Supplementary Material 1

Contents

1.1 Plasmid sequence (pcDNA3.1/ Hygro(-))

Length: 5596 bp
MCS with XhoI and BamHI restriction site

GACGGATCGGGAGATCTCCCGATCCCCTATGGTGCACTCTCAGTACAATCTGCT
CTGATGCCGCATAGTTAAGCCAGTATCTGCTCCCTGCTTGTGTGTTGGAGGTCG
CTGAGTAGTGCGCGAGCAAAATTTAAGCTACAACAAGGCAAGGCTTGACCGACA
ATTGCATGAAGAATCTGCTTAGGGTTAGGCGTTTTGCGCTGCTTCGCGATGTAC
GGGCCAGATATACGCGTTGACATTGATTATTGACTAGTTATTAATAGTAATCAAT
TACGGGGTCATTAGTTCATAGCCCATATATGGAGTTCCGCGTTACATAACTTACG
GTAAATGGCCCGCCTGGCTGACCGCCCAACGACCCCCGCCCATTGACGTCAAT
AATGACGTATGTTCCCATAGTAACGCCAATAGGGACTTTCCATTGACGTCAATG
GGTGGAGTATTTACGGTAAACTGCCCACTTGGCAGTACATCAAGTGTATCATAT
GCCAAGTACGCCCCCTATTGACGTCAATGACGGTAAATGGCCCGCCTGGCATTA
TGCCCAGTACATGACCTTATGGGACTTTCCTACTTGGCAGTACATCTACGTATTA
GTCATCGCTATTACCATGGTGATGCGGTTTTGGCAGTACATCAATGGGCGTGGA
TAGCGGTTTGACTCACGGGGATTTCCAAGTCTCCACCCCATTGACGTCAATGGG
AGTTTGTTTTGGCACCAAAATCAACGGGACTTTCCAAAATGTCGTAACAACTCCG
CCCCATTGACGCAAATGGGCGGTAGGCGTGTACGGTGGGAGGTCTATATAAGC
AGAGCTCTCTGGCTAACTAGAGAACCCACTGCTTACTGGCTTATCGAAATTAATA
CGACTCACTATAGGGAGACCCAAGCTGGCTAGCGTTTAAACGGGCCCTCTAGA
CTCGAGCGGCCGCCACTGTGCTGGATATCTGCAGAATTCCACCACACTGGACT
AGTGGATCCGAGCTCGGTACCAAGCTTAAGTTTAAACCGCTGATCAGCCTCGAC
TGTGCCTTCTAGTTGCCAGCCATCTGTTGTTTGCCCCTCCCCCGTGCCTTCCTT
GACCCTGGAAGGTGCCACTCCCACTGTCCTTTCCTAATAAAATGAGGAAATTGC
ATCGCATTGTCTGAGTAGGTGTCATTCTATTCTGGGGGGTGGGGTGGGGCAGG
ACAGCAAGGGGGAGGATTGGGAAGACAATAGCAGGCATGCTGGGGATGCGGT
GGGCTCTATGGCTTCTGAGGCGGAAAGAACCAGCTGGGGCTCTAGGGGGTATC
CCCACGCGCCCTGTAGCGGCGCATTAAGCGCGGCGGGTGTGGTGGTTACGCG
CAGCGTGACCGCTACACTTGCCAGCGCCCTAGCGCCCGCTCCTTTCGCTTTCTT

CCCTTCCTTTCTCGCCACGTTCGCCGGCTTTCCCCGTCAAGCTCTAAATCGGGG
GCTCCCTTTAGGGTTCCGATTTAGTGCTTTACGGCACCTCGACCCCAAAAAACT
TGATTAGGGTGATGGTTCACGTAGTGGGCCATCGCCCTGATAGACGGTTTTTCG
CCCTTTGACGTTGGAGTCCACGTTCTTTAATAGTGGACTCTTGTTCCAAACTGGA
ACAACACTCAACCCTATCTCGGTCTATTCTTTTGATTTATAAGGGATTTTGGCCAT
TTCGGCCTATTGGTTAAAAAATGAGCTGATTTAACAAAAATTTAACGCGAATTAAT
TCTGTGGAATGTGTGTCAGTTAGGGTGTGGAAAGTCCCCAGGCTCCCCAGCAG
GCAGAAGTATGCAAAGCATGCATCTCAATTAGTCAGCAACCAGGTGTGGAAAGT
CCCCAGGCTCCCCAGCAGGCAGAAGTATGCAAAGCATGCATCTCAATTAGTCAG
CAACCATAGTCCCGCCCCTAACTCCGCCCATCCCGCCCCTAACTCCGCCCAGTT
CCGCCCATTCTCCGCCCCATGGCTGACTAATTTTTTTTATTTATGCAGAGGCCGA
GGCCGCCTCTGCCTCTGAGCTATTCCAGAAGTAGTGAGGAGGCTTTTTTGGAG
GCCTAGGCTTTTGCAAAAAGCTCCCGGGAGCTTGTATATCCATTTTCGGATCTG
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TGATCGAAAAGTTCGACAGCGTCTCCGACCTGATGCAGCTCTCGGAGGGCGAA
GAATCTCGTGCTTTCAGCTTCGATGTAGGAGGGCGTGGATATGTCCTGCGGGTA
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CGGCCGCGCTCCCGATTCCGGAAGTGCTTGACATTGGGGAATTCAGCGAGAGC
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GAAACCGAACTGCCCGCTGTTCTGCAGCCGGTCGCGGAGGCCATGGATGCGAT
CGCTGCGGCCGATCTTAGCCAGACGAGCGGGTTCGGCCCATTCGGACCGCAA
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CATGTGTATCACTGGCAAACTGTGATGGACGACACCGTCAGTGCGTCCGTCGC
GCAGGCTCTCGATGAGCTGATGCTTTGGGCCGAGGACTGCCCCGAAGTCCGGC
ACCTCGTGCACGCGGATTTCGGCTCCAACAATGTCCTGACGGACAATGGCCGC
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GGTCGCCAACATCTTCTTCTGGAGGCCGTGGTTGGCTTGTATGGAGCAGCAGA
CGCGCTACTTCGAGCGGAGGCATCCGGAGCTTGCAGGATCGCCGCGGCTCCG
GGCGTATATGCTCCGCATTGGTCTTGACCAACTCTATCAGAGCTTGGTTGACGG
CAATTTCGATGATGCAGCTTGGGCGCAGGGTCGATGCGACGCAATCGTCCGAT
CCGGAGCCGGGACTGTCGGGCGTACACAAATCGCCCGCAGAAGCGCGGCCGT
CTGGACCGATGGCTGTGTAGAAGTACTCGCCGATAGTGGAAACCGACGCCCCA
GCACTCGTCCGAGGGCAAAGGAATAGCACGTGCTACGAGATTTCGATTCCACC
GCCGCCTTCTATGAAAGGTTGGGCTTCGGAATCGTTTTCCGGGACGCCGGCTG
GATGATCCTCCAGCGCGGGGATCTCATGCTGGAGTTCTTCGCCCACCCCAACTT
GTTTATTGCAGCTTATAATGGTTACAAATAAAGCAATAGCATCACAAATTTCACAA
ATAAAGCATTTTTTTCACTGCATTCTAGTTGTGGTTTGTCCAAACTCATCAATGTA
TCTTATCATGTCTGTATACCGTCGACCTCTAGCTAGAGCTTGGCGTAATCATGGT
CATAGCTGTTTCCTGTGTGAAATTGTTATCCGCTCACAATTCCACACAACATACG
AGCCGGAAGCATAAAGTGTAAAGCCTGGGGTGCCTAATGAGTGAGCTAACTCA
CATTAATTGCGTTGCGCTCACTGCCCGCTTTCCAGTCGGGAAACCTGTCGTGCC
AGCTGCATTAATGAATCGGCCAACGCGCGGGGAGAGGCGGTTTGCGTATTGGG
CGCTCTTCCGCTTCCTCGCTCACTGACTCGCTGCGCTCGGTCGTTCGGCTGCG
GCGAGCGGTATCAGCTCACTCAAAGGCGGTAATACGGTTATCCACAGAATCAG
GGGATAACGCAGGAAAGAACATGTGAGCAAAAGGCCAGCAAAAGGCCAGGAAC
CGTAAAAAGGCCGCGTTGCTGGCGTTTTTCCATAGGCTCCGCCCCCCTGACGA
GCATCACAAAAATCGACGCTCAAGTCAGAGGTGGCGAAACCCGACAGGACTAT
AAAGATACCAGGCGTTTCCCCCTGGAAGCTCCCTCGTGCGCTCTCCTGTTCCGA
CCCTGCCGCTTACCGGATACCTGTCCGCCTTTCTCCCTTCGGGAAGCGTGGCG
CTTTCTCATAGCTCACGCTGTAGGTATCTCAGTTCGGTGTAGGTCGTTCGCTCC
AAGCTGGGCTGTGTGCACGAACCCCCCGTTCAGCCCGACCGCTGCGCCTTATC

