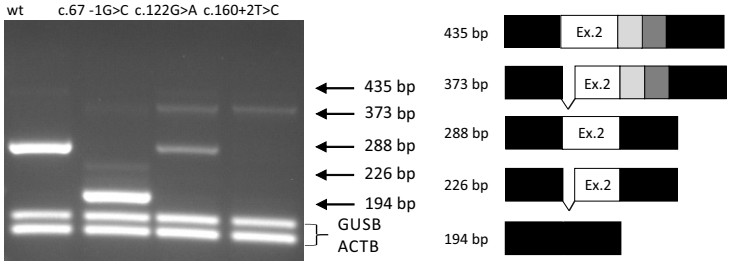
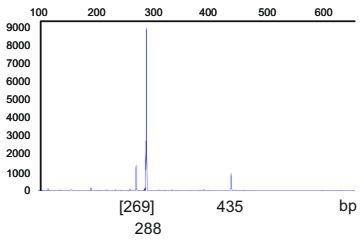
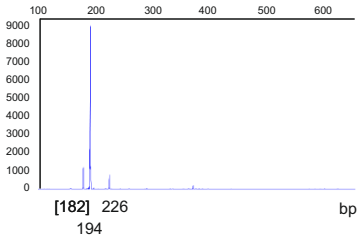
Exon 2



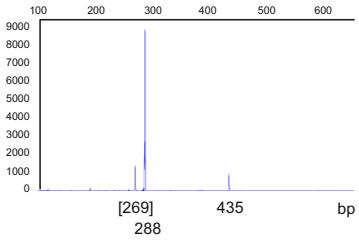
wt



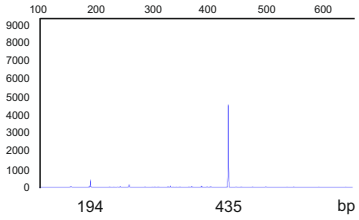
c.67-1G>C



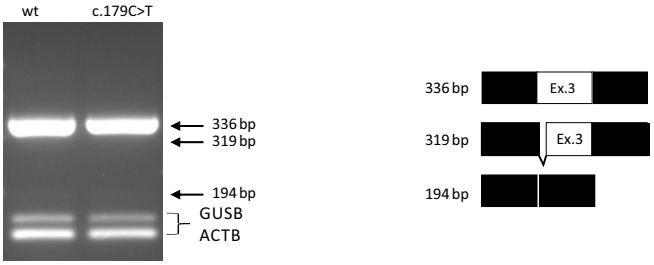
c.122G>A



c.160+2T>C



Exon 3



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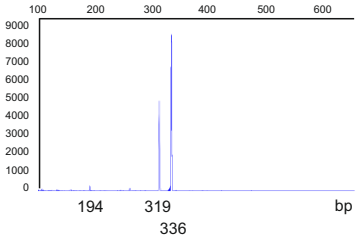
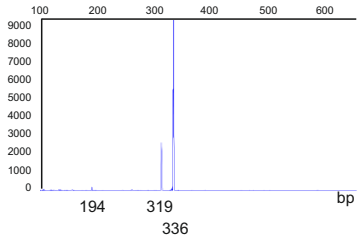
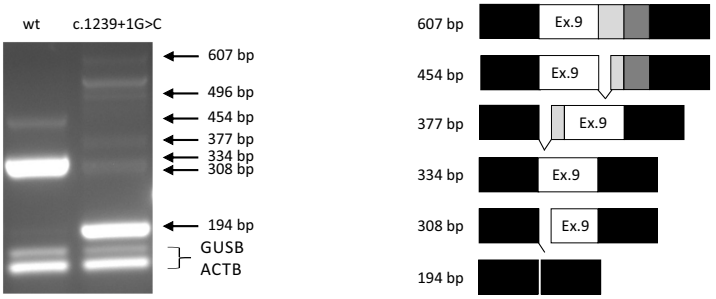


