Supplemental Data

The Mitochondrial Disulfide Relay System Protein GFER Is Mutated in Autosomal-Recessive Myopathy with Cataract and Combined Respiratory-Chain Deficiency

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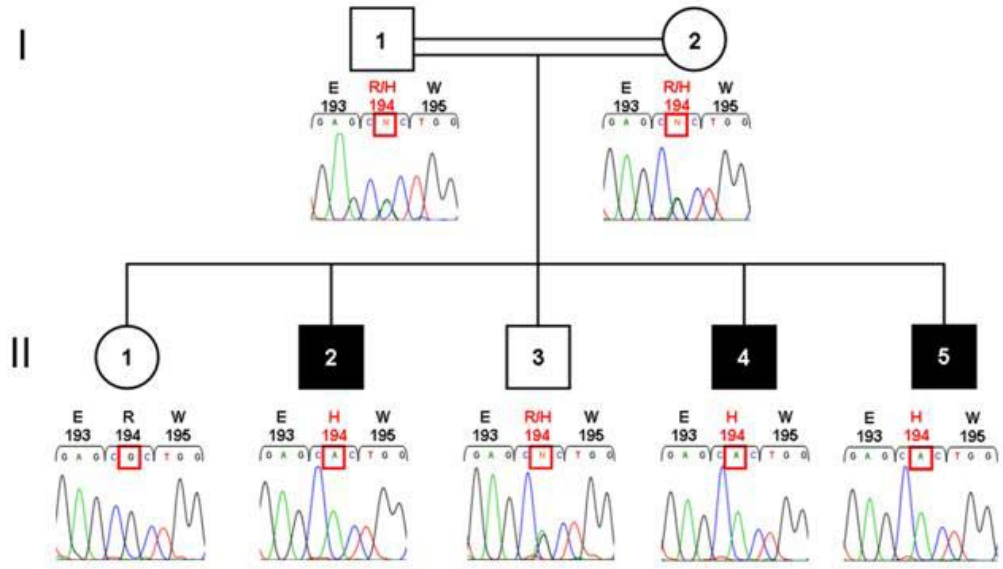
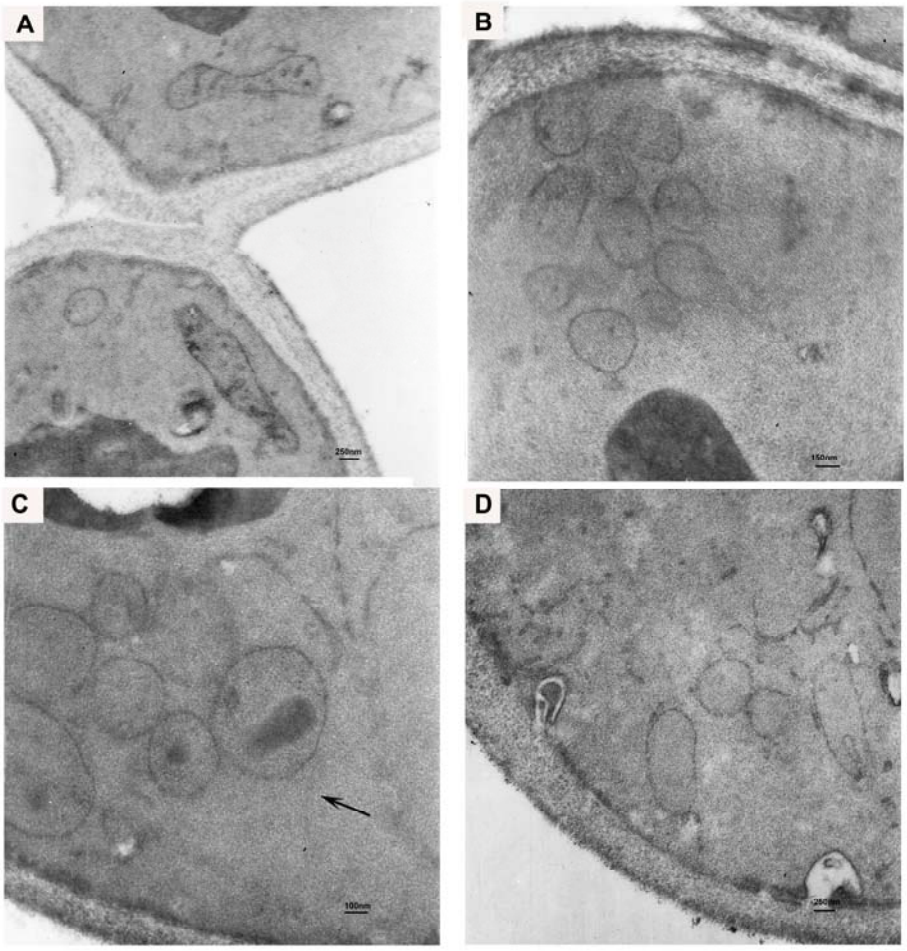
Figure S1. Pedigree Diagram of the Genotyped Family



Black symbols denote affected individuals, open symbols denote unaffected individuals. Electropherograms of the genomic DNA sequence of *GFER* show the segregation of c.581 G>A and predicted amino acid substitutions (in red).

Figure S2. Yeast Ultrastructural Analysis



(A) Wild-type yeast showing normal mitochondria with normal cristae. (B-C) Yeast carrying the *erv1R182H* allele after an 8-hour shift to 37°C. Note the clusters of swollen mitochondria with no cristae, showing both small and large electron dense inclusions (arrow). (D) Mitochondria with no cristae and thin membranes in mutated yeast at 16 h.

Table S1. Oligonucleotides Used for the PCR Amplification of *GFER* and the Sequence Analysis of Genomic DNA and cDNA

GFER_1aF	TGGCTCCGCCTCCACACG
GFER_1aR	TCCACGTCTTGAAGTCGACG
GFER_1bF	TGACCCGGCAGGCCTTGC
GFER_1bR	AGGTTCAGGGACAGGCAACC
GFER_2F	GATCGTCTGCAGGACTTTGG
GFER_2R	CATCCGTTTCTCTACGTTCC
GFER_3F	GGTGTAGTTCACAGCAGTGC
GFER_3R	TGAGACACAACAGGCTCTGG
cDNA_1F	CCTTGCGCGGGCAACATG
cDNA_3R	CTAGTCACAGGAGCCATCCTTC
cDNA_1RX>L*	CAAGTCACAGGAGCCATCCTTC

* This primer was used in the creation of the GFER-GFP fusion protein.