CGGTAACTATCGTCTTGAGTCCAACCCGGTAAGACACGACTTATCGCCACTGGC
AGCAGCCACTGGTAACAGGATTAGCAGAGCGAGGTATGTAGGCGGTGCTACAG
AGTTCTTGAAGTGGTGGCCTAACTACGGCTACACTAGAAGAACAGTATTTGGTA
TCTGCGCTCTGCTGAAGCCAGTTACCTTCGGAAAAAGAGTTGGTAGCTCTTGAT
CCGGCAAACAAACCACCGCTGGTAGCGGTTTTTTTGTTTGCAAGCAGCAGATTA
CGCGCAGAAAAAAAGGATCTCAAGAAGATCCTTTGATCTTTTCTACGGGGTCTG
ACGCTCAGTGGAACGAAAACTCACGTTAAGGGATTTTGGTCATGAGATTATCAA
AAAGGATCTTCACCTAGATCCTTTTAAATTAAAAATGAAGTTTTAAATCAATCTAA
AGTATATATGAGTAAACTTGGTCTGACAGTTACCAATGCTTAATCAGTGAGGCAC
CTATCTCAGCGATCTGTCTATTTCGTTCATCCATAGTTGCCTGACTCCCCGTCGT
GTAGATAACTACGATACGGGAGGGCTTACCATCTGGCCCCAGTGCTGCAATGAT
ACCGCGAGACCCACGCTCACCGGCTCCAGATTTATCAGCAATAAACCAGCCAG
CCGGAAGGGCCGAGCGCAGAAGTGGTCCTGCAACTTTATCCGCCTCCATCCAG
TCTATTAATTGTTGCCGGGAAGCTAGAGTAAGTAGTTCGCCAGTTAATAGTTTGC
GCAACGTTGTTGCCATTGCTACAGGCATCGTGGTGTCACGCTCGTCGTTTGGTA
TGGCTTCATTCAGCTCCGGTTCCCAACGATCAAGGCGAGTTACATGATCCCCCA
TGTTGTGCAAAAAAGCGGTTAGCTCCTTCGGTCCTCCGATCGTTGTCAGAAGTA
AGTTGGCCGCAGTGTTATCACTCATGGTTATGGCAGCACTGCATAATTCTCTTAC
TGTCATGCCATCCGTAAGATGCTTTTCTGTGACTGGTGAGTACTCAACCAAGTCA
TTCTGAGAATAGTGTATGCGGCGACCGAGTTGCTCTTGCCCGGCGTCAATACG
GGATAATACCGCGCCACATAGCAGAACTTTAAAAGTGCTCATCATTGGAAAACG
TTCTTCGGGGCGAAAACTCTCAAGGATCTTACCGCTGTTGAGATCCAGTTCGAT
GTAACCCACTCGTGCACCCAACTGATCTTCAGCATCTTTTACTTTCACCAGCGTT
TCTGGGTGAGCAAAAACAGGAAGGCAAAATGCCGCAAAAAAGGGAATAAGGGC
GACACGGAAATGTTGAATACTCATACTCTTCCTTTTTCAATATTATTGAAGCATTT
ATCAGGGTTATTGTCTCATGAGCGGATACATATTTGAATGTATTTAGAAAAATAAA
CAAATAGGGGTTCCGCGCACATTTCCCCGAAAAGTGCCACCTGACGTC

1.2 Inserted SDHB minigene sequence

Total length minigene plasmid: 8441 bp
Total length of insert: 2905 bp

Amplicon 1 (693bp):

5' end - (plasmid sequence): TAGCGTTTAAACGGGCCCTCTAGAC
(202 bp 3' flanking Intron 1):
GCCCCTAATACCATGTCTGATATATTAGCATATGATAGGTGCTTAATACTTGTTG
GATATTGAATGCCTGCCTTTTCTAAGAAGATTATGTGTCAGTAATGTGTGAGTTT
ATATCCAGCGTTACATCTGTTGTGCCAGCAAAATGGAATTATCTTGTATTTCTAAT
TTTTTTTTCCTTTTTGTGAACTTTAAAAAATTTCAG
(128 bp Exon 2):
GCCTCCCGAGGAGCCCAGACAGCTGCAGCCACAGCTCCCCGTATCAAGAAATT
TGCCATCTATCGATGGGACCCAGACAAGGCTGGAGACAAACCTCATATGCAGAC
TTATGAAGTTGACCTTAATAA
(325 bp 5' flanking Intron 2):

