Supplemental Figure 1

A

| Exon 1 | Exon 2 |
|--------|--------|
| Mother | ...GAG TGT GAG gtg... | ATT AAG ...
| Subject 1 | ...GAG TGT GAG tgt... | ATT AAG ...
| c.78+1G>T |

B

[Genetic sequence and explanation]

C

| Exon 1 | Exon 2 |
|--------|--------|
| Mother | ...CGC GAG TGT GAG ATT AAG TAC ... |
| Subject 1 | ...CGC GAG TAT TAA R E Y |
| p.(Cys25Tyrfs2) |