



MVK (NM_000431.4) - cDNA + Protein - 2024-05-14

GCTCTGGGTT GTGGGAGTTG GGGAGCTGCT CCGGCTTCGG CGCGGAGGGG -45

CGGCGGCCGG GGAGGCGGCG GCGGCGGCAG GATTCCCAGG AGCCATGTTG 6 del EXON 2 M1L Met1?

MetLeu 2

TCAGAAGTCC TACTGGTGTG TGCTCCGGGG AAAGTCATCC TTCATGGAGA 56 E4ter V5A L6fs V8L V8M V8E A10V P11S G11R K13X K13Q K13Nfs*68 G18R E19K

SerGluValL euLeuValSe rAlaProGly LysValIleL euHisGlyGl 19

ACATGCCGTG GTACATGGCA AGGTAGCACT GGCTGTATCC TTGAACTTGA 106 H20N H20P H20R H20Q A21V V22M H24P G25fs G25V G25G del EXON 3 A28T L29fs L35S

uHisAlaVal ValHisGlyL ysValAlaLe uAlaValSer LeuAsnLeuA 36

GAACATTCCI CCGGCTTCAA CCCACAGCA ATGGGAAAGT GGACCTCAGC 156 R36T L39P R40W R40L L41P L41R H44fs L51E S52N

rgThrPheLe uArgLeuGln ProHisSerA snGlyLysVa lAspLeuSer 52

TTACCCAACA TTGGTATCAA GCGGGCCTGG GATGTGGCCA GGCTTCAGTC 206 I56V W62X(c.185) W62X(c.186) S69T

LeuProAsnI leGlyIleLy sArgAlaTrp AspValAlaA rgLeuGlnSe 69

ACTGGACACA AGCTTTCTGG AGCAAGGTGA TGTCACAACA CCCACCTCAG 256 D79N D79Y V80I F83C S85*

rLeuAspThr SerPheLeuG luGlnGlyAs pValThrThr ProThrSerG 86

AGCAAGTGGA GAAGCTAAAG GAGGTTGCAG GCITGCCTGA CGACTGTGCT 306 E93fs L97fs D100N

luGlnValGl uLysLeuLys GluValAlaG lyLeuProAs pAspCysAla 102

GTCACCGAGC GCCTGGCTGT GCTGGCCTTT CTTIACCTAI ACCIGTCCAT 356 V109L Y114fs Y116H L117P

ValThrGluA rgLeuAlaVa lLeuAlaPhe LeuTyrLeuT yrLeuSerIl 119

CTGCCGGAAG CAGAGGGCCC TGCCGAGCCT GGATATCGTA GTGTGGTGG 406 I119M R124W del EXON 5 P127L S128Pfs* D130G V132I V132Efs*25 W134X S135L S135S

eCysArgLys GlnArgAlaL euProSerLe uAspIleVal ValTrpSerG 136

AGCTGCCCCC CGGGGCGGGC TTGGGCTCCA GCGCCGCCCTA CTCGGTGTGT 456 G140fs A141fs (dupG) A141fs (delG) G142D G144V S146N A147T A147A A148T

447 448insGCCTAC A148V Y149X S150L C152fs S150S V151M C152Y

luLeuProPr oGlyAlaGly LeuGlySerS erAlaAlaTy rSerValCys 152

CTGGCAGCAG CCCTCCTGAC TGTGTGCGAG GAGATCCCAA ACCCGCTGAA 506 T159fs C161RfsX25 C161R I164fs N166K P165L P167L L168fs

LeuAlaAlaA laLeuLeuTh rValCysGlu GluIleProA snProLeuLy 169

GGACGGGGAT TCGTCAACA GGTGGACCAA GGAGGATTTG GAGCTAATTA 556 D170D G171R D172D C173R C173Y E180K L182E

sAspGlyAsp CysValAsnA rgTrpThrLy sGluAspLeu GluLeuIleA 186

ACAAGTGGGC CTTCCAAAGGG GAGAGAATGA TTCACGGGAA CCCCTCCGGA 606 W188X A189V Q190fs G192E H197H S201F G202R G202Q

snLysTrpAl aPheGlnGly GluArgMetI leHisGlyAs nProSerGly 202

GTGGACAATG CTGTCAGCAC CTGGGGAGGA GCCCTCCGAT ACCATCAAGG 656 V203fs V203A D204E N205D T209A G211A G211E G212R G211del L214Hfs*63 R215X R215G R215Q