c.179C>T



Exon 9



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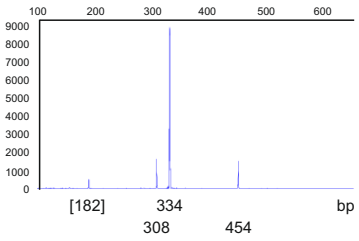
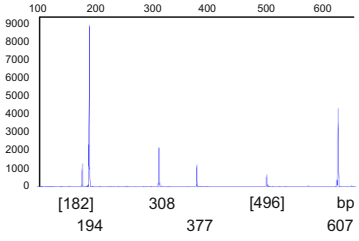
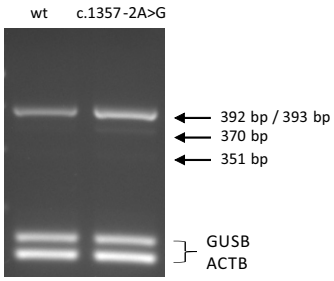
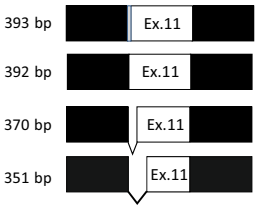


c.1239+1G>C



Exon 11





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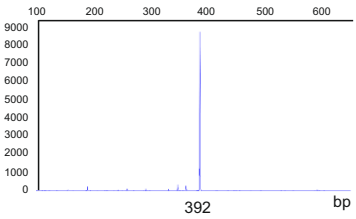
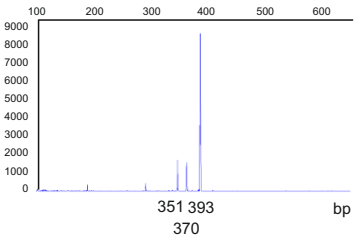
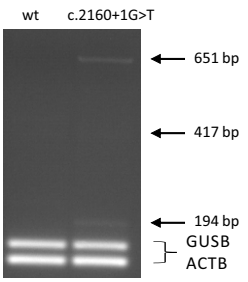
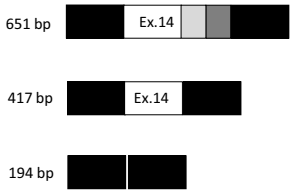


c.1357-2A>G

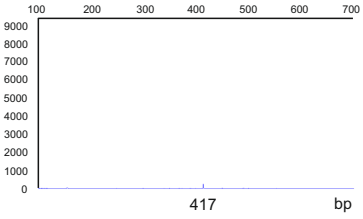


Exon 14

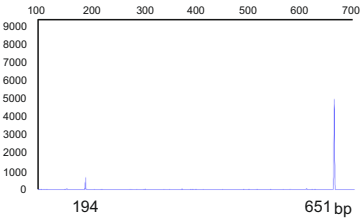




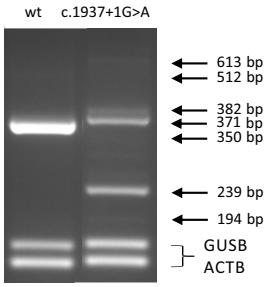
wt

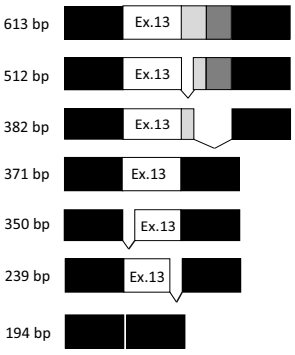


c.2160+1G>T



Exon 13





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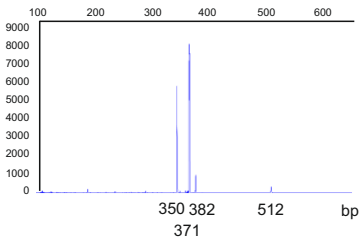
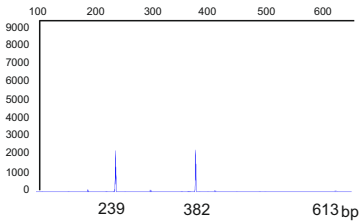
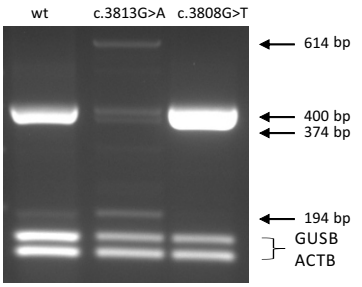
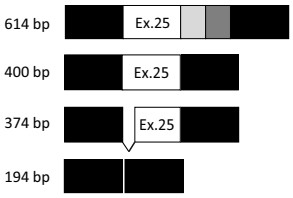


