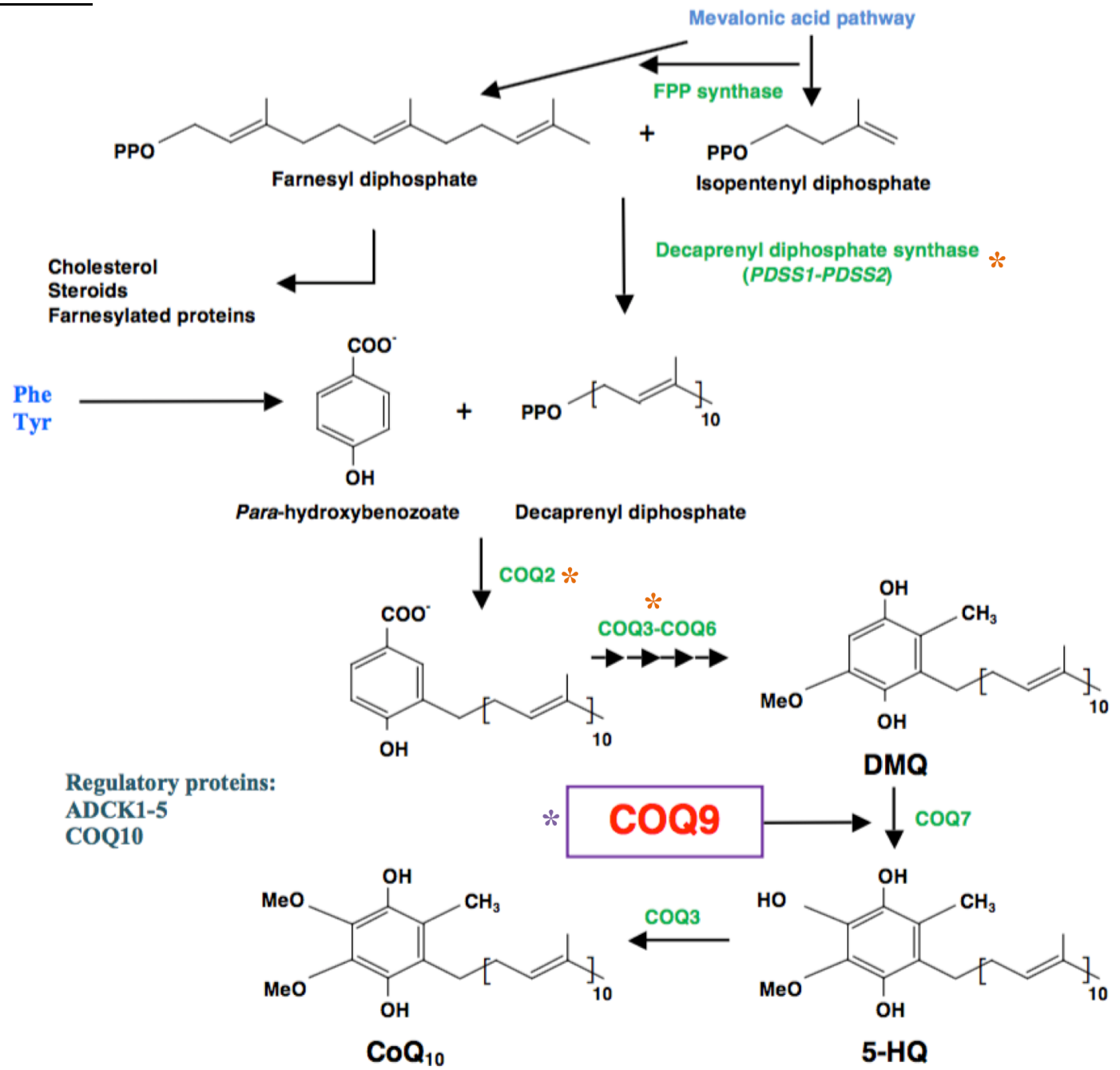
Appendix Figure S1. CoQ biosynthetic pathway. Purple asterisk indicates the dysfunctional protein in the mouse models; orange asterisks indicate the dysfunctional proteins in the patients' skin fibroblasts. COQ9 is needed for the reaction catalyzed by COQ7. PDSS2 and COQ2 are catalytic enzymes. COQ4 may play a structural role in the multiprotein complex for CoQ biosynthesis.