H217P Q218X G219W

ValAspAsnA laValSerTh rTrpGlyGly AlaLeuArgT yrHisGlnGl 219

GAAGATTTCA TCCTIAAGA GGTGCGCAGC TCICAGATC CTGCTGACCA 706 L224* R226K P228L P228P L230P L234P

yLysIleSer SerLeuLysA rgSerProAl aLeuGlnIle LeuLeuThra 236

ACACCAAAGT CCCTCGCAAT ACCAGGGCCC ITGTGGCTGG CGTCAGAAAC 756 T237S T237N R241C T243I L246P V247fs V250I V250F N252S

snThrLysVa lProArgAsn ThrArgAlaL euValAlaGl yValArgAsn 252

AGGCTGCICA AGITCCCAGA GATCGTGGCC CCCCTCCIGA CCTCAATAGA 806 L255P F257I I260I V261A A262P P263P L264E c.790del L265P L265R I268T I268K D269H
ArgLeuLeuL ysPheProGl uIleValAla ProLeuLeuT hrSerIleAs 269

TGCCATCTCC CTGGAGTGTG AGCGCGTIGCT GGGAGAGATG GGGGAAGCCC 856 S272P S272F S272Fdelins R277C R277G R277H R277R V278A L279P M282T E284Kfs*17
c.853insA

pAlaIleSer LeuGluCysG luArgValLe uGlyGluMet GlyGluAlaP 286

CAGCCC CGGA GCAGTACCTC GTGCTGGAAG AGCTCATTGA CATGAAC CAG 906 P286L P288L Q290H Y291D V293M del exons 10-11 E296G I298T D299N M300V N301T N301Tfs*
Q302*

roAlaProGl uGlnTyrLeu ValLeuGluG luLeuIleAs pMetAsnGln 302

CACCATCTGA ATGCCCTCGG CGTGGGCCAC GCCTCTCTGG ACCAGCTCTG 956 L308L G309S G309R G309V V310M V310L G311R H311R S314S L315V L315Gfs*51 c.955T>C C319S
HisHisLeuA snAlaLeuGl yValGlyHis AlaSerLeuA spGlnLeuCy 319

CCAGGTGACC AGGGCCCGCG GACTTCACAG CAAGCTGACT GGCGCAGGGCG 1006 V321A T322S T322N A324V R325R G326R S329N S329R G333G A334T G335S G335D G335A G335G
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sGlnValThr ArgAlaArgG lyLeuHisSe rLysLeuThr GlyAlaGlyG 336

GTGGTGGCTG TGGCATCACAC CTCCTCAAGC CAGGGCTGGA GCAGCAGAA 1056 G338S G338D C339S T342A T342I L343I L343P G347R P351S
lyGlyGlyCy sGlyIleThr LeuLeuLysP roGlyLeuGl uGlnProGlu 352

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

ValGluAlaT hrLysGlnAl aLeuThrSer CysGlyPheA spCysLeuGl 369

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

uThrSerIle GlyAlaProG lyValSerIl eHisSerAla ThrSerLeuA 386

ACAGCCGAGT CCAGCAAGCC CTGGATGGCC TCIGAGAGGA GCCCACGACA *15 [R388X](#) [Q390*](#) [Q390P](#) [Stop397R](#) [c.1202C>T](#)
spSerArgVa lGlnGlnAla LeuAspGlyL euStop

CTGCAGCCCC ACCCAGATGC CCCTTTCTGG ATTATTCTGG GGGCTGCAGT *65 [c.*35C>T](#) [1245-1246INS](#)
TCGACTCTGT GCTGGCCAGC GAGCGCCAG 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 GGTAAATAA GGCAGGGTTT ATATGCACTT *615 [*571G>A](#)
TCTTCCGATC TGTACCTGAG AGGTTTGTGG AAAAGATGGC AAATGGGGAA *665
TAAAAAGATT TTGTGTCAAC AGTAGAGACT CCAGGCCACC AGCACCTCCC *715
TCTGTCCCTG TCCCCTCTCC AGCTGTTTCC TCCATGGAGC TCTTCAGCAA *765
TGGAGGGAAG TAGGGTTTGG GGTCACTTTG TTGTGCGTCT TGGGGATGAG *815
GTGGCTTTTC CCAGATGGCC CTTGCTGGAG AGGGAAGTGG ACACGGCTCT *865
CAGTCCATCA GCACAACTCT AGGCTGCTGC TGCAGGAGGA GAAGTTGAGC *915
TTCCTAGCTC CAGAATCACA AGCACCACG AGAGCACAGA CCTGTGTAAG *965
ACAGGAAAGC AGAACCTGCC ATCGCTCCTG GGGCGCGCCT TCCTTTCTGA *1015
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TGTAGTTGGT GCCTTGCCAG CCAGAACCTC TGGGACCCAC GGACCTGCAA *1215
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GCCTTCCCTG CTGTTGTCTT CCTGCAGGGT GAGAGGAGCA GGAGCCGAGC *1365
TCCACCCCCA CGCCAGCCTT GGGCCCGGCC TGGGATCACT GCTGGGAACG *1415
TGAGAGTGAA GGGAGGACGC CTACCCAGC TTAATTGTA GAAATGGCCC *1465
CAGATCACTG ATGGCTGTTT CCTGCCCCTT CCCTTCAAAA CACAACGCAT *1515
AAAGCAGTAA TACTAATTAA TACTGAACGC TCA

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