**Supplementary Figure S1. Pedigrees carrying LHON mutations**

**A.** Pedigrees harboring m.11778G>A mutation. **B.** Pedigrees harboring m.3460G>A mutation.

Affected are indicated by filled symbols; Carriers are indicated by white symbols. Percentage of mutant alleles for each available subject is also indicated.

HET, heteroplasmy; n.a, not available.
Supplementary Figure S2. PCR-RFLP analysis by gel electrophoresis of the two LHON mutations

Mutation proportions as *ratio* between mutated *versus* total (wild-type and mutated) fragments estimated as intensities of bands in the gel using Quantity One-4.4.1 imaging software (Bio-Rad Laboratories).

A. m.11778G>A detected by *MaeIII* digestion of the 320-bp amplicon.

B. m.3460G>A detected by *HglI* digestion of the 750-bp amplicon. Two *Affected*, II-3 *FAM-A3* and II-1 *FAM-B3* are not shown.