GTGAGTATCTCTGTGAAAGCCAGCTATTGAAGGAGAGTTCTTGATTTGATTTAGG
GACATGCTTTTCACATCCTTGGAAGGCTTAAAAATCTGAGATCATCCGATGGCTC
TGTTTTTAAAACAAGAATGAGCTTGGGCCGGGCACGGTGGCTCACGCCTATAAT
CCCAGCACTTTGGGAGGCTCAGGCGGGCAGATCATCTGAGGTTGGGAGTTCAA
GACCAATGTGGCCAACATGGTGAAACTCCCCTCTCTACTAAAAATACAGAAATTA
GCTGGGTATAGTGGCTCATGCCTGTAATCCCAGCCACTCAGGAGGCTGGGGCA

Amplicon 2 (759):

(325 bp 3’ flanking Intron 2):
AAATTATTTGGAAAAACTTTGCAGGGAGCTTCAGAAGTAAAAAACAAAAACTCGG
CCACTGTATGGAAGTTGTGTCATACTTGTCTACTACTACTTTAGCTTTCCATTTCC
TAGAAATGTGTTTGCACTGTGATCATCAAAATAGAACTGTGCAGTACTTAAAATG
TAAGAATCTGCAAAAACAAAACTACAGAACAAATTCCGAAGGTGACCTGAGAAG
ACCAAATGGATAAGCTAATACATCCAGGTGTCTCCGATTATATTATGATAAAGTG
TAGGGAGGTTGAACTTTACATAAATACCACTGGATATTTTTCTTTGTTAG
(86 bp Exon 3):
ATGTGGCCCCATGGTATTGGATGCTTTAATCAAGATTAAGAATGAAGTTGACTCT
ACTTTGACCTTCCGAAGATCATGCAGAGAAG
(325 bp 5’ flanking Intron 3):
GTGAGCATTTCATTCCTGTTGGGCTCCAGATACTTGTGGTCTTCCAAAGAGGCT
TGGGCTGGCCAAAGCTGATAGAGACATCTGGAAAACAGCTGCAGCTCTCTGCT
GGGTGAACTTGCTGTGCAACCTAATTCCTTTTCTTGGGTAAATGCATCCAAAATT
CTAACCATGGTGCAAGTTCTGGTTTCTGAGTTTACTTTCCTTCAGGAGCATTCTA
TCCTGTGGTTACTGCATCTGGCGGTCCCTGTGAGCTGCTCCACGCTTCAAGGC
CTAGCTAGGACCTGCTGGCTGGCACCTTCCATGCTGCTGAGCTCTTTTATTAAG

Amplicon 3 (1553bp):

(325 bp 3’ flanking Intron 3):
TGGAGCTAGAACTGCTGTGGAAACTCCATGGTGCCAGCAGCGCCCAGGCCTCT
GGCCTCCTCCAGTGCCATACTTGGCATGTGGCTTCTAACCTCAAGGTTGCCACA
TGGTCATAGAGTAGTTTTTGGAACTCAAGCCATTAGAACCACATTCGAGGCAGG
AAGAAGGAGGGAAGGGAGAAAAGCCAACAGGCACCTCTGTCAGAGGAATGTTG
CATGTCAGTGCTGCCCCTGATGGCACAGCAAGGAGGATCCAGAAGAAAGTATTT
GGGGCAGGACTGATTCCGGATATGGGTGAGGATGTGTTAAATGTGTGTCTCTTT
CAG
(137 bp Exon 4):
GCATCTGTGGCTCTTGTGCAATGAACATCAATGGAGGCAACACTCTAGCTTGCA
CCCGAAGGATTGACACCAACCTCAATAAGGTCTCAAAAATCTACCCTCTTCCACA
CATGTATGTGATAAAGGATCTTGTTCCC
(734 bp Intron 4):
GTGAGTTTCTGCATCTCTCTGGTTTTGTTTTTATTTGCATGGGGGGCAGTGCTAT
GTGTGTTACGCTATTAGTTTTTCAGGGCAGGATTTGTTTGAGTATTTAAGGCTCC
TCTTCTAGTCAGATAGTAAATAGATGGGACTGTTTCCTATATTATGGCAGGAGGA
GGACCACAGTGTCACTGAGAGATTAGGAAAATCTCCAGATCTGAGACAGTGTGG
CCAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAGAAAATCCTC
CAGCAGCTCATTTGGTGAGGTTTGTTTGAGAATAATGGGGCACAGAATTTTAGA
ATTGGAACATTCCCAGAAATGAATAATTTGGCCATAACTACAAAGAAATCATGTG
GAAAATGCCAGTTGGGAAAGCAACAGCTGTTTATGAGTCCCACTTTAATTTCTAC
CTCCCTATATTTGAACACCTTTGCTGACGTGGTGTTTTCAAGGGCAGAAAGGAC

TGTGGGATTGGCAGGTTTCCCAAGTAGGTGAGGGAGAGGTTTCCTAACCATGCT
TAACATTGTGACCATTTTTCCTTTTGTAATCTTTGCATATATTCATGACAAAGGAA
AAGAGTCTGGCTTCTGCCTGGCATAGAGTGGACGAGTAGTCAGTGTCCAAGAAA
TGGGGTAAATAAAGCTGAGGTGATGATGGAATCTGATCCTTTTCTTCTTCTTCTT
CTTCTTCTTCCTCTTAACCACAG
(117 bp Exon 5):
GATTTGAGCAACTTCTATGCACAGTACAAATCCATTGAGCCTTATTTGAAGAAGA
AGGATGAATCTCAGGAAGGCAAGCAGCAGTATCTGCAGTCCATAGAAGAGCGT
GAGAAACTG
(202 bp 5' flanking Intron 5):
GTCATTAGTCCCTATTTATTGTTTCAATCTGAAGAATTTATTGCAAAGATGATTGC
CAGGAGTGTGGCTTGGCAGGGAACATAAGCTGATGCAAGCCATTTAGTGTATTC
TGGAGAGGAACTGGCACAAGGGTGGGGCACTCATCCTCTTAGGGCCGGGTGA
GTAGTGTGAACCCGTGAATGGCCCAGAGTGGACCAAGGAG
(flanking plasmid sequence): CGAGCTCGGTACCAAGCTTAAGTTT **– 3' end**

1.3 Primer sequences

Primers were ordered and produced by metabion (https://www.metabion.com/)