c.1937+1G>A

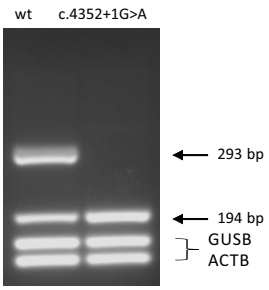


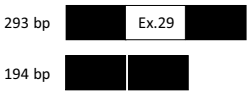
Exon 25





Exon 29





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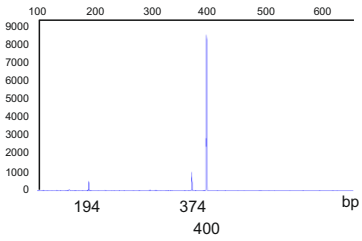
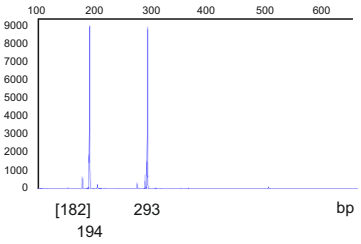
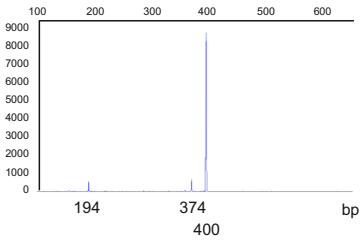


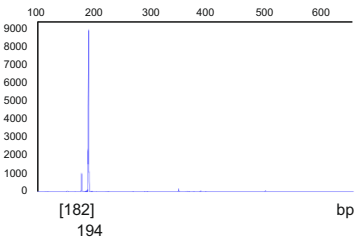
wt



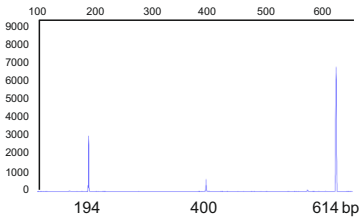
c.3808G>T



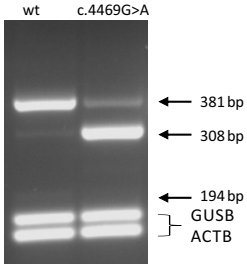
c.4352+1G>A

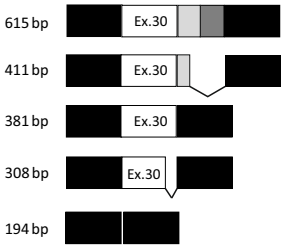


c.3813G>A



Exon 30





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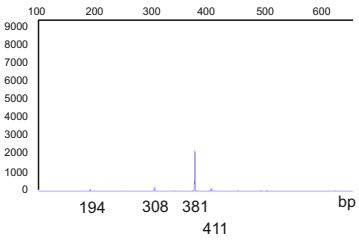
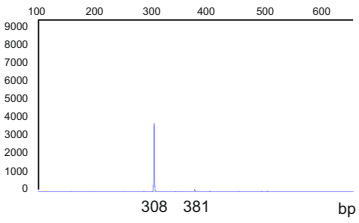
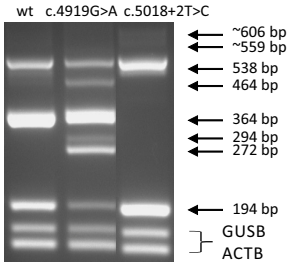
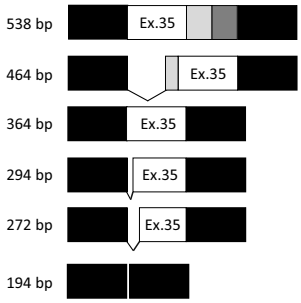