Appendix Figure S2. Glutathione system in muscle and kidney of *Coq9^{R239X}* mice. Total GSH in cytosol and mitochondria of muscle (A) and kidney (B) of *Coq9^{+/+}* and *Coq9^{R239X}* mice. Cytosolic GPx and GRd activities in muscle (C) and kidney (D) of *Coq9^{+/+}* and *Coq9^{R239X}* mice. Data are expressed as mean $\pm$ SD. * $P < 0.05$; *Coq9^{R239X}* mice versus *Coq9^{+/+}* mice (*t* test; $n = 5$ for each group).

Appendix Figure S3. Glutathione redox status in cerebrum, muscle and kidneys of *Coq9^{R239X}* mice. GSSG / GSH ratio in cytosol (A) and mitochondria (B) from cerebrum, muscle and kidneys of *Coq9^{+/+}* and *Coq9^{R239X}* mice (*t* test; $n = 5$ for each group).

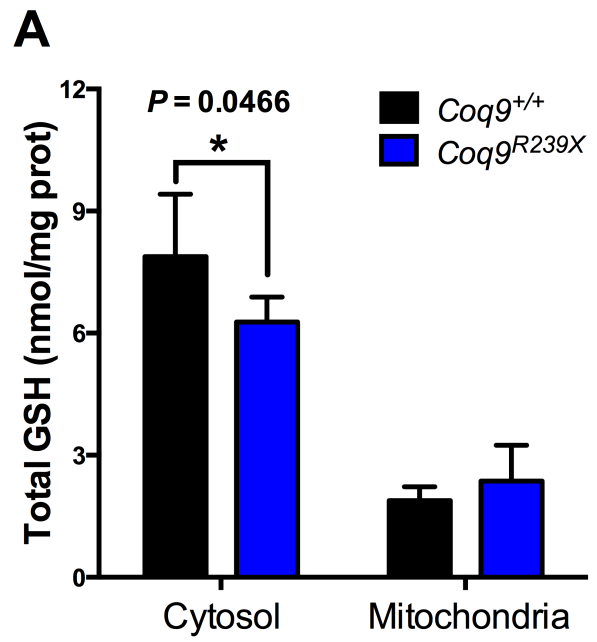
Appendix Figure S4. Consequences of SQR deficiency in cellular GSH levels. Levels of Sqr mRNA (A) SQR protein (B) and GSH (C) in Hepalclc7 cells after culture of SQR siRNA during 48 and/or 72 h. Levels of GSH (D) in fibroblasts of controls (C1-C3) and patients (P1-4) with primary CoQ₁₀ deficiency. Data are expressed as mean $\pm$ SD. *** $P < 0.001$; SQR siRNA versus negative siRNA control (*t* test or one-way ANOVA with a Tukey's *post hoc* test; $n = 4$ for each group).

