Supplementary Figure 8. Sequence of the cDNA of patient 9 with the splice site mutation c.4460-1G>C, downstream of the sequence primer g.Ex_34F in intron 34 and including exon 34. Since the start position of the inclusion of intron 33 was not known, all possible reading frames were examined for stop codons. In total, nine stop codons were present (indicated in red), with premature termination affecting exon 34 in all three reading frames.

Intron 33/exon 34 splice site mutation in P9

Original reading frame (1 – 0)

```
tggggaaatattgaaaacacgtctattcagatgaggattctgacagcctccattgtccccc
catagatctaacttgccatttgctgctctcgttgcccttacACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCCTGTCTCCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACTCCCTGCTGACCTCCACCATCAACGGCATTAA
GAAAGTCCTGAAA
```

Alternative reading frame 1 (0 - 2)

```
tggggaaatattgaaaacacgtctattcagatgaggattctgacagcctccattgtccccc
catagatctaacttgccatttgctgctctcgttgcccttacACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCCTGTCTCCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACTCCCTGCTGACCTCCACCATCAACGGCATTAA
GAAAGTCCTGAAA
```

Alternative reading frame 2 (2 - 1)

```
tggggaaatattgaaaacacgtctattcagatgaggattctgacagcctccattgtccccc
catagatctaacttgccatttgctgctctcgttgcccttacACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCCTGTCTCCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACTCCCTGCTGACCTCCACCATCAACGGCATTAA
GAAAGTCCTGAAA
```

TAA; TAG; TGA; stopcodons
C; splice acceptor site mutation IVS33+3753G>C
tggggaaatattgaaaacacg; primer g.Ex_34F
Capital letters indicate exon 34