c.4469G>A

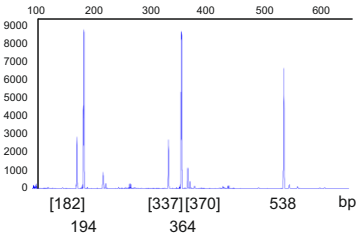


Exon 35

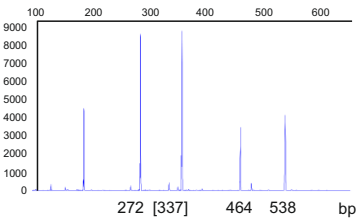




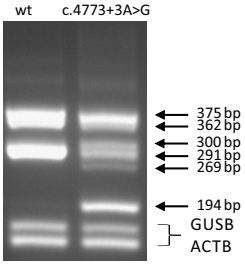
wt

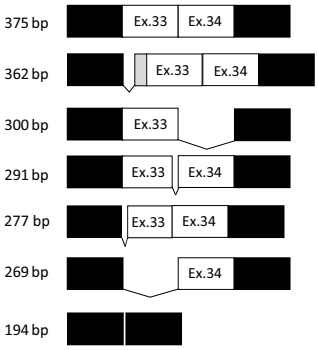


c.4919G>A

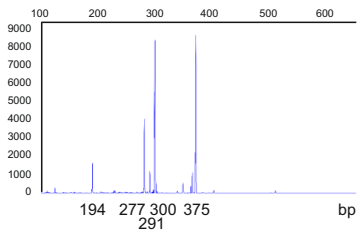


Exon 33 und 34

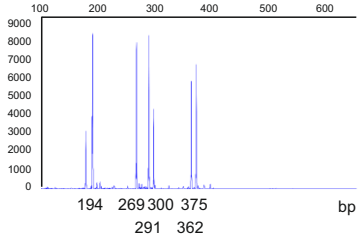




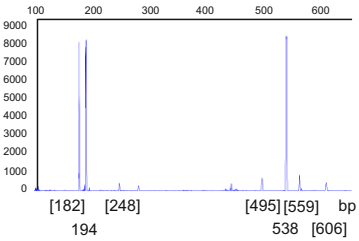
wt



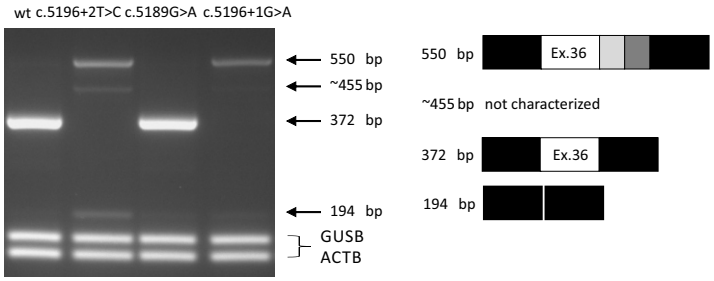
c.4773+3A>G



c.5018+2T>C



Exon 36



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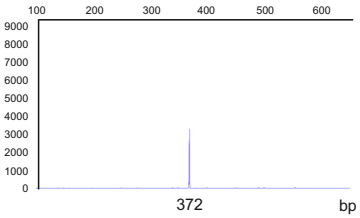
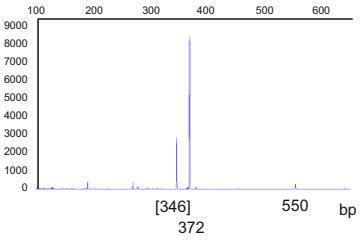
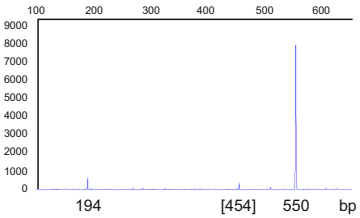


