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GCTCTGGGTT GTGGGAGTTG GGGAGCTGCT CCGGCTTCGG CGCGGAGGGG	-45	
CGGCGGCCGG GGAGGCGGCG GCGGCGGCAG GATTCCCAGG AGCCATGTTG	6	del EXON 2 M1L Met1?
TCAGAAGTCC TACTGGTGTC TGCTCCGGGG AAAGTCATCC TTCATGGAGA	56	E4ter V5A L6fs V8L V8M V8E A10V P11S G11R K13X K13Q K13Nfs*68 G18R E19K
ACATGCCGTG GTACATGGCA AGGTAGCACT GGCTGTATCC TTGAACTTGA	106	H20N H20P H20R H20Q A21V V22M H24P G25fs G25V G25G del EXON 3 A28T L29fs L35S
GAACATTCCT CCGGCTTCAA CCCCACAGCA ATGGGAAAGT GGACCTCAGC	156	R36T L39P R40W R40L L41P L41R H44fs L51F S52N
TTACCCAACA TTGGTATCAA GCGGGCCTGG GATGTGGCCA GGCTTCAGTC	206	I56V W62X(c.185) W62X(c.186) S69T
ACTGGACACA AGCTTTCTGG AGCAAGGTGA TGTCACAACA CCCACCTCAG	256	D79N D79Y V80I F83C S85*
AGCAAGTGGA GAAGCTAAAG GAGGTTGCAG GCTTGCCTGA CGACTGTGCT	306	E93fs L97fs D100N
GTCACCGAGC GCCTGGCTGT GCTGGCCTTT CTTTACTTAT ACCTGTCCAT	356	V109L Y114fs Y116H L117P
CTGCCGGAAG CAGAGGGCCC TGCCGAGCCT GGATATCGTA GTGTGGTCGG	406	I119M R124W del EXON 5 P127L S128Pfs* D130G V132I V132Efs*25 W134X S135L S135S
AGCTGCCCCC CGGGGCGGGC TTGGGCTCCA GCGCCGCCTA CTCGGTGTGT	456	G140fs A141fs (dupG) A141fs (delG) G142D G144V S146N A147T A147A A148T
447_448insGCCTAC A148V Y149X S150L C152fs S150S V151M C152Y		
CTGGCAGCAG CCCTCCTGAC TGTGTGCGAG GAGATCCCAA ACCCGCTGAA	506	T159fs C161RfsX25 C161R I164fs N166K P165L P167L L168fs
GGACGGGGAT TGCGTCAACA GGTGGACCAA GGAGGATTTG GAGCTAATTA	556	D170D G171R D172D C173R C173Y E180K L182F
ACAAGTGGGC CTTCCAAGGG GAGAGAATGA TTCACGGGAA CCCCTCCGGA	606	W188X A189V Q190fs G192E H197H S201F G202R G202Q
GTGGACAATG CTGTCAGCAC CTGGGGAGGA GCCCTCCGAT ACCATCAAGG	656	V203fs V203A D204E N205D I209A G211A G211E G212R G211del L214Hfs*63 R215X R215G R215Q
H217P Q218X G219W		
GAAGATTTCA TCCTTAAAGA GGTCGCCAGC TCTCCAGATC CTGCTGACCA	706	L224* R226K P228L P228P L230P L234P
ACACCAAAGT CCCTCGCAAT ACCAGGGCCC TTGTGGCTGG CGTCAGAAAC	756	T237S T237N R241C T243I L246P V247fs V250I V250F N252S
AGGCTGCTCA AGTTCCCAGA GATCGTGGCC CCCCTCCTGA CCTCAATAGA	806	L255P F257I I260I V261A A262P P263P L264F c.790del L265P L265R I268I I268K D269H
TGCCATCTCC CTGGAGTGTG AGCGCGTGCT GGGAGAGATG GGGGAAGCCC	856	S272P S272F S272Fdelins R277C R277G R277H R277R V278A L279P M282T E284Kfs*17

c.853insA

CAGCCCCGGA GCAGTACCTC GTGCTGGAAG AGCTCATTGA CATGAACCAG 906 P286L P288L Q290H Y291D V293M del exons 10-11 E296G I298T D299N M300V N301T N301Tfs* Q302*

CACCATCTGA ATGCCCTCGG CGTGGGCCAC GCCTCTCTGG ACCAGCTCTG 956 L308L G309S G309R G309V V310M V310L G311R H311R S314S L315V L315Gfs*51 c.955T>C C319S

CCAGGTGACC AGGGCCCGCG GACTTCACAG CAAGCTGACT GGCGCAGGCG 1006 V321A T322S T322N A324V R325R G326R S329N S329R G333G A334T G335S G335D G335A G335G G336S

