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Supplemental Data

COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ_{10} Deficiency

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SUPPLEMENTAL DATA

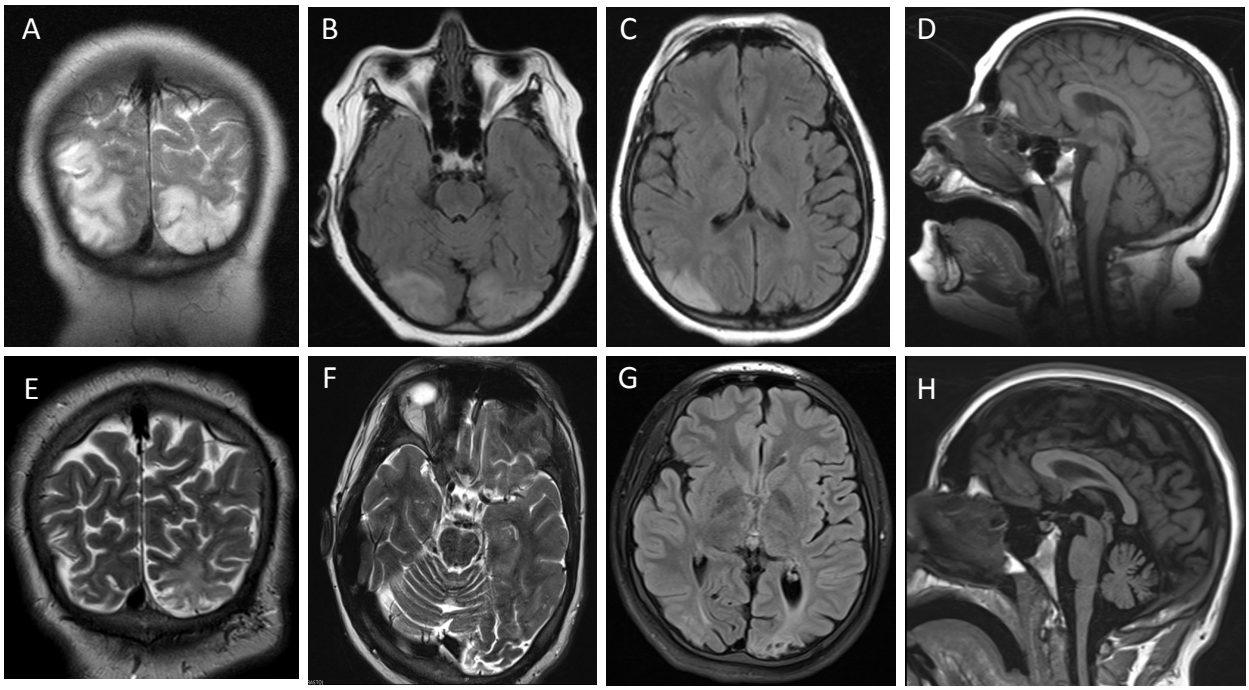

Figure S1. Brain MRI of subject 5.

Two serial brain MRI images of subject S5 obtained at age 12 years (A-D) and at age 17 years (E-H). A and E are coronal T2 weighted sections to show abnormal hyperintense cortical areas corresponding to infarct-like lesions of the occipital lobes at two different stages of progression to cortical atrophy with scars (E-G); The same occipital abnormal hyperintense cortical areas are shown in axial sections B and C (FLAIR weighted), F (T2 weighted) and G (FLAIR weighted). Notice that no abnormality is present in basal ganglia and brainstem. Finally, figures D and H are sagittal T1 sections showing progressive cerebellar atrophy of the vermis.

Id	Type	Reads	Mapped	Percent	Seq (Gb)	on bait	Avg cov	Cov 1x	Cov 4x	Cov 8x	Cov 20x
S1	SureSelect50Mbv5	106627996	105933934	99.35	10.77	78.48	132.44	99.92	99.72	99.35	97.38
S2	SeqCapEZ V1	130889538	125611825	95.97	13.81	62.71	155.83	98.36	97.32	96.41	93.80
S4	SureSelect50Mbv5	90821087	90214603	99.33	9.17	78.99	113.62	99.80	99.52	99.05	96.55
S5	SureSelect50Mbv4	125822167	124979728	99.33	12.71	73.98	146.50	99.95	99.80	99.55	98.22

Table S1: Exome Sequencing Statistics

Avg: average; Cov: coverage

Nucleotide Change	Amino acid Change	Subject	Status	Polyphen2	SIFT	PMUT	Mutation taster
c.[433C>G]	p.[Arg145Gly]	S1	Homozygous	Probably damaging	Deleterious	Pathological	Disease causing
c.[718C>T]	p.[Arg240Cys]	S2	CE with a nonsense mutation p.[Arg141*]	Probably damaging	Deleterious	Pathological	Disease causing
c.[155T>C]	p.[Leu52Ser]	S3, S4	CE with a deletion p.[Thr174del]	Probably damaging	Deleterious	Neutral	Disease causing
c.[190C>T]	p.[Pro64Ser]	S5	Homozygous	Probably damaging	Deleterious	Neutral	Disease causing

Table S2: In silico prediction of pathogenicity for *COQ4* mutations

CE: compound heterozygous.