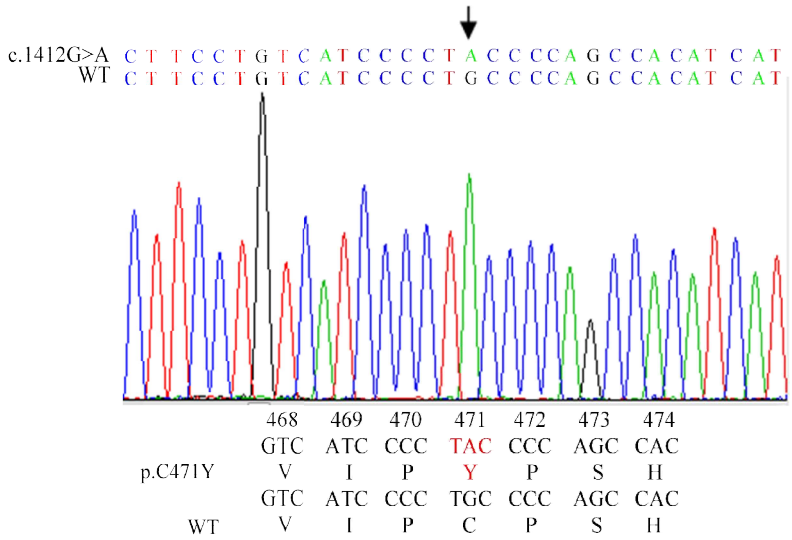
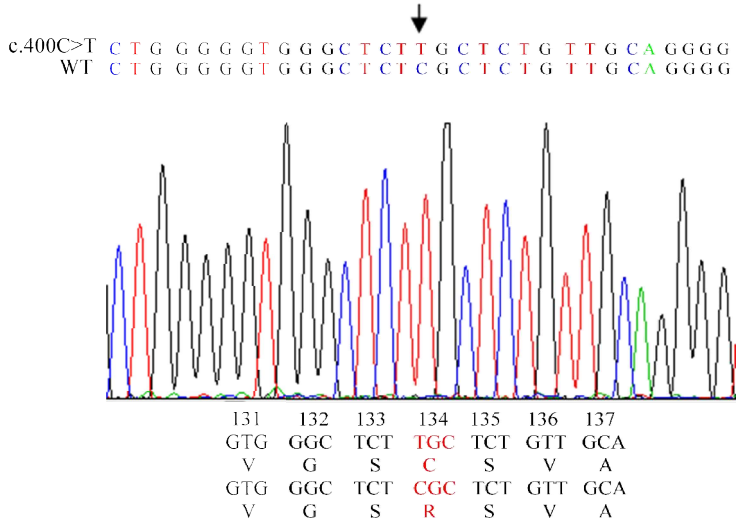
A


c.1412G>A C T T C C T G T C A T C C C C T A C C C C A G C C A C A T C A T
WT C T T C C T G T C A T C C C C T G C C C C A G C C A C A T C A T
468 469 470 471 472 473 474
GTC ATC CCC TAC CCC AGC CAC
V I P Y P S H
p.C471Y
GTC ATC CCC TGC CCC AGC CAC
V I P C P S H
WT

B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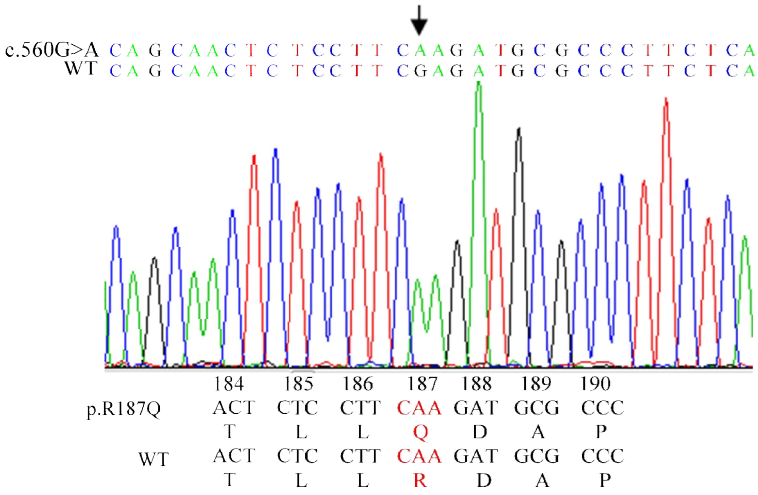
WT C T G G G G G T G G G C T C T C G C T C T G T T G C A G G G G
131 132 133 134 135 136 137
GTG GGC TCT TGC TCT GTT GCA
V G S C S V A
GTG GGC TCT CGC TCT GTT GCA
V G S R S V A

