

Supplementary Figures

Mousson de Camaret B, et al.

Supplementary Figure 1. Genotype data

(A) *DGUOK* mutations in patient 1 and his family

(B) *DGUOK* mutations in patient 2

(C) *DGUOK* mutations in patient 3 and her parents

Supplementary Figure 2. Skipping of exon 4 in patient 2

(A) Amplification and sequencing of exons 3-5 from patient 2

(B) Amplification of exons 4-5 from patient 2

Supplementary Figure 3. Aminoacid interactions

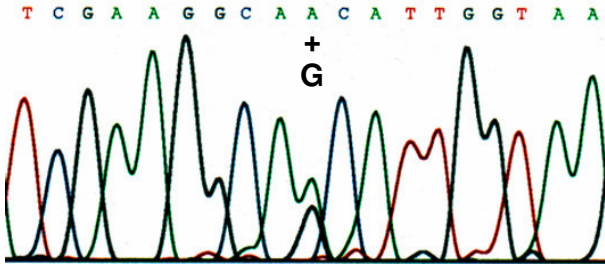
(A) With wild-type N46 and mutant S46

(B) With wild-type L266 and mutant R266

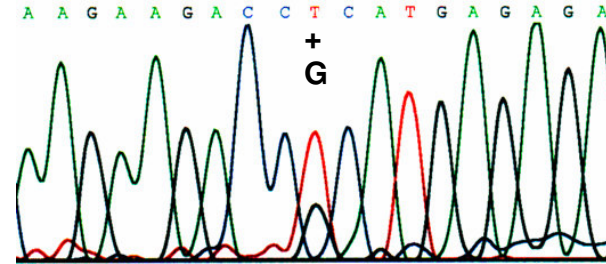
Supplementary Figure 1. Genotype data

(A) Sequencing of *DGUOK* gene. Detection of mutations in patient 1 and in his family