GTGGTGGCTG TGGCATCACA CTCCTCAAGC CAGGGCTGGA GCAGCCAGAA 1056 G338S G338D C339S T342A T342I L343I L343P G347R P351S

GTGGAGGCCA CGAAGCAGGC CCTGACCAGC TGTGGCTTTG ACTGCTTGGA 1106 c.1057delTGGAGGCCACGAAG V353del T356M T356R L357fs Q358P S362I F365I F364S D366fs C367S

AACCAGCATC GGTGCCCCCG GCGTCTCCAT CCACTCAGCC ACCTCCCTGG 1156 I372M G376S G376V V377I S378P I379N H380R A382P S384F D386N

ACAGCCGAGT CCAGCAAGCC CTGGATGGCC TCTGAGAGGA GCCCACGACA *15 R388X Q390* Q390P Stop397R c.1202C>T

CTGCAGCCCC ACCCAGATGC CCCTTTCTGG ATTATTCTGG GGGCTGCAGT *65 c.*35C>T 1245-1246INSG

TCGACTCTGT GCTGGCCAGC GAGCGCCCAG CTCCTGACAC TGCTGGAGAG *115

GCCCCAGCCG CTTGGCGATG CCAGCCAAGC TCTGCAGTCC CAGCGGTGGG *165

ACCTAGGGAG GCATGGTCTG CCCTCTGCAT CCTCTGGAGC CAGCCGAGCA *215

GGAGGCCTAG GAGGGTCCTC TGAGACTCCA GACCTGAGGC GAGAAGGGCT *265

GCTTCCCTGA AGCTCCCACA GTCCCATCTG CTTCAGGCCC CCGCCTTGGC *315

CTGTGTTCTT CCTGGCCGCC TGGGTCCAAT GCTCAGGTGC TGGGGCCTGG *365

TTCCCGGAGA AGTGTGCCTT CTCTCTCCCT TTTCAGGGAC CGCCCCCTGT *415

CTCTCAGGGC CAGGCCTCTC CCTCCTCCAG GAAGCCTTCC CCTACCCCTT *465

GTCGCCCCTC CCTCCCAGAG CACCTGCTGT CTGGGTGGCT CACTCAGCAC *515

TTGGTGTGGC CTTCCCTTCT ACCTAGCGGG ATGGGGCTCC CCCAGGGGCT *565

GTCCCGGAGG CGGTGGGCCT GGTTAAATAA GGCAGGGTTT ATATGCACTT *615 *571G>A

TCTTCCGATC TGTACCTGAG AGGTTTGTGG AAAAGATGGC AAATGGGGAA *665

TAAAAAGATT TTGTGTCAAC AGTAGAGACT CCAGGCCACC AGCACCTCCC *715

TCTGTCCCTG TCCCCTCTCC AGCTGTTTCC TCCATGGAGC TCTTCAGCAA *765

TGGAGGGAAA TAGGGTTTGG GGTCACTTTG TTGTGCGTCT TGGGGATGAG *815

GTGGCTTTTC CCAGATGGCC CTTGCTGGAG AGGGACTGGG ACACGGCTCT *865

CAGTCCATCA GCACAACTCT AGGCTGCTGC TGCGGAGGGA GAAGTTGAGC *915

TTCCTAGCTC CAGAATCACA AGCACCCACG AGAGCACAGA CCTGTGTAAG *965

ACAGGAAAGC AGAACCTGCC ATCGCTCCTG GGGCGCGCCT TCCTTTCTGA *1015

AATGAACTGG CTGGATGGAG AAAACAGACT CAAATGTTCT GGCCCGGGTG *1065

CCTGGCACTC CCCACCCCCG CCCCCCACCG GCCCTATTTG AACTTTATAT *1115

TGCAGTCAGC TTGGTGCTTT CCGAAATGCC ATTAGCCATC AGGAAACCCT *1165

TGTAGTTGGT GCCTTGCCAG CCAGAACCTC TGGGACCCAC GGACCTGCAA *1215

AGAGGCCGAG TGGAAAGGTG GGGGCCGGCG CAGGGATTTC AGGATGAGGT *1265

GAAAGCGATT CAGTGCGCGT CTGCCCTTGG CCACTAGGGG GCAGCTGGCG *1315

GCCTTCCCTG CTGTTGTCTT CCTGCAGGGT GAGAGGAGCA GGAGCCGAGC *1365

TCCACCCCCA CGCCAGCCTT GGGCCCGGCC TGGGATCACT GCTGGGAACG *1415

TGAGAGTGAA GGGAGGACGC CTACCCCAGC TTAACTTGTA GAAATGGCCC *1465

CAGATCACTG ATGGCTGTTC CCTGCCCCTT CCCTTCAAAA CACAACGCAT *1515

AAAGCAGTAA TACTAATTAA TACTGAACGC TCA

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