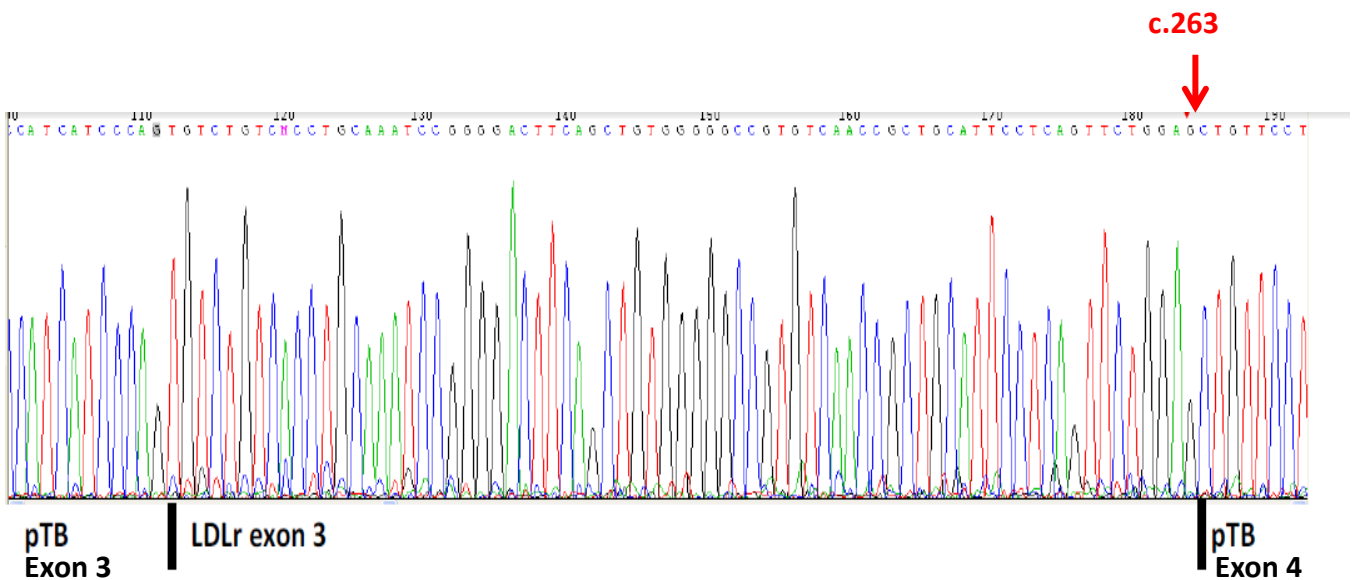


Figure S2. Sequence analysis of the RT-PCR products from plasmid constructions containing the three *LDLR* variants c.313+5G>T (A), c.686A>T (B) and c.2311G>A (C)