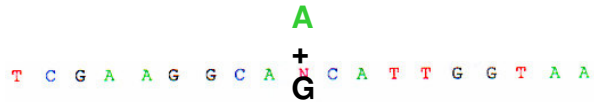
Patient
exon 1: c.137A>G



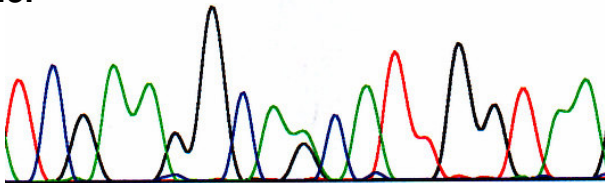
Patient
exon 6: c.797T>G



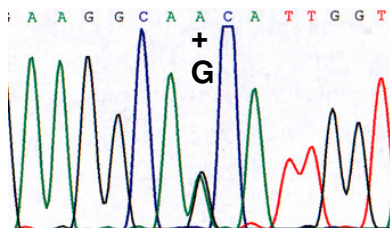
Mother and Brother
exon 1: c.137A>G



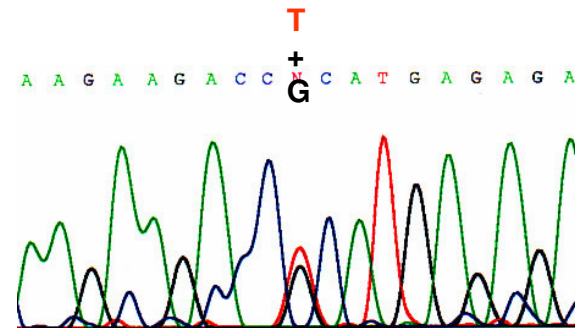
Mother



Brother

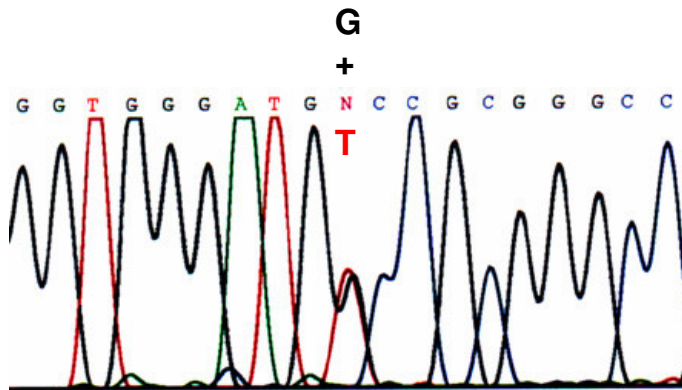


Father
exon 6: c.797T>G

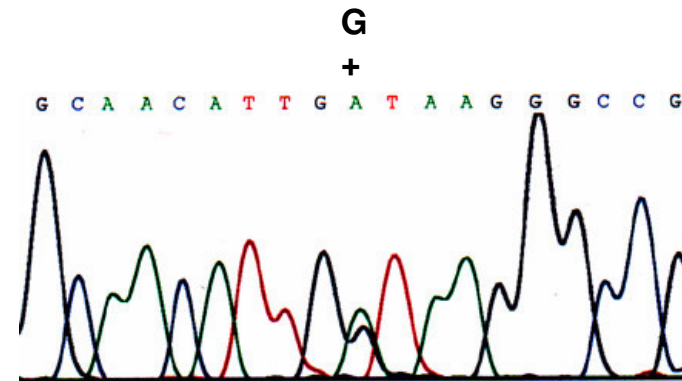


(B) Sequencing of *DGUOK* gene. Detection of mutations in patient 2

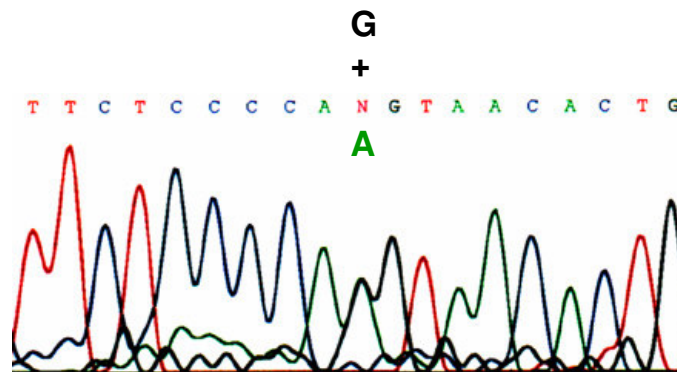
exon 1: c.4G>T (A2S)



intron 1: c.142+1G>A

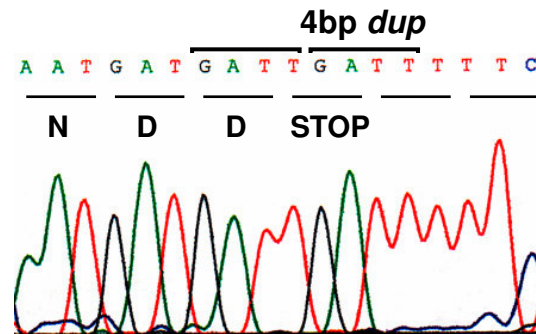


exon 4: c.591G>A

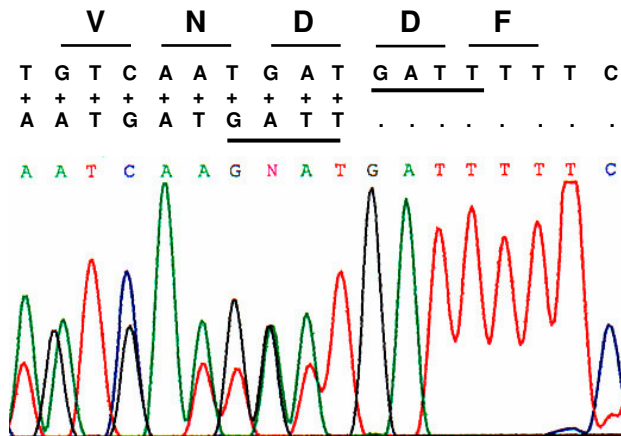


(C) Sequencing of *DGUOK* gene. Detection of mutation in patient 3 and her parents