Name	T_m [°C]	Sequence 5'-3'	Use
Exon 2_HYG (-)_Fwd	63	tagcgtttaaacgggccctctagacGCCCCTAATAC CATGTCTGATATATTAGCATATGATA	Amplicon PCR Minigene
Exon 2_rev	75	ttccaaataatttTGCCCCAGCCTCCTGAGTGG	Amplicon PCR Minigene
Exon 3_fwd	58	gaggctggggcaAAATTATTTGGAAAAACTTT GCAG	Amplicon PCR Minigene
Exon 3_rev	58	gttctagctccaCTTAATAAAAGAGCTCAGCAG	Amplicon PCR Minigene
Exon 4.5_fwd	67	ctcttttattaagTGGAGCTAGAACTGCTGTGG	Amplicon PCR Minigene
Exon 4.5_Rev HYG(-)	61	aaacttaagcttggtaccgagctcgCTCCTTGGTCC ACTCTGGGC	Amplicon PCR Minigene
MG_Val_Fwd	58	TAATACGACTCACTATAGGGAGACCC	MG plasmid insert validation
MG Val Rev	58	CTGGCAACTAGAAGGCACAG	MG plasmid insert validation
RT_PCR_plasmid_fwd	58	AAGCTGGCTAGCGTTTAAACG	RT-PCR
RT_PCR_plasmid_rev	57	TAAACTTAAGCTTGGTACCGAGC	RT-PCR
MG_V1_Fwd	70	CCGAAGATCATGCAGAGAAGATGAGCAT TTCATTCCTGTTGGGCTCCAGA	c.286+1G>A
MG_V1_Rev	67	CAACAGGAATGAAATGCTCATCTTCTCTG CATGATCTTCGGAAGGTCAAA	c.286+1G>A
MG_V2_Fwd	63	TATGAAGTTGACCTTAATAAATGAGTATCT CTGTGAAAGCCAGCTATTGA	c.200+1G>A
MG_V2_Rev	64	GGCTTTCACAGAGATACTCATTTATTAAG GTCAACTTCATAAGTCTGCAT	c.200+1G>A
MG_V3_Fwd	69	GTTAAATGTGTGTCTCTTTCGGGCATCTG TGGCTCTTGTGCAATGAACAT	c.287-2A>T
MG_V3_Rev	68	GCACAAGAGCCACAGATGCCCGAAAGAG ACACACATTTAACACATCCTCA	c.287-2A>T

MG_V4_Fwd	65	GGCCCCATGGTATTGGATGCCTTAATCAAGATTAAGAATGAAGTTGACTC	c.225T>C
MG_V4_Rev	65	TCATTCTTAATCTTGATTAAGGCATCCAATACCATGGGGCCACATCTAAC	c.225T>C
MG_V5_Fwd	70	CTTTCAGGCATCTGTGGCTCCTGTGCAATGAACATCAATGGAGGCAACAC	c.300T>C
MG_V5_Rev	70	CCATTGATGTTCATTGCACAGGAGCCACAGATGCCTGAAAGAGACACACA	c.300T>C
MG_V7_Fwd	64	GACTTATGAAGTTGACCTTAGTAAGTGAGTATCTCTGTGAAAGCCAGCTA	c.197A>G
MG_V7_Rev	62	TTCACAGAGATACTCACTTACTAAGGTCAACTTCATAAGTCTGCATATGA	c.197A>G
MG_V8_Fwd	62	AAGTTGACCTTAATAAGTGACTATCTCTGTGAAAGCCAGCTATTGAAGGA	c.200+5G>C
MG_V8_Rev	65	AGCTGGCTTTCACAGAGATAGTCACTTATTAAGGTCAACTTCATAAGTCT	c.200+5G>C
MG_V9_Fwd	64	GAGAAGACCAAATGGATAAGGTAATACATCCAGGTGTCTCCGATTATATT	c.201-91C>G
MG_V9_Rev	66	GGAGACACCTGGATGTATTACCTTATCCATTTGGTCTTCTCAGGTCACCT	c.201-91C>G
MG_V10_Fwd	65	TACATAAATACCACTGGATAGTTTTCTTTGTTAGATGTGGCCCCATGGTA	c.201-14T>G
MG_V10_Rev	61	GCCACATCTAACAAAGAAAACTATCCAGTGGTATTTATGTAAAGTTCAAC	c.201-14T>G
MG_V11_Fwd	64	TTACATAAATACCACTGGATATTTTTCTTAGTTAGATGTGGCCCCATGGT	c.201-6T>A
MG_V11_Rev	64	ACCATGGGGCCACATCTAACTAAGAAAAATATCCAGTGGTATTTATGTAA	c.201-6T>A
MG_V12_Fwd	66	ACTGGATATTTTTCTTTGTTTGATGTGGCCCCATGGTATTGGATGCTTTA	c.201-2A>T
MG_V12_Rev	64	CAATACCATGGGGCCACATCAAACAAAGAAAAATATCCAGTGGTATTTAT	c.201-2A>T
MG_V13_Fwd	64	ACTGGATATTTTTCTTTGTTACATGTGGCCCCATGGTATTGGATGCTTTA	c.201-1G>C
MG_V13_Rev	65	CCAATACCATGGGGCCACATGTAACAAAGAAAAATATCCAGTGGTATTTA	c.201-1G>C
MG_V14_Fwd	62	TGGTATTGGATGCTTTAATCTAGATTAAGAATGAAGTTGACTCTACTTTG	c.232A>T
MG_V14_Rev	65	GTCAACTTCATTCTTAATCTAGATTAAAGCATCCAATACCATGGGGCCAC	c.232A>T
MG_V15_Fwd	69	TCCGAAGATCATGCAGAGAACGTGAGCATTTCATTCCTGTTGGGCTCCAG	c.286G>C
MG_V15_Rev	68	AACAGGAATGAAATGCTCACGTTCTCTGCATGATCTTCGGAAGGTCAAAG	c.286G>C
MG_V16_Fwd	69	CGAAGATCATGCAGAGAAGGAGAGCATTTCATTCCTGTTGGGCTCCAGAT	c.286+2T>A
MG_V16_Rev	68	CCAACAGGAATGAAATGCTCTCCTTCTCTGCATGATCTTCGGAAGGTCAA	c.286+2T>A
MG_V17_Fwd	67	GAAGATCATGCAGAGAAGGTCAGCATTTCATTCCTGTTGGGCTCCAGATA	c.286+3G>C
MG_V17_Rev	69	CCCAACAGGAATGAAATGCTGACCTTCTCTGCATGATCTTCGGAAGGTCA	c.286+3G>C
MG_V18_Fwd	69	AAGATCATGCAGAGAAGGTGTGCATTTCATTCCTGTTGGGCTCCAGATAC	c.286+4A>T
MG_V18_Rev	69	GCCCAACAGGAATGAAATGCACACCTTCTCTGCATGATCTTCGGAAGGTC	c.286+4A>T
MG_V19_Fwd	68	AGATCATGCAGAGAAGGTGACCATTTCATTCCTGTTGGGCTCCAGATACT	c.286+5G>C
MG_V19_Rev	71	AGCCCAACAGGAATGAAATGGTCACCTTCTCTGCATGATCTTCGGAAGGT	c.286+5G>C

