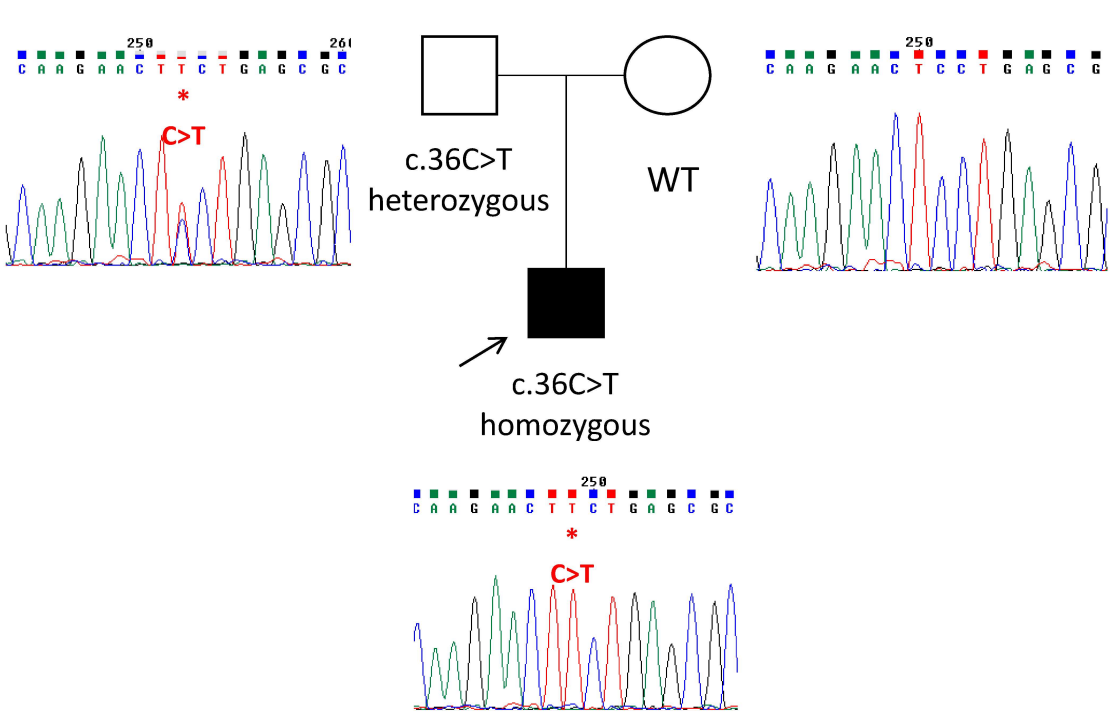
Supplementary data

Genetic analysis

NGS analysis on the proband resulted in the identification of a homozygous SNV (c.36C>T). (A) Family segregation studies showed that the mother was heterozygous for the same variant, while the father was WT. This led to the hypothesis that the second allele in the proband was missing. This hypothesis was confirmed by MLPA analysis, which showed a heterozygous CNV (B), which was originated de novo in the proband.

A



B