Appendix Figure S1

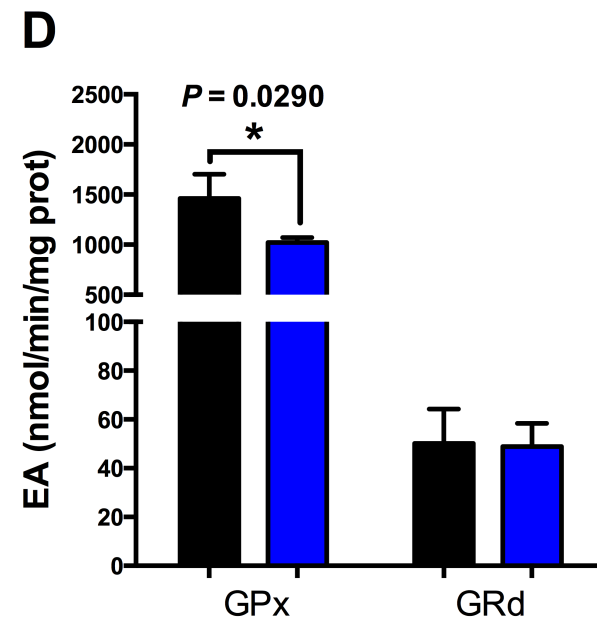
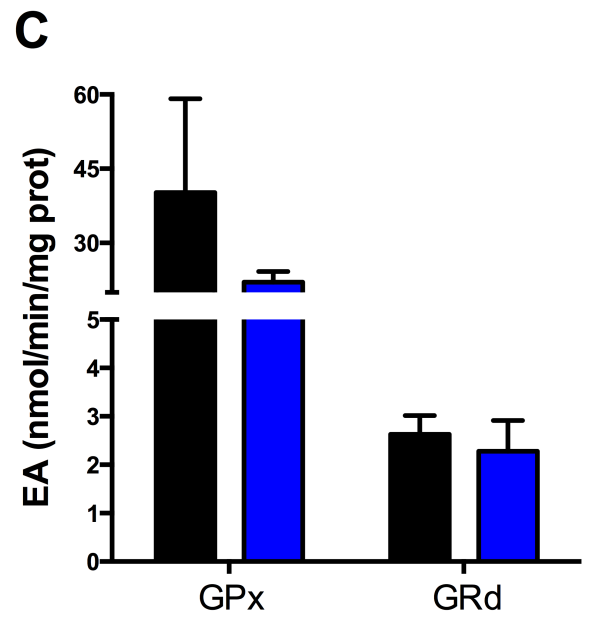
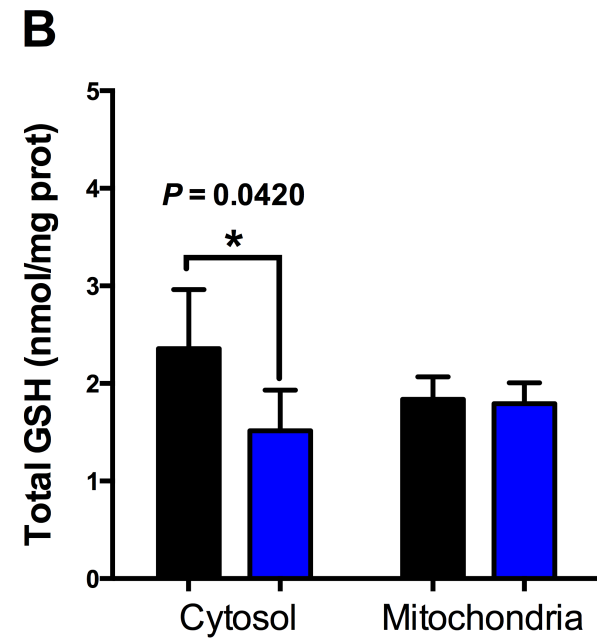