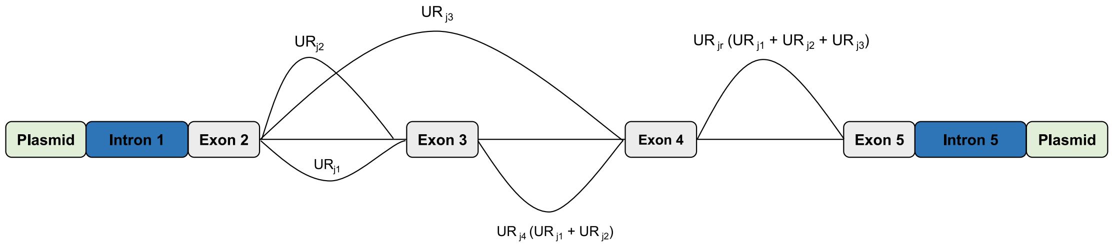
MG_V20_Fwd	68	GTGAGGATGTGTTAAATGTGAGTCTCTTTCAGGCATCTGTGGCTCTTGTG	c.287-12T>A
MG_V20_Rev	67	CACAGATGCCTGAAAGAGACTCACATTTAACACATCCTCACCCATATCCG	c.287-12T>A
MG_V21_Fwd	70	GAGGATGTGTTAAATGTGTGGCTCTTTCAGGCATCTGTGGCTCTTGTGCA	c.287-10T>G
MG_V21_Rev	67	GCCACAGATGCCTGAAAGAGCCACACATTTAACACATCCTCACCCATATC	c.287-10T>G
MG_V22_Fwd	69	GGATGTGTTAAATGTGTGTCGCTTTCAGGCATCTGTGGCTCTTGTGCAAT	c.287-8T>G
MG_V22_Rev	68	GAGCCACAGATGCCTGAAAGCGACACACATTTAACACATCCTCACCCATA	c.287-8T>G
MG_V23_Fwd	68	ATGTGTTAAATGTGTGTCTCGTTCAGGCATCTGTGGCTCTTGTGCAATGA	c.287-6T>G
MG_V23_Rev	69	AAGAGCCACAGATGCCTGAACGAGACACACATTTAACACATCCTCACCCA	c.287-6T>G
MG_V24_Fwd	68	TGTGTTAAATGTGTGTCTCTGTCAGGCATCTGTGGCTCTTGTGCAATGAA	c.287-5T>G
MG_V24_Rev	69	CAAGAGCCACAGATGCCTGACAGAGACACACATTTAACACATCCTCACCC	c.287-5T>G
MG_V25_Fwd	68	TGTTAAATGTGTGTCTCTTTGAGGCATCTGTGGCTCTTGTGCAATGAACA	c.287-3C>G
MG_V25_Rev	68	CACAAGAGCCACAGATGCCTCAAAGAGACACACATTTAACACATCCTCAC	c.287-3C>G
MG_V26_Fwd	66	TTAAATGTGTGTCTCTTTCATGCATCTGTGGCTCTTGTGCAATGAACATC	c.287-1G>T
MG_V26_Rev	69	TGCACAAGAGCCACAGATGCATGAAAGAGACACACATTTAACACATCCTC	c.287-1G>T
MG_V27_Fwd	67	AAGGATTGACACCAACCTCAGTAAGGTCTCAAAAATCTACCCTCTTCCAC	c.365A>G
MG_V27_Rev	67	GGTAGATTTTTGAGACCTTACTGAGGTTGGTGTCAATCCTTCGGGTGCAA	c.365A>G
MG_V28_Fwd	67	GACACCAACCTCAATAAGGTATCAAAAATCTACCCTCTTCCACACATGTA	c.372C>A
MG_V28_Rev	66	GGAAGAGGGTAGATTTTTGATACCTTATTGAGGTTGGTGTCAATCCTTCG	c.372C>A
MG_V29_Fwd	67	AATCTACCCTCTTCCACACAGGTATGTGATAAAGGATCTTGTTCCCGTGA	c.398T>G
MG_V29_Rev	64	CAAGATCCTTTATCACATACCTGTGTGGAAGAGGGTAGATTTTTGAGACC	c.398T>G
MG_V30_Fwd	66	ATCTACCCTCTTCCACACATCTATGTGATAAAGGATCTTGTTCCCGTGAG	c.399G>C
MG_V30_Rev	64	ACAAGATCCTTTATCACATAGATGTGTGGAAGAGGGTAGATTTTTGAGAC	c.399G>C
MG_V31_Fwd	69	TCTACCCTCTTCCACACATGGATGTGATAAAGGATCTTGTTCCCGTGAGT	c.400T>G
MG_V31_Rev	63	AACAAGATCCTTTATCACATCCATGTGTGGAAGAGGGTAGATTTTTGAGA	c.400T>G
MG_V32_Fwd	68	CTACCCTCTTCCACACATGTTTGTGATAAAGGATCTTGTTCCCGTGAGTT	c.401A>T
MG_V32_Rev	64	GAACAAGATCCTTTATCACAAACATGTGTGGAAGAGGGTAGATTTTTGAG	c.401A>T
MG_V33_Fwd	67	TACCCTCTTCCACACATGTAAGTGATAAAGGATCTTGTTCCCGTGAGTTT	c.402T>A
MG_V33_Rev	63	GGAACAAGATCCTTTATCACTTACATGTGTGGAAGAGGGTAGATTTTTGA	c.402T>A
MG_V34_Fwd	67	ACCCTCTTCCACACATGTATTTGATAAAGGATCTTGTTCCCGTGAGTTTC	c.403G>T
MG_V34_Rev	64	GGGAACAAGATCCTTTATCAAATACATGTGTGGAAGAGGGTAGATTTTTG	c.403G>T

