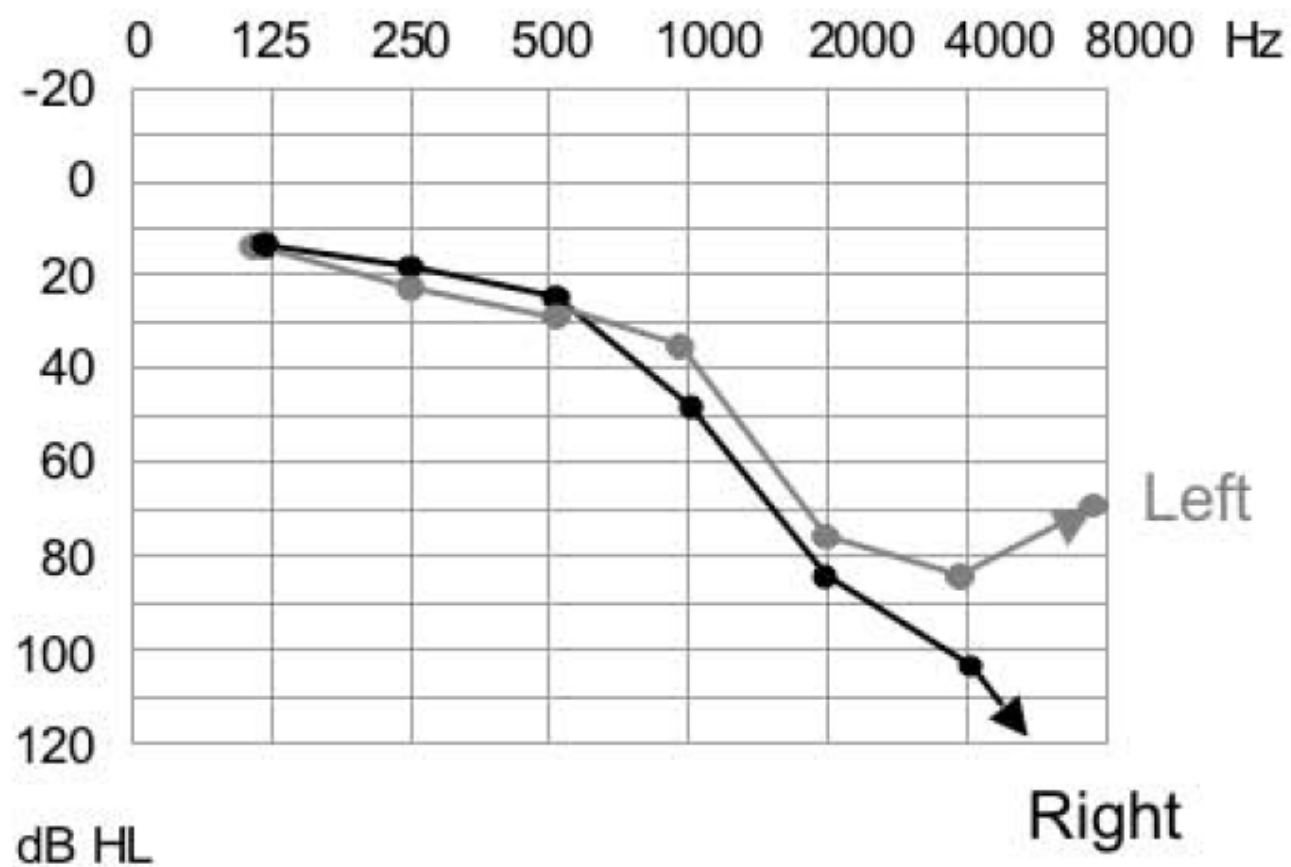


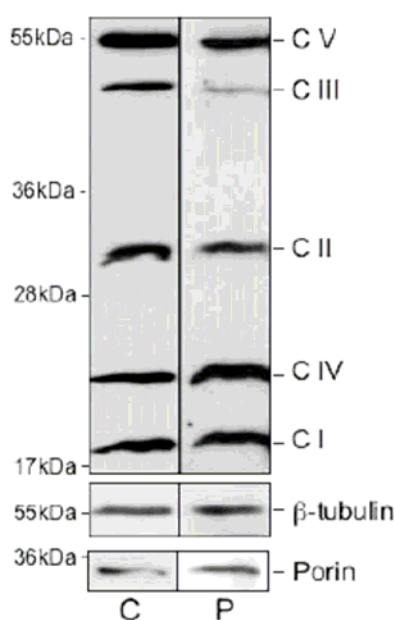
SUPPLEMENTARY DATA

Supplementary Figure 1. Audiogram of the patient.



SUPPLEMENTARY DATA

Supplementary Figure 2. Western blotting of fibroblast homogenates. Equal amounts (50 μ g) of protein extracts from a control adult individual (C) and the patient (P) were subjected to SDS-PAGE and blotted onto a PVDF membrane prior to incubation with a cocktail of anti human total OXPHOS complexes monoclonal antibodies. Antibodies against β -tubulin (Sigma, St Quentin Fallavier, France) were used at 1/10.000. Antibodies against porin (Mitosciences, Eugene, USA) were used at 1/1000. They correspond respectively to loading and mitochondrial controls.



Supplementary Figure 3. Identification of mtDNA mismatches, secondary to m.14535_14536insC mutation, by Surveyor Nuclease digestion. After amplification, the PCR product (2002 bp) was digested with Surveyor Nuclease in a final volume of 60 μ l. For control, undigested PCR product was systematically mixed with water in the same volume. Digested (+) and undigested (-) products were analyzed on a 1.5% agarose gel. According to the position of forward (nt13491-nt13511) and reverse (nt15493-nt15473) primers, the digestion of the amplicon bearing an insertion at position 14535 leads to 2 fragments, 1044 bp and 958 bp in length. Arrows indicate the location of cleavage fragments, which are not found in lanes corresponding to undigested PCR product from the patient and to digested and undigested PCR products from a control individual. M1, M2 : size markers.