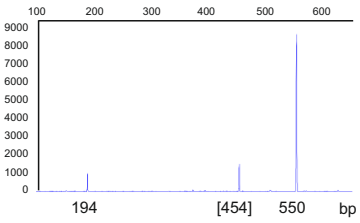
c.5189G>A



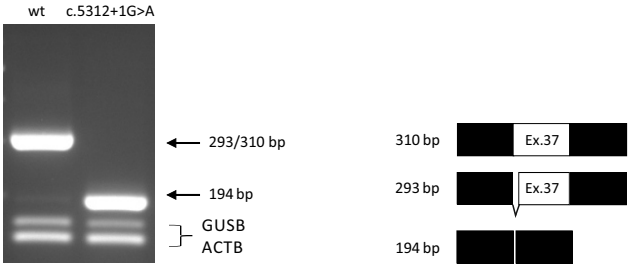
c.5196+1G>A



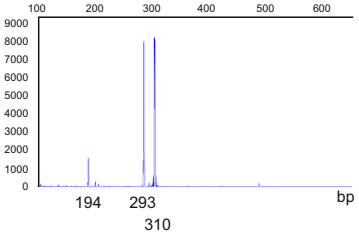
c.5196+2T>C



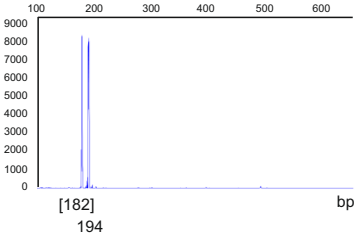
Exon 37



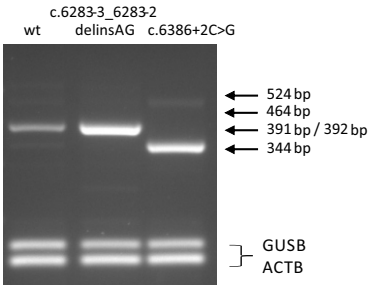
wt

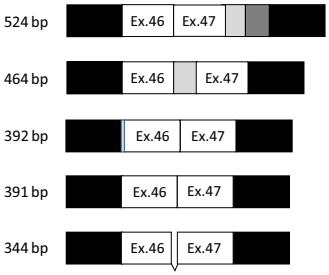


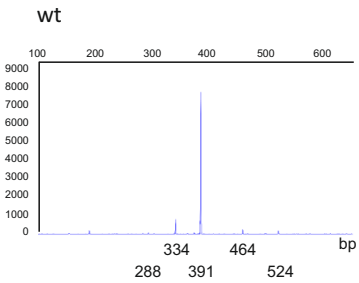
c.5312+1G>A

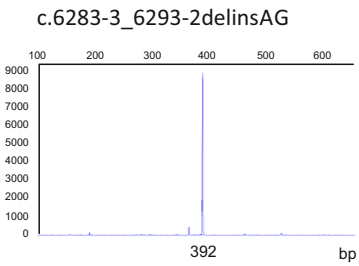


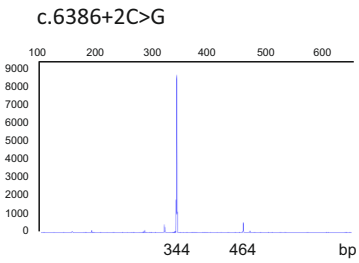
Exon 46 and 47



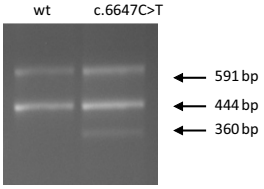


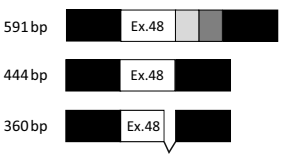






Exon 48





Supplemental Figure 1: Analysis of putative *ABCA4* splicing mutations by *in vitro* splicing assays. Exons carrying putative *ABCA4* splicing mutations (Table 2) were cloned into the pSPL3 vector and the constructs were transfected into HEK293 cells. RNA isolated from these cells was RT-PCR amplified using pSPL3-specific vector primer SD6 and SA4 and the PCR products were directly Sanger sequenced. The sizes of the PCR products were determined by fragment length analysis (except for variants in exon 48). Some of the faint PCR products could not be directly sequenced, their numbers are given in brackets if the nature of the transcripts could not be predicted *in silico*. Images of PCR products separated by gel electrophoresis and electropherograms of fluorescent PCR products are shown for mutants and controls. RT-PCR amplification of GUSB and ACTB served as control. Schematic illustration of the splice products is depicted on the right. Black and dark-grey rectangles represent exonic sequence from the vector, white rectangles are ABCA4 exons and light-grey rectangles indicate ABCA4 specific intronic sequence. Please note that the RNA of cells transfected with a construct containing exon 14 of a control person repeatedly yielded only a very faint band of a 417 bp RT-PCR product which represents the correctly spliced transcript. Analysis of the mutant exon 14, however, obtained misspliced products of 194 bp and 165 bp and was therefore classified as pathogenic.