MG_V35_Fwd	66	TGATAAAGGATCTTGTTCCCTTGAGTTTC TGCATCTCTCTGGTTTTGTTT	c.423+1G>T
MG_V35_Rev	68	CAGAGAGATGCAGAAACTCAAGGGAACA AGATCCTTTATCACATACATGT	c.423+1G>T
MG_V36_Fwd	68	GATAAAGGATCTTGTTCCCGGGAGTTTCT GCATCTCTCTGGTTTTGTTTT	c.423+2T>G
MG_V36_Rev	66	CCAGAGAGATGCAGAAACTCCCGGGAAC AAGATCCTTTATCACATACATG	c.423+2T>G
MG_V37_Fwd	65	ATAAAGGATCTTGTTCCCGTTAGTTTCTG CATCTCTCTGGTTTTGTTTTT	c.423+3G>T
MG_V37_Rev	67	ACCAGAGAGATGCAGAAACTAACGGGAA CAAGATCCTTTATCACATACAT	c.423+3G>T
MG_V38_Fwd	66	TAAAGGATCTTGTTCCCGTGTGTTTCTGC ATCTCTCTGGTTTTGTTTTTA	c.423+4A>T
MG_V38_Rev	67	AACCAGAGAGATGCAGAAACACACGGGA ACAAGATCCTTTATCACATACA	c.423+4A>T
MG_V39_Fwd	64	AAAGGATCTTGTTCCCGTGACTTTCTGCA TCTCTCTGGTTTTGTTTTTAT	c.423+5G>C
MG_V39_Rev	67	AAACCAGAGAGATGCAGAAAGTCACGGG AACAAGATCCTTTATCACATAC	c.423+5G>C
MG_V40_Fwd	68	AAGGATCTTGTTCCCGTGAGGTTCTGCAT CTCTCTGGTTTTGTTTTTATT	c.423+6T>G
MG_V40_Rev	65	AAAACCAGAGAGATGCAGAACCTCACGG GAACAAGATCCTTTATCACATA	c.423+6T>G
MG_V41_Fwd	65	TTCCTTTTGTAATCTTTGCAGATATTCATG ACAAAGGAAAAGAGTCTGGC	c.424-151T>G
MG_V41_Rev	62	TTTTCCTTTGTCATGAATATCTGCAAAGAT TACAAAAGGAAAAATGGTCAC	c.424-151T>G
MG_V42_Fwd	68	GGGACTGTTTCCTATATTATTGCAGGAGG AGGACCACAGTGTCACTGAGA	c.423+156G>T
MG_V42_Rev	64	CACTGTGGTCCTCCTCCTGCAATAATATA GGAAACAGTCCCATCTATTTA	c.423+156G>T
MG_V43_Fwd	66	TCTTCTTCTTCTTCCTCTTAGCCACAGGA TTTGAGCAACTTCTATGCACA	c.424-7A>G
MG_V43_Rev	65	AAGTTGCTCAAATCCTGTGGCTAAGAGGA AGAAGAAGAAGAAGAAGA	c.424-7A>G
MG_V44_Fwd	67	CACAGTGTCACTGAGAGATTCGGAAAATC TCCAGATCTGAGACAGTGTGG	c.423+190A>C
MG_V44_Rev	68	CTCAGATCTGGAGATTTTCCGAATCTCTC AGTGACACTGTGGTCCTCCTC	c.423+190A>C
MG_V45_Fwd	68	CCAGCAGCTCATTTGGTGAGTTTTGTTTG AGAATAATGGGGCACAGAATT	c.423+294G>T
MG_V45_Rev	68	CCCCATTATTCTCAAACAAAACTCACCAA ATGAGCTGCTGGAGGATTTTC	c.423+294G>T
MG_V46_Fwd	68	AGCAGCTCATTTGGTGAGGTATGTTTGAG AATAATGGGGCACAGAATTTT	c.423+296T>A
MG_V46_Rev	69	TGCCCCATTATTCTCAAACATACCTCACC AAATGAGCTGCTGGAGGATTT	c.423+296T>A
MG_V47_Fwd	68	GACCTGAGAAGACCAAATGGGTAAGCTA ATACATCCAGGTGTCTCCGATT	c.201-96A>G
MG_V47_Rev	68	CACCTGGATGTATTAGCTTACCCATTTGG TCTTCTCAGGTCACCTTCGGA	c.201-96A>G
MG_V48_Fwd	67	CACTGGATATTTTTCTTTGTAAGATGTGG CCCCATGGTATTGGATGCTTT	c.201-3T>A
MG_V48_Rev	64	AATACCATGGGGCCACATCTTACAAAGAA AAATATCCAGTGGTATTTATG	c.201-3T>A
MG_V49_Fwd	65	GGATATTTTTCTTTGTTAGAAGTGGCCCC ATGGTATTGGATGCTTTAATC	c.202T>A
MG_V49_Rev	65	ATCCAATACCATGGGGCCACTTCTAACAA AGAAAAATATCCAGTGGTATT	c.202T>A

MG_V50_Fwd	66	GTGATAAAGGATCTTGTTCCGGTGAGTTT CTGCATCTCTCTGGTTTTGTT	c.423C>G
MG_V50_Rev	64	AGAGAGATGCAGAAACTCACCGGAACAA GATCCTTTATCACATACATGTG	c.423C>G
MG_V51_Fwd	70	ATGTGTGTCTCTTTCAGGCAGCTGTGGCT CTTGTGCAATGAACATCAATG	c.290T>G
MG_V51_Rev	68	TCATTGCACAAGAGCCACAGCTGCCTGA AAGAGACACACATTTAACACAT	c.290T>G
MG_V52_Fwd	66	TAAATGTGTGTCTCTTTCAGTCATCTGTG GCTCTTGTGCAATGAACATCA	c.287G>T
MG_V52_Rev	69	TTGCACAAGAGCCACAGATGACTGAAAG AGACACACATTTAACACATCCT	c.287G>T
MG_V53_Fwd	67	ACACATGTATGTGATAAAGGGTCTTGTTC CCGTGAGTTTCTGCATCTCTC	c.413A>G
MG_V53_Rev	68	AGAAACTCACGGGAACAAGACCCTTTATC ACATACATGTGTGGAAGAGGG	c.413A>G
MG_V54_Fwd	65	AGATGTGGCCCCATGGTATTAGATGCTTT AATCAAGATTAAGAATGAAGT	c.219G>A
MG_V54_Rev	64	TTAATCTTGATTAAAGCATCTAATACCATG GGGCCACATCTAACAAAGAA	c.219G>A
MG_V55_Fwd	62	TTAATCTTGATTAAAGCATCTAATACCATG GGGCCACATCTAACAAAGAA	c.245A>T
MG_V55_Rev	64	AGGTCAAAGTAGAGTCAACTACATTCTTA ATCTTGATTAAAGCATCCAAT	c.245A>T
MG_V56_Fwd	65	ACTGGATATTTTTCTTTGTTCGATGTGGC CCCATGGTATTGGATGCTTTA	c.201-2A>C
MG_V56_Rev	64	CAATACCATGGGGCCACATCGAACAAAG AAAAATATCCAGTGGTATTTAT	c.201-2A>C
MG_V57_Fwd	65	TCTTCTTCCTCTTAACCACAAGATTTGAG CAACTTCTATGCACAGTACAA	c.424-1G>A
MG_V57_Rev	65	GCATAGAAGTTGCTCAAATCTTGTGGTTA AGAGGAAGAAGAAGAAGAAGA	c.424-1G>A
MG_V58_Fwd	66	TTAAATGTGTGTCTCTTTCACGCATCTGT GGCTCTTGTGCAATGAACATC	c.287-1G>C
MG_V58_Rev	69	TGCACAAGAGCCACAGATGCGTGAAAGA GACACACATTTAACACATCCTC	c.287-1G>C