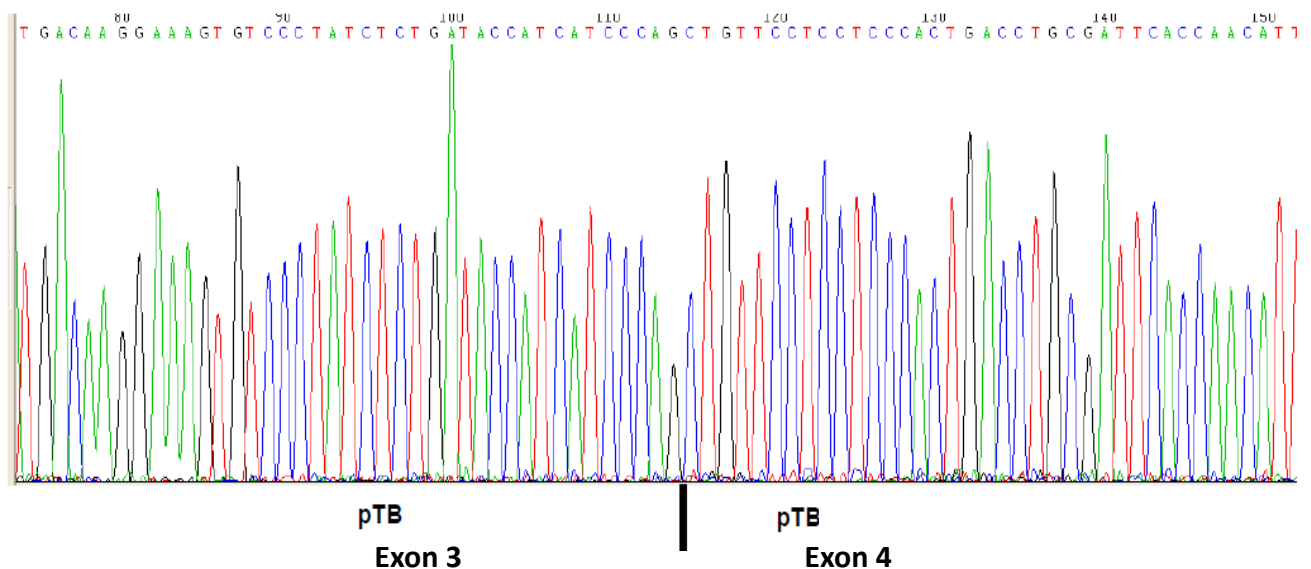
(A) Minigene 313+5A>T

RT-PCR product of the wild type (not shown) = 369 bp.

a) Using of the cryptic site c.263: RT-PCR product = 318 bp.

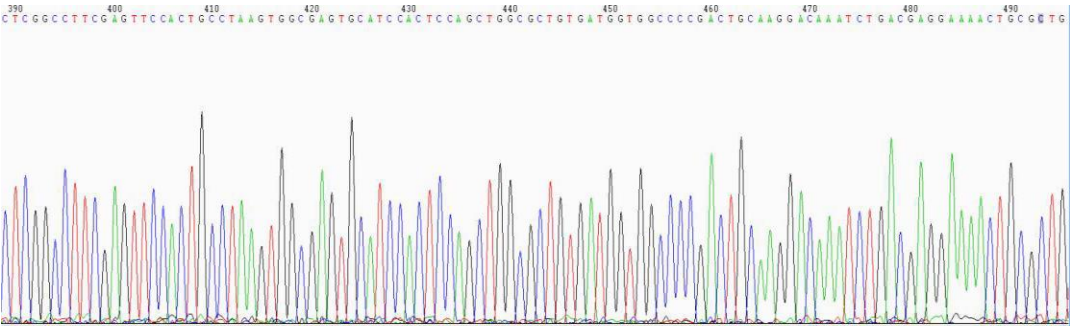


b) Exon skipping: RT-PCR product = 248 bp.



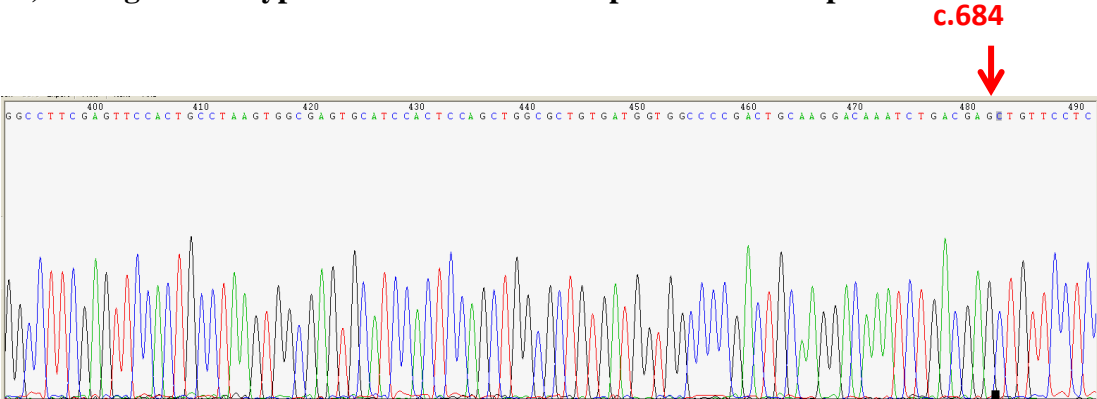
(B) Minigene 686A>T

a) RT-PCR product of the wild type (nature splice site in c.694) = 627 bp.



