MVK (NM_000431.4) - cDNA + Protein - 2026-11-24

GCTCTGGGTT GTGGGAGTTG GGGAGCTGCT CCGGCTTCGG CGCGGAGGGG -45

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

MetLeu 2

TCAGAAGTCC TACTGGTGTC 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

GAACATTCCT CCGGCTTCAA CCCCACAGCA ATGGGAAAGT GGACCTCAGC 156 R36T L39P R40W R40L L41P L41R H44fs L51F 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 GCTTGCCTGA CGACTGTGCT 306 E93fs L97fs D100N

luGlnValGl uLysLeuLys GluValAlaG lyLeuProAs pAspCysAla 102

GTCACCGAGC GCCTGGCTGT GCTGGCCTTT CTTTACTTAT ACCTGTCCAT 356 V109L Y114fs Y116H L117P

ValThrGluA rgLeuAlaVa lLeuAlaPhe LeuTyrLeuT yrLeuSerIl 119

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

eCysArgLys GlnArgAlaL euProSerLe uAspIleVal ValTrpSerG 136

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

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 TGCGTCAACA GGTGGACCAA GGAGGATTTG GAGCTAATTA 556 D170D G171R D172D C173R C173Y E180K L182F

sAspGlyAsp CysValAsnA rgTrpThrLy sGluAspLeu GluLeuIleA 186

ACAAGTGGGC CTTCCAAGGG 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 TCCTTAAAGA GGTCGCCAGC TCTCCAGATC CTGCTGACCA 706 L224* R226K P228L P228P L230P L234P

yLysIleSer SerLeuLysA rgSerProAl aLeuGlnIle LeuLeuThrA 236

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

snThrLysVa lProArgAsn ThrArgAlaL euValAlaGl yValArgAsn 252

AGGCTGCTCA AGTTCCCAGA GATCGTGGCC CCCCTCCTGA CCTCAATAGA 806 L255P F257I I260I V261A A262P P263P L264F c.790del L265P L265R I268T I268K D269H

ArgLeuLeuL ysPheProGl uIleValAla ProLeuLeuT hrSerIleAs 269

TGCCATCTCC CTGGAGTGTG AGCGCGTGCT GGGAGAGATG GGGGAAGCCC 856 S272P S272F S272Fdelins E276G R277C R277G R277H R277R V278A L279P M282T E284Kfs*17 c.853insA

pAlaIleSer LeuGluCysG luArgValLe uGlyGluMet GlyGluAlaP 286

CAGCCCCGGA GCAGTACCTC GTGCTGGAAG AGCTCATTGA CATGAACCAG 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 GGCGCAGGCG 1006 V321A T322S T322N A324V R325H R325R G326R S329N S329R G333G A334T G335S G335D G335A G335G G336S

sGlnValThr ArgAlaArgG lyLeuHisSe rLysLeuThr GlyAlaGlyG 336

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

lyGlyGlyCy sGlyIleThr LeuLeuLysP roGlyLeuGl uGlnProGlu 352

GTGGAGGCCA 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 TCTGAGAGGA GCCCACGACA *15 R388X Q390* Q390P Stop397R c.1202C>T

spSerArgVa lGlnGlnAla LeuAspGlyL euStop

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

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GCCCCAGCCG CTTGGCGATG CCAGCCAAGC TCTGCAGTCC CAGCGGTGGG *165

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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

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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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