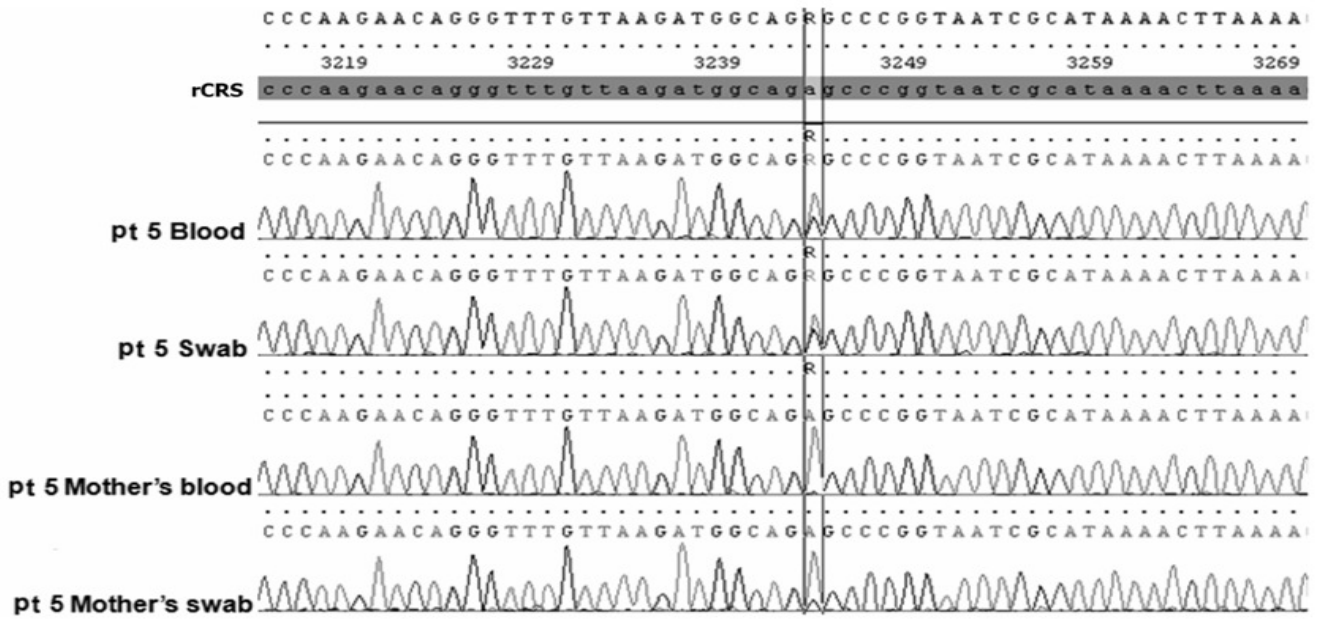


Figure S1. Sequence analysis showing the heteroplasmic 3243A>G mtDNA mutation detected in the patient 5 (pt 5) and her mother (m 5) (swab and blood).



Levels of heteroplasmy of 3243A>G mutation, displayed for each sample between the two lines, were higher in mtDNA from pt 5 than in mtDNA from m5 in both swab and blood samples. rCRS: revised Cambridge Reference Sequence; R: unidentified nucleotide between adenosine and guanosine.