End of LDLR exon 4 | pTB
Exon 4

b) Using of the cryptic site c.684: RT-PCR product = 617 bp.



c.684

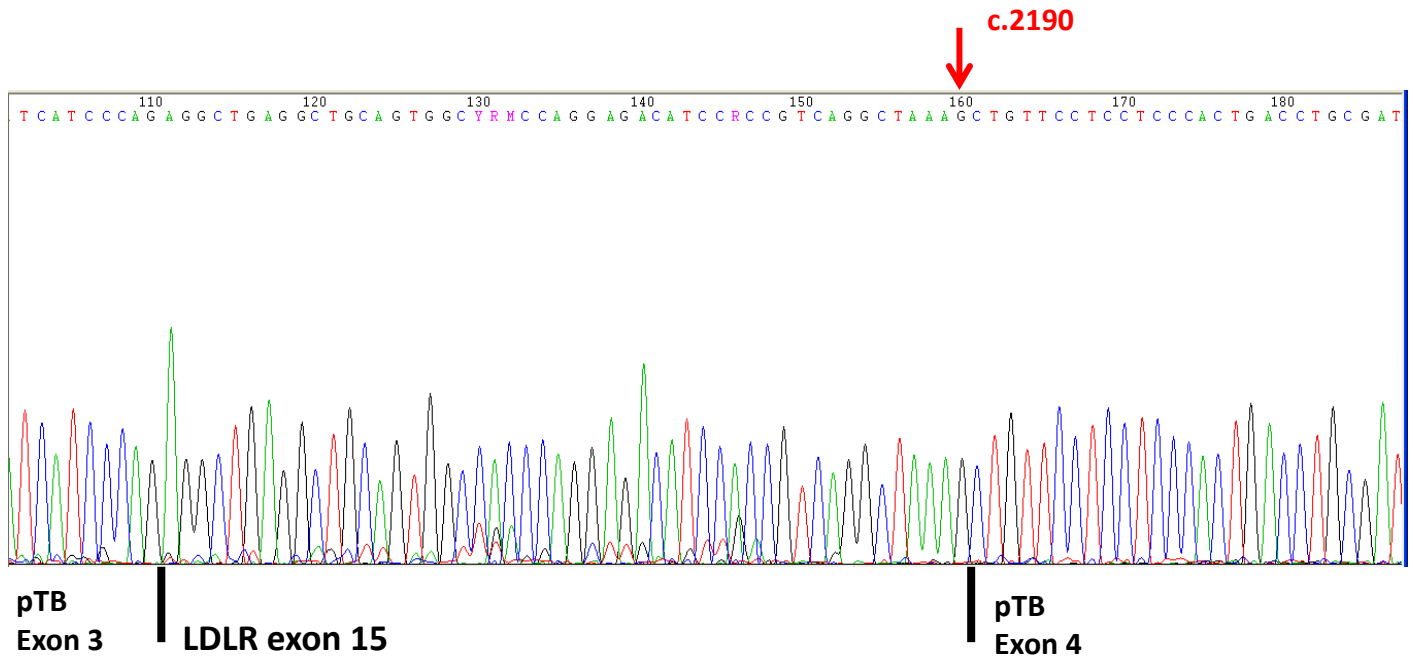


pTB
Exon 4

(C) Minigene 2311G>A

RT-PCR product of the wild type (not shown) = 417 bp.

a) Using of the cryptic site c.2190: RT PCR product =296 bp.



b) Using of the cryptic site c.2311+90: RT PCR product = 507 bp.