1.4 Transcript analysis (PSI)

Relative transcript proportions were calculated using the Percent-spliced in concept (PSI) adapted to splice-junction-based quantification. The PSI value is a quantitative metric used to describe the relative frequency at which a particular exon or splice junction is included in transcripts. PSI represents the proportion of inclusion-supporting reads compared to all reads that cover the corresponding alternative splicing event. Conceptually, PSI is calculated by dividing the number of reads consistent with exon inclusion by the total of inclusion- and exclusion-supporting reads, and is expressed as a percentage (Schafer et al., 2015). For this study, the PSI principle was adapted to the STAR SJ output to estimate transcript proportions for each variant.



$$TR_{j_x} = \frac{UR_{j_x}}{UR_{j_r}}$$

$$RN_{j_x} = \frac{UR_{j_x}}{\sum_{n=1}^{i} UR_{jn}}$$

UR = uniquely mapping reads crossing a splice junction
TR = Transcript ratio
RN = normalized transcript ratio
j_x = transcript *(x)* defining splice junction
j_r = reference splice junction
i = total number of splice junctions defining the transcripts

Before calculation, all transcript defining splice junctions (j_x) were identified in the prior step of SJ annotation (section 3.9.2.). In case of pseudo-exon inclusion, two splice junctions could be considered as transcript defining. At least one read must span both junctions to distinguish true pseudo-exon inclusion from isolated donor or acceptor gain. If two confirming junctions were present but read coverage differently, the junction with lower read count was used to represent transcript. Following the identification of all relevant splice junctions for each variant (i), a reference splice junction (j_r) was selected. This junction corresponded to the full-length transcript and was shared across all isoforms within one variant, thereby representing the total pool of inclusion- and exclusion-supporting reads. If multiple reference junctions were suitable the junction with the highest number of uniquely mapping reads was chosen. The relative transcript ratio (TR_{jx}) was calculated by dividing the number of uniquely mapping reads crossing the defining splice junction UR_{jx} by the number of uniquely mapping reads crossing the reference splice junction UR_{jr}:

$$TR_{jx} = \frac{UR_{jx}}{UR_{jr}}$$

If the sum of all transcript-defining junction counts did not equal the reference count ($\sum UR_{jn} \neq UR_{jr}$), an internal normalization step was applied. For each transcript-defining junction, a normalized ratio (RN_{jn}) was calculated as:

$$RN_{jx} = \frac{UR_{jx}}{\sum_{n=1}^{i} UR_{jn}}$$

This adjustment ensured that all transcript ratios collectively summed up to 1.

UR uniquely mapping reads crossing a splice junction
TR transcript ratio
RN normalized transcript ratio
j_x transcript (x) defining splice junction
j_r reference splice junction
i total number of splice junctions defining the transcripts

If the proportion of transcript-defining reads relative to the reference ($\frac{\sum UR_{jn}}{UR_{jr}}$) was <0.9, this indicated a potential whole-intron retention, which could not be detected by STAR algorithm as it produced no splice junctions. In case, a whole intron retention was confirmed in reference alignment (IGV), its proportion was estimated from read coverage across the retained intron to coverage of the corresponding wild-type exon. If intron coverage was <1% of maximal exon coverage, missing reads were attributed to mapping imprecision and background noise, and ratios of transcripts were normalized to add up to 1. For whole intron retentions indicating >1% coverage, an estimated transcript ratio (ETR_{jx}) was derived from coverage profiles and converted into an estimated read count (EUR_{jx}) to integrate into TR normalization.

$$EUR_{jx} = ETR_{jx} \cdot UR_{jr}$$

EUR_{jx}estimated read count
ETR_{jx} estimated transcript ratio

These estimated read counts were then incorporated into the normalization pipeline described above.

1.5 HGVS nomenclature for transcripts with pseudo exon inclusion/InDel/double insertion/multi exon skipping

ORF were located using https://www.bioinformatics.org/sms2/orf_find.html
AA = first changed aminoacid
If upstream exon ends with incomplete base triplet (B or BB) those bases are put in front of Pseudo exon Sequence and included in ORF finder analysis

Variant (HGVSc)	RNA HGVS (effect)	Inserted/deleted sequence	Orf finder output	Protein HGVS (NP_002991.2)
c.201-91C>G **Transcript 9b**	r.200-201ins[201-211_201-92]	Exon 2 end: AA (Lys 67) AAATGTGTTTGCACTGTGATCATCAAAATAGAACTGTGCAGTACTTAAAATGTAAGAATCTGCAAAAACAAAACTACAGAACAAATTCCGAAGGTGACCTGAGAAGACCAAATGGATAAG Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 21. AAAAATGTGTTTGCACTGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. KNVFAL*	Ter after 6 AA p.Cys68AsnTer6
c.201-91C>G **Transcript 9d**	r.200-201ins[201-226_201-92]	Exon 2 end: AA CTTTCCATTTCCTAGAAATGTGTTTGCACTGTGATCATCAAAATAGAACTGTGCAGTACTTAAAATGTAAGAATCTGCAAAAACAAAACTACAGAACAAATTCCGAAGGTGACCTGAGAAGACCAAATGGATAAG Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 36. AACTTTCCATTTCCTAGAAATGTGTTTGCACTGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. NFPFPRNVFAL*	Ter after 12 AA p.Lys67AsnTer12

