Supporting Information

Zollo et al. 10.1073/pnas.1118594109

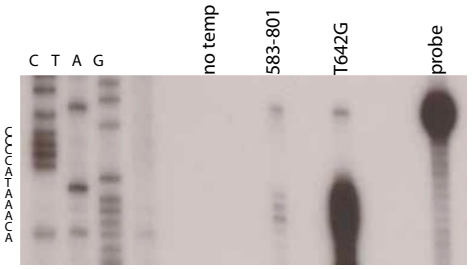

Fig. S1. The size of the transcript produced by transcription from the T642G template was compared with WT using S1 as described in Fig. 2. Although transcription from T642G was considerably more robust, the end positioning was the same. Controls include undigested probe and a transcription reaction run without DNA.

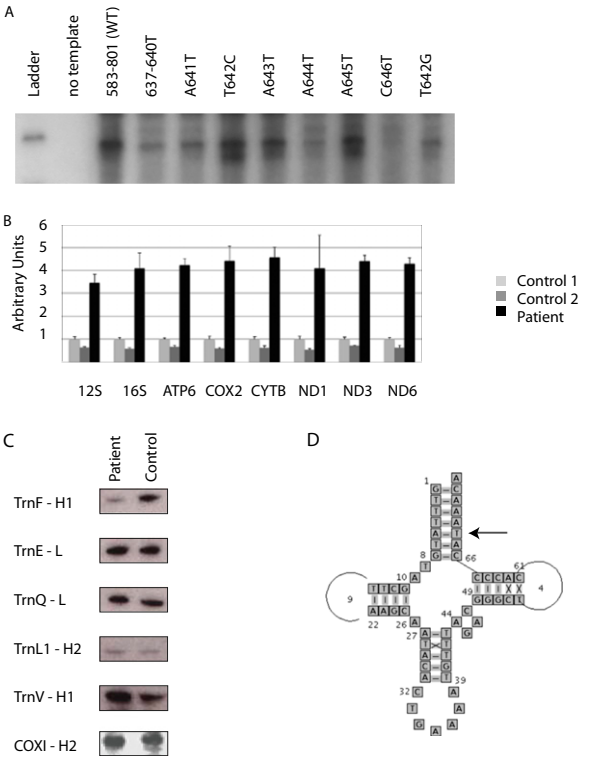

Fig. S2. Analysis of muscle RNA from a patient with the m.642T > C mutation. (*A*) In vitro transcription of heavy-strand promoter 2 (HSP2) mutations by partially purified mitochondrial extract from HeLa. (*B*) Transcription in frozen muscle biopsy specimens was evaluated using real-time PCR. Studies were normalized to GAPDH expression, and the control 1 sample was set as the reference sample. Expression of mitochondrial transcripts was globally up-regulated in the m.642T > C patient, with the effects seen in transcripts derived from all three precursor RNAs. (*C*) RNA was extracted from frozen patient and control muscle samples and 200 ng RNA as resolved on 15% acrylamide Tris-borate-EDTA–urea gels for tRNA blotting and formaldehyde-agarose gel for cytochrome c oxidase subunit I blotting. (*D*) Cloverleaf structure of mitochondrial phenylalaninyl tRNA (MTTF) showing the location of the m.642T > C mutation. Illustration from Mamit-tRNA (http://mamit-tRNA.u-strasbg.fr).