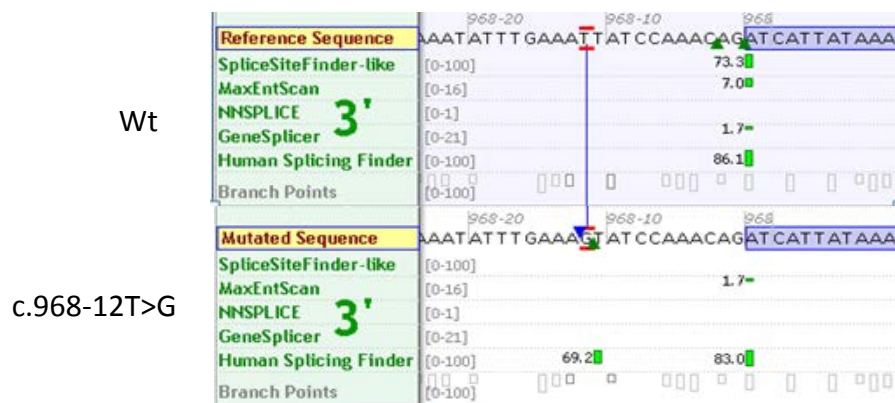
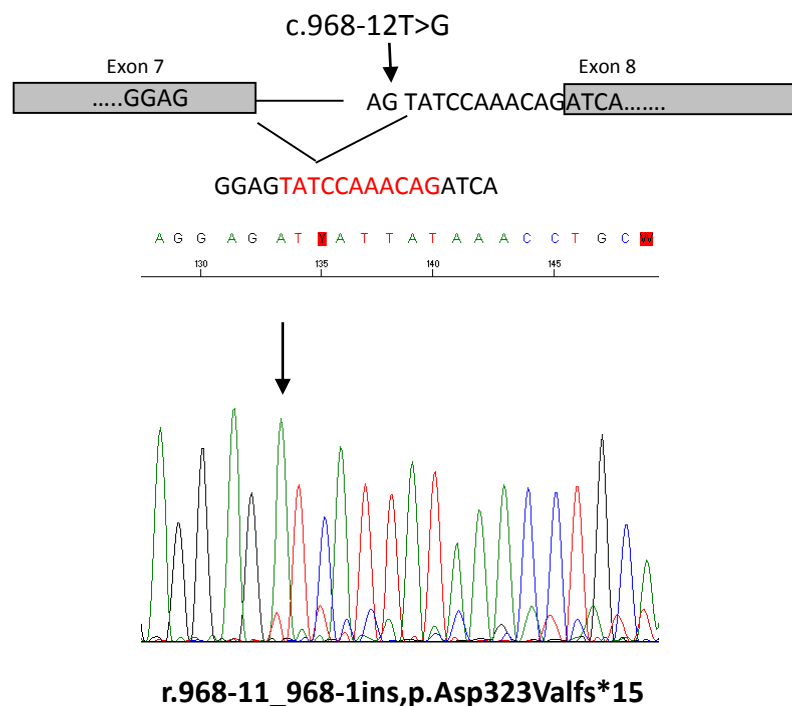


A.



B.



Supplementary figure 1. Analysis of splicing variant c.968-12T>G.(A) In silico analysis by splicing predictions tools Splice Site Finder, MaxEntScan, NnsplICE, GeneSplicer and Human Splicing Finder. The c.968-12T>G variant was predicted to create a cryptic acceptor site by one prediction tool (Human Splicing Finder) (B) Consequences at the cDNA level of the c.968-12T>G variant. This variant leads to the inclusion of the last 11 bases of intron 7 into the mature mRNA. The signal of the alternatively spliced mRNA is low compared to the normal mRNA likely because of its degradation by non-sense mediated mRNA decay.