c.201-96A>G **Transcript 47b**	r.200-201ins[201-211_201-97]	Exon 2 end: AA (Lys 67) AAATGTGTTTGCACTGTGATCATCAAAATAGAACTGTGCAGTACTTAAAATGTAAGAATCTGCAAAAACAAAACTACAGAACAAATTCCGAAGGTGACCTGAGAAGACCAAATGG Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 21. AAAAATGTGTTTGCACTGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. KNVFAL*	Ter after 6 AA p.Cys68AsnTer6
c.201-96A>G **Transcript 47c**	r.200-201ins[201-226_201-97]	Exon 2 end: AA (Lys 67) CTTTCCATTTCCTAGAAATGTGTTTGCACTGTGATCATCAAAATAGAACTGTGCAGTACTTAAAATGTAAGAATCTGCAAAAACAAAACTACAGAACAAATTCCGAAGGTGACCTGAGAAGACCAAATGG Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 36. AACTTTCCATTTCCTAGAAATGTGTTTGCACTGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. NFPFPRNVFAL*	Ter after 12 AA p.Lys67AsnTer12
c.423+156G>T **Transcript 42b**	r.423_424ins[423+161_423+288]	Exon 4 end: CCC (Pro 141) GAGGAGGACCACAGTGTCACTGAGAGATTAGGAAAATCTCCAGATCTGAGACAGTGTGGCCAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAGAAAATCCTCCAGCAGCTCATTTG Exon 5 beginning: GAT (Asp 142)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 105. GAGGAGGACCACAGTGTCACTGAGAGATTAGGAAAATCTCCAGATCTGAGACAGTGTGGC CAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAG >Translation of ORF number 1 in reading frame 1 on the direct strand. EEDHSVTERLGKSPDLRQCGQSLIKEFEWQKINA*	Ter after 35 AA p.Asp142GluTer35
c.423+190A>C **Transcript 44b**	r.423_424ins[423+161_423+288]	Exon 4 end: CCC (Pro 141) GAGGAGGACCACAGTGTCACTGAGAGATTCGGAAAATCTCCAGATCTGAGACAGTGTGGCCAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAGAAAATCCTCCAGCAGCTCATTTG Exon 5 beginning: GAT (Asp 142)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 105. GAGGAGGACCACAGTGTCACTGAGAGATTCGGAAAATCTCCAGATCTGAGACAGTGTGGC CAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAG >Translation of ORF number 1 in reading frame 1 on the direct strand. EEDHSVTERFGKSPDLRQCGQSLIKEFEWQKINA*	Ter after 35 AA p.Asp142GluTer35
c.423+294G>T **Transcript 45b**	r.423_424ins[423+161_423+288]	Exon 4 end: CCC (Pro 141) GAGGAGGACCACAGTGTCACTGAGAGATTAGGAAAATCTCCAGATCTGAGACAGTGTGGCCAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAGAAAATCCTCCAGCAGCTCATTTG Exon 5 beginning: GAT (Asp 142)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 105. GAGGAGGACCACAGTGTCACTGAGAGATTAGGAAAATCTCCAGATCTGAGACAGTGTGGC CAGAGTTTAATTAAAGAATTTGAATGGCAGAAGATAAACGCCTAG	Ter after 35 AA p.Asp142GluTer35

			>Translation of ORF number 1 in reading frame 1 on the direct strand. EEDHSVTERLGKSPDLRQ CGQSLIKEFEWQKINA*	
c.423+296T>A **Transcript 46b**	r.423_424ins[423+161_423+293]	Exon 4 end: CCC (Pro 141) GAGGAGGACCACAGTGTCACTGAGAGAT TAGGAAAATCTCCAGATCTGAGACAGTG TGGCCAGAGTTTAATTAAAGAATTTGAAT GGCAGAAGATAAACGCCTAGAAAATCCT CCAGCAGCTCATTTGGTGAG Exon 5 beginning: GAT (Asp 142)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 105. GAGGAGGACCACAGTGT CACTGAGAGATTAGGAAA ATCTCCAGATCTGAGACA GTGTGGC CAGAGTTTAATTAAAGAAT TTGAATGGCAGAAGATAA ACGCCTAG >Translation of ORF number 1 in reading frame 1 on the direct strand. EEDHSVTERLGKSPDLRQ CGQSLIKEFEWQKINA*	Ter after 35 AA p.Asp142GluTer35
c.200+5G>C **Transcript 8g**	r.201_286delins200+1_200+236	Exon 2 end: AA (Lys 67) GTGAGTATCTCTGTGAAAGCCAGCTATT GAAGGAGAGTTCTTGATTTGATTTAGGG ACATGCTTTTCACATCCTTGGAAGGCTTA AAAATCTGAGATCATCCGATGGCTCTGTT TTTAAAACAAGAATGAGCTTGGGCCGGG CACGGTGGCTCACGCCTATAATCCCAGC ACTTTGGGAGGCTCAGGCGGGCAGATC ATCTGAGGTTGGGAGTTCAAGACCAATG TGGCCAACATG Exon 4 beginning: G (Next base due to Exon 3 skipping) Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 6. AAGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. K*	p.Cys68Ter
c.200+5G>C **Transcript 8c**	r.200_201ins[200+1_200+30; 201-2_201-1] -> 200+5 G>C included?	Exon 2 end: AA (Lys 67) GTGACTATCTCTGTGAAAGCCAGCTATT GA Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 6. AAGTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. K*	p.Cys68Ter
c.200+1G>A **Transcript 2d**	r.200_201ins[a; 200+2_200+30; 201-2_201-1]	Exon 2 end: AA (Lys 67) ATGACTATCTCTGTGAAAGCCAGCTATTG A Exon 3 beginning: A TGT (Cys 68)	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 6. AAATGA >Translation of ORF number 1 in reading frame 1 on the direct strand. K*	p.Cys68Ter
c.287-1G>T (Transcript 26e) **c.287-1G>C (Transcript 58b)** **c.287-2A>T (Transcript 3b)**	r.201_423del	Exon 2 end: AA (Lys 67) Exon 3 beginning: A TGT (Cys 68) Exon 5 beginning: GAT (Asp 142) Exon 5 sequence: (GAT)TTGAGCAACTTCTATGCACAGTACA AATCCATTGAGCCTTATTTGAAGAAGAAG GA	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 9. AAGATTTGA >Translation of ORF number 1 in reading frame 1 on the direct strand. KI*	p.Cys68IleTer2

c.287-3C>G (Transcript 25c) **c.287-5T>G (Transcript 24c)**				
c.200+5G>C (Transcript 8f)	r.73_286del	Exon 1 end: CAG (Gln 24) Exon 2 beginning: GCC (Ala 25) S26 R27 Exon 4 beginning: GC Exon 4 sequence: (GC)ATCTGTGGCTCTTGTGCAATGAACATCAATGGAGGCAACACTCTAGCTTGCACCCGAAGGATTGACACCAACCTCAATAAGGTCTCAAAAATCTACCCTCTTCCACACATGTATGTGATAAAGGATCTTGTTCCC	>ORF number 1 in reading frame 1 on the direct strand extends from base 1 to base 24. GCATCTGTGGCTCTTGTGCAATGA >Translation of ORF number 1 in reading frame 1 on the direct strand. ASVALVQ*	p.Arg27ValTer6