C


c.560G>A C A G C A A C T C T C C T T C A A G A T G C G C C C T T C T C A
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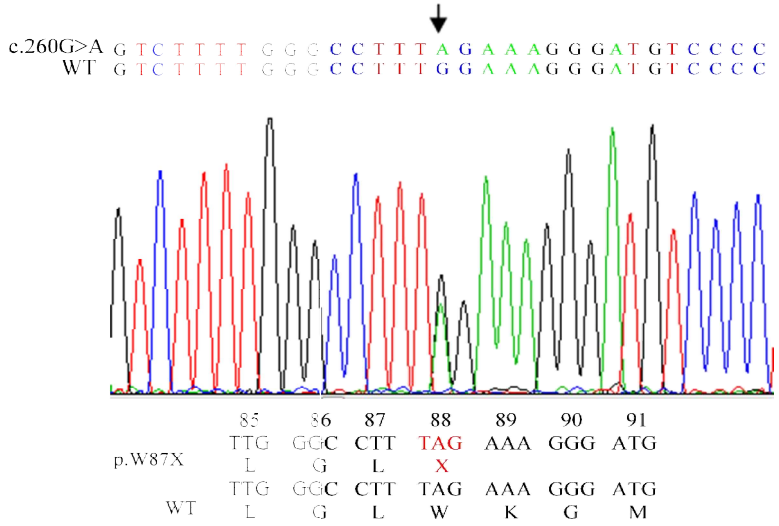
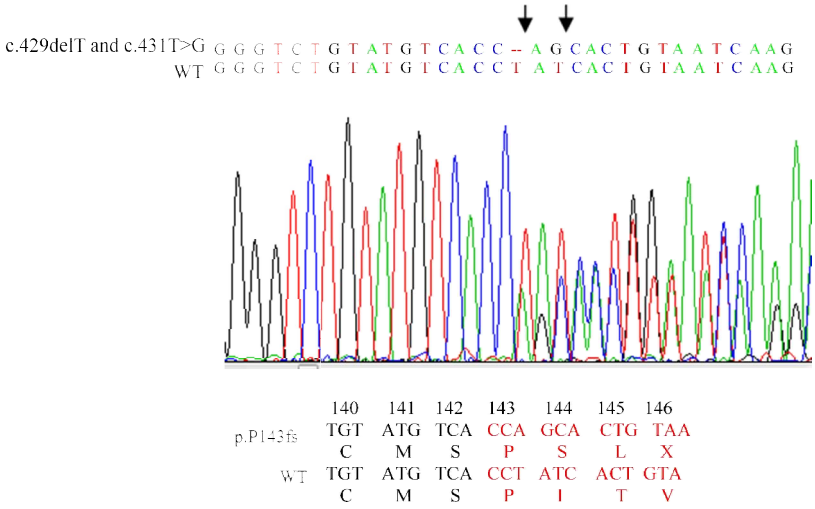
184 185 186 187 188 189 190
ACT CTC CTT CAA GAT GCG CCC
T L L Q D A P
p.R187Q
ACT CTC CTT CAA GAT GCG CCC
T L L R D A P
WT

D



E



Additional file 4. Chromatograms of the mutations found in the patients, as confirmed by Sanger sequencing. (A) Homozygous missense mutation c.1412G>A of ALAS2 in Patient No. 3. (B) Homozygous missense mutation c.400C> T of SLC25A38 in Patient No. 4. (C) Homozygous mis-sense mutation c.560G>A of SLC25A38 in Patient No. 5. (D) Heterozygous nonsense mutation c.260G>A of SLC25A38 in Patient No. 6. (E) Heterozygous nonsense mutations c.429delT and c.431T>G of SLC25A38 in Patient No. 6.