Patient
 exon 6: c.763_766dupGATT



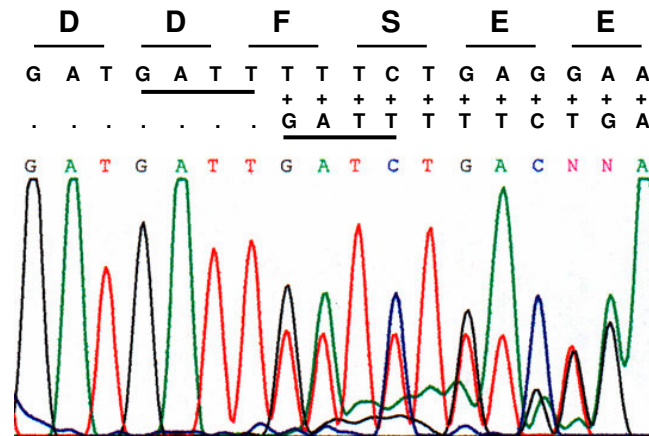
Mother



Antisense sequencing

Normal
 sequence
 Superimposed
 sequence

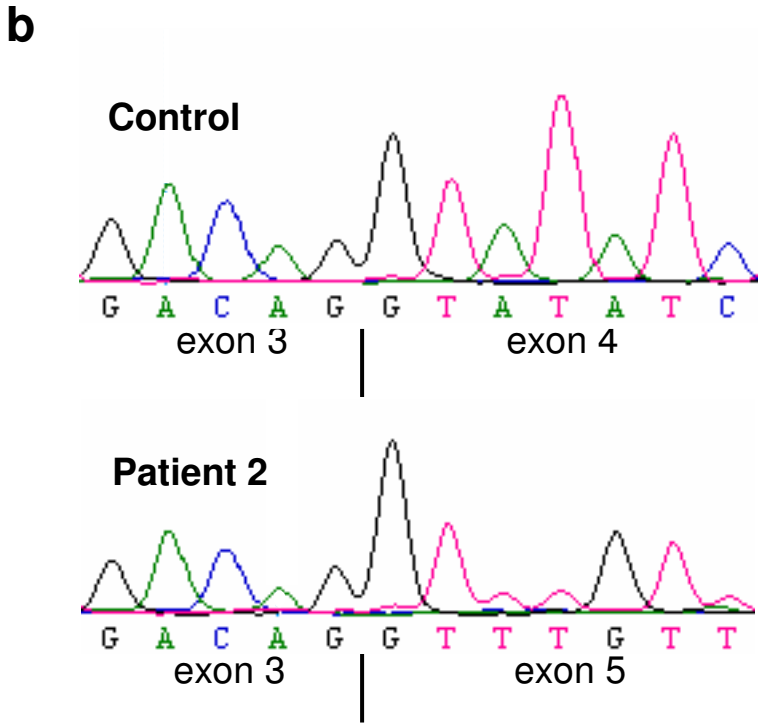
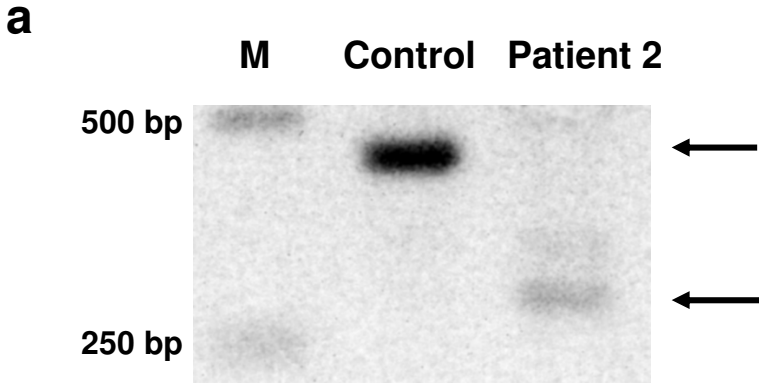
Father