Appendix Figure S2

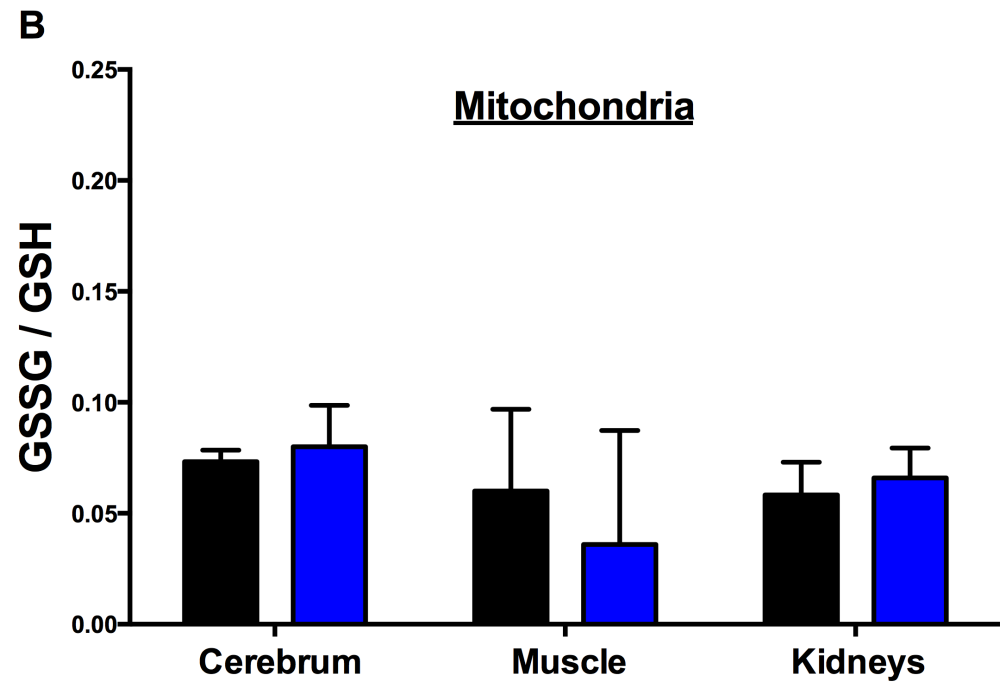
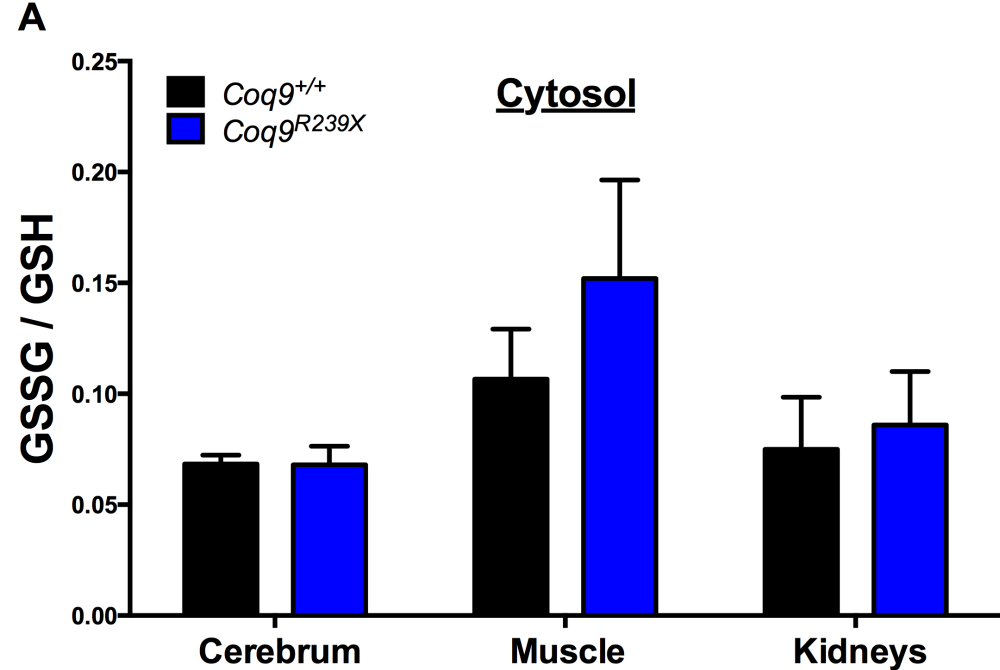
Muscle