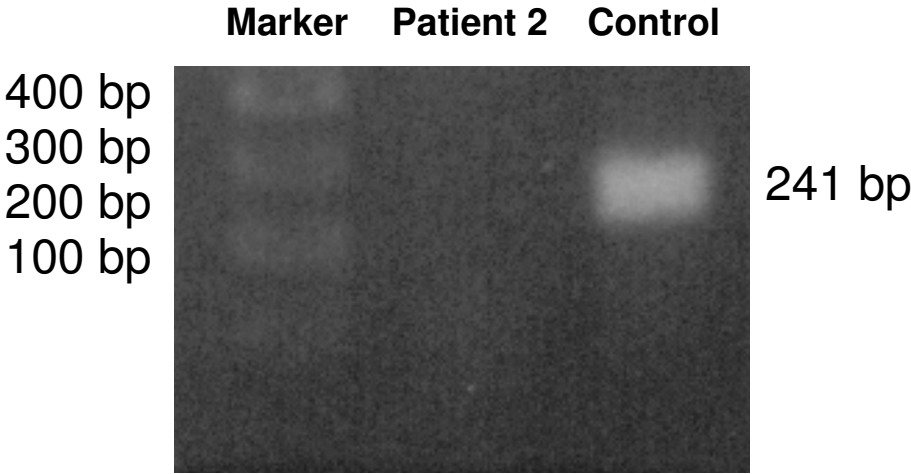
Sense sequencing

Supplementary Figure 2. Skipping of exon 4 in patient 2

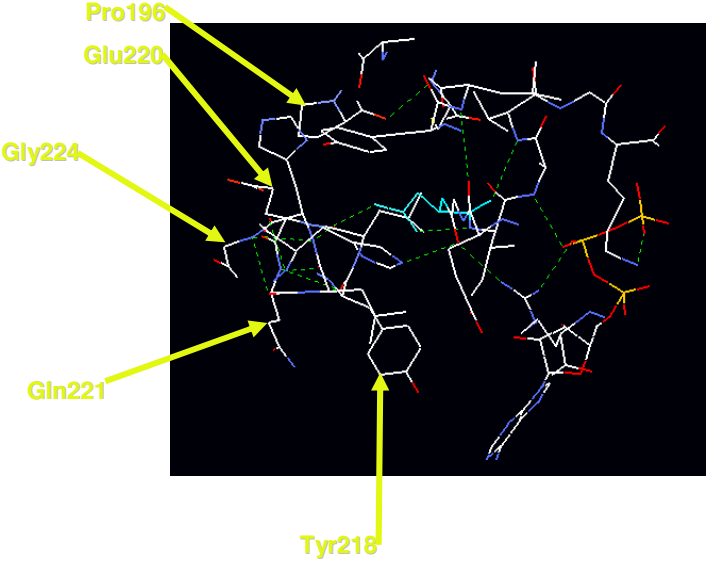
(A) Amplification and sequencing of exons 3-5 from patient 2



(B) Amplification of exons 4-5 from patient 2

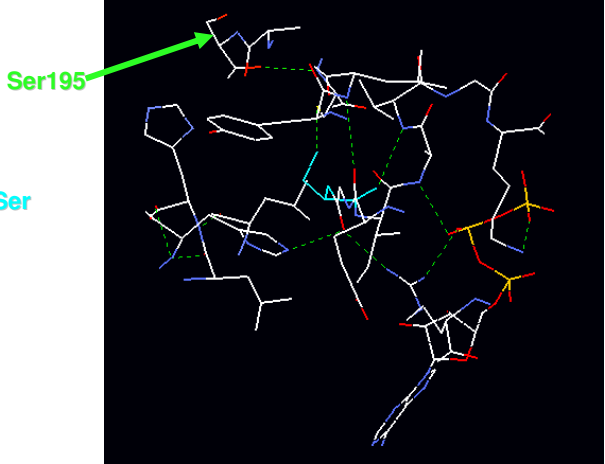


**Supplementary Figure 3. Aminoacid interactions
(A) with wild-type N46 and mutant S46**

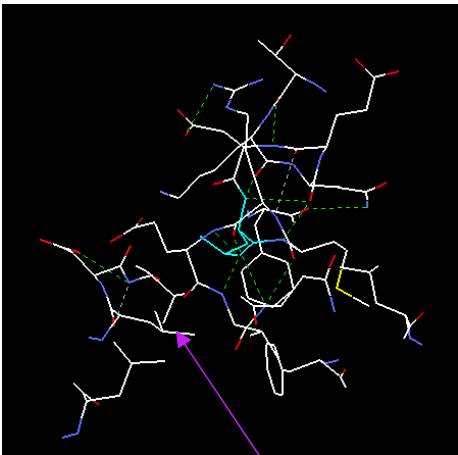


The amino acids in the vicinity of the **Asn 46 (N46)** residue

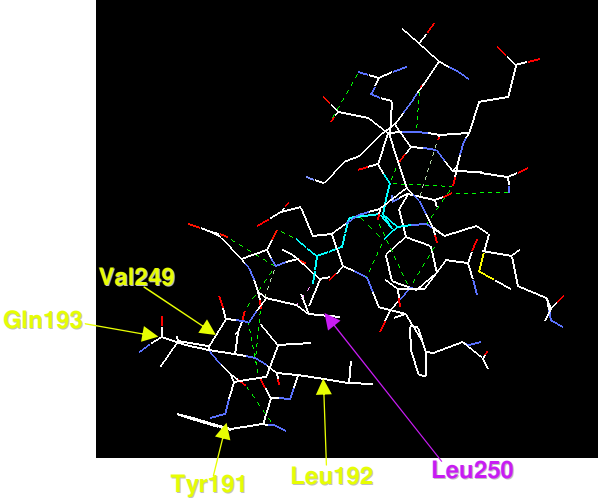
The amino acids in the vicinity of the **mutant Ser 46 (S46)** residue



(B) with wild-type L266 and mutant R266



The amino acids in the vicinity of the **Leu266 (L266)** residue



The amino acids in the vicinity of the **mutant Arg266 (R266)** residue