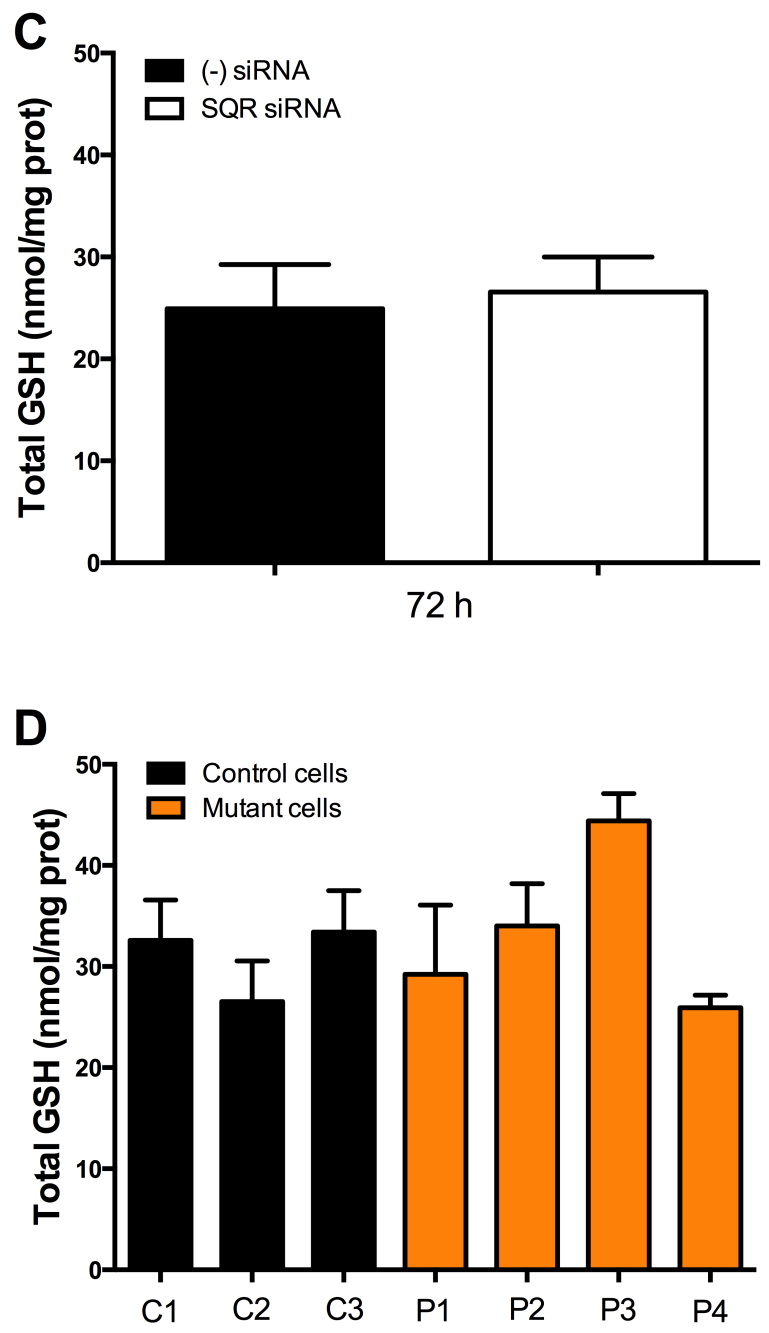
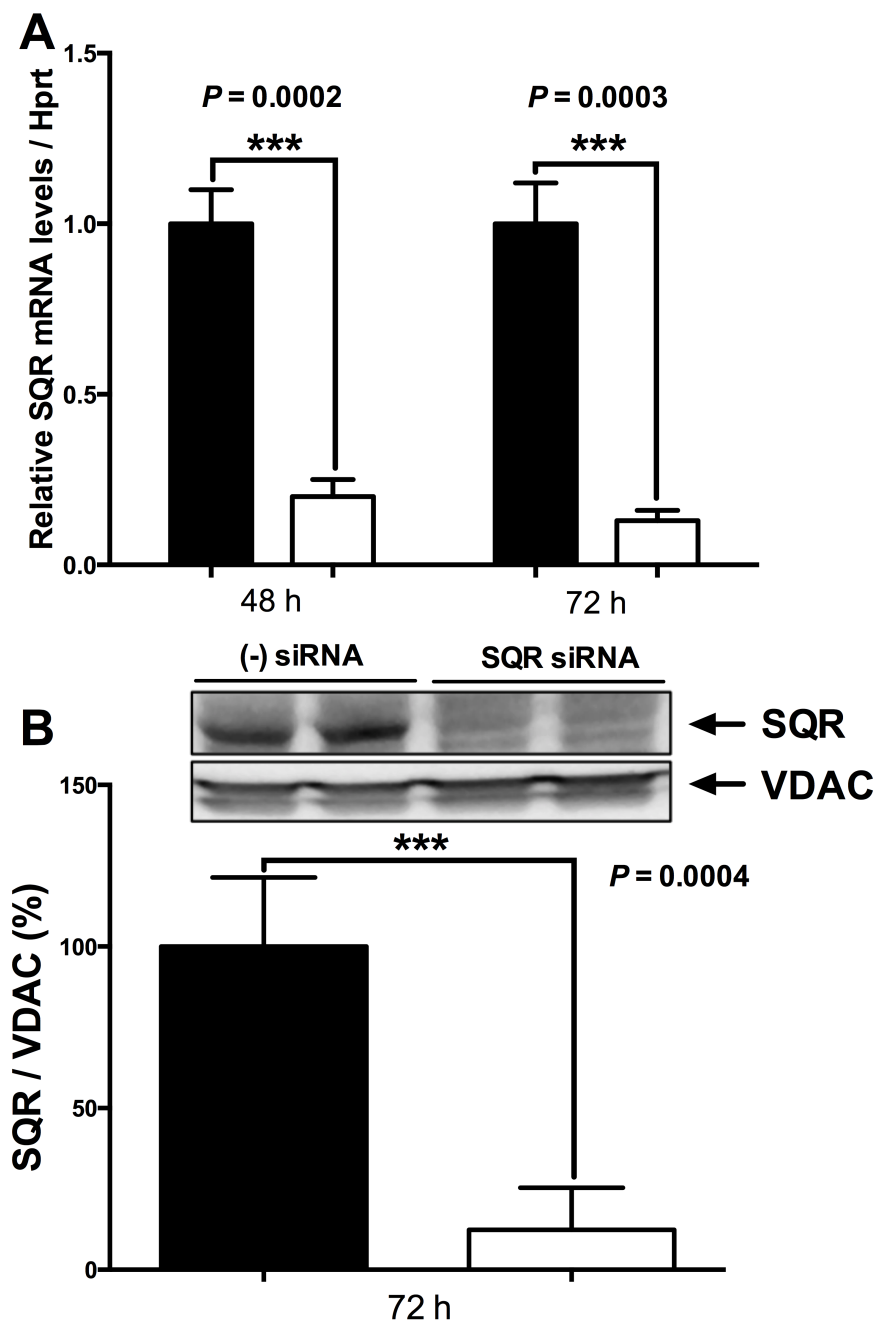
Kidneys



Appendix Figure S3



Appendix Figure S4



Appendix Table S1

Sample	Gene	Sequence Ref	Mutations	Protein change	Reference
P1	<i>PDSS2</i>	NM_020381.3	c.[964C>T];[1145C>T]	p.[Gln332Stop];[Ser238Leu]	López et al., 2006
P2	<i>COQ2</i>	NM_015697.7	c.760T>A	p.Trp254Arg	
P3	<i>COQ4</i>	NM_016035.3	c.[155T>C];[518 520delCCA]	p.[Leu52Ser];[Thr174del]	Brea-Calvo et al., 2015
P4	<i>COQ9</i>	NM_020312.3	c.521+1delG		Danhauser et al., 2015

Table S1. Identified mutations in the human skin fibroblasts used in this study.