



## MEFV (NM\_000243.3) - cDNA + Protein - 2024-05-17

CTACCAAAG CCAGACAGCT GGCTCGAGCC TCTCTGGTC AGCACCCATGG 4      [chr16:g.3320350\\_3256171dup -12C>G](#)

MetA 2

CTAAGACCCC TAGTGACCAT CTGCTGTCCA CCCCTGGAGGA GCTGGTGCCC 54      [L9L T12I](#)

IaLysThrPr oSerAspHis LeuLeuSerT hrLeuGluGl uLeuValPro 18

TATGACTTCG AGAAGTCAAGCTG CAGAACACCA GTGTGCAGAA 104      [Y19C K25R L28V Q34P K35R](#)

TyrAspPheG luLysPheLy sPheLysLeu GlnAsnThrS erValGlnLy 35

GGAGCACTCC AGGATCCCC GGAGCCAGAT CCAGAGAGCC AGGCCGGTGA 154      [R39G R42W P50L](#)

sGluHisSer ArgIleProA rgSerGlnIl eGlnArgAla ArgProValL 52

AGATGGCCAC TCTGCTGGTC ACCTACTATG GGGAAAGAGTA CGCCGTGCAG 204      [L57L G62W Y65Y V67E](#)

ysMetAlaTh rLeuLeuVal ThrTyrTyrG lyGluGluTy rAlaValGln 68

CTCACCCCTGC AGGTCCTGCG GCCATCAAC CAGGGCCTGC TGGCCGAGGA 254      [R75Q E84K](#)

LeuThrLeuG lnValLeuAr gAlaIleAsn GlnArgLeuL euAlaGluGl 85

GCTCCACAGG GCAGCCATTC AGGAATATTC CACCAAGAA AACGGCACAG 304      [H87R A89T Q97K Q97X Q97R N99N](#)

uLeuHisArg AlaAlaIleG lnGluTyrSe rThrGlnGlu AsnGlyThrA 102

ATGATTCCGC AGCGTCCAGC TCCCTGGGGG AGAACAAGC CAGGAGCTG 354      [D102D D103D S104C A105E S108R S108G L110P L110L G111R G111E G111G 334\\_335insG P115T](#)

P115R R116S

spAspSerAl aAlaSerSer SerLeuGlyG luAsnLysPr oArgSerLeu 118

AAGACTCCAG ACCACCC CGA GGGGAACGAG GGGAAC GCC CTCGGCCTA 404 T120I H123Q P124P E125E E128 N130del 390\_391insGAGGGAAC N130N Y135H

LysThrProA spHisProGl uGlyAsnGlu GlyAsnGlyP roArgProTy 135

CGGGGGCGGA GCTGCCAGCC TGCGGTGCAG CCAGCCCGAG GCCGGGAGGG 454 G136R G136W G136E G136G G138G S141I R143P S145G E148RfsX5 E148Q E148V E148D G150G R151S G152R

rGlyGlyGly AlaAlaSerL euArgCysSe rGlnProGlu AlaGlyArgG 152

GGCTGTCGAG GAAGCCCCTG AGCAAACGCA GAGAGAAGGC CTCGGAGGGC 504 S154P R155T S159N E163A E163D A165T A165A S166L S166S E167D  
lyLeuSerAr gLysProLeu SerLysArgA rgGluLysAl aSerGluGly 168

CTGGACGCGC AGGGCAAGCC TCGGACCCGG AGCC CGGCC TGCCGGCCGG 554 L169V Q172P P175H T177I S179I S179N P180R P180P P183T P183P  
LeuAspAlaG lnGlyLysPr oArgThrArg SerProAlaL euProGlyGl 185

GAGAAGCCCC GGCCCCTGCA GGGCGCTAGA GGGGGGCCAG GCCGAGGTCC 604 S187N P188P A193V L194P E195E G196W G196R G197R  
yArgSerPro GlyProCysA rgAlaLeuGl uGlyGlyGln AlaGluValA 202

GGCTGCGCAG AAACGCCAGC TCCCGCGGGGA GGCTGCAGGG GCTGGCGGG 654 R202Q I203P R204H N206S N206K 606\_621dup S208C S208T S209S G211G G218A  
rgLeuArgAr gAsnAlaSer SerAlaGlyA rgLeuGlnGl yLeuAlaGly 218

GGCGCCCCGG GGCAGAAGGA GTGCAGGCC TTCGAAGTGT ACCTGCCCTC 704 G219G P221P G222R K224del E225G E225D E230K E230Q Y232H c.698\_700dupTGC P234P S235L  
GlyAlaProG lyGlnLysGl uCysArgPro PheGluValT yrLeuProSe 235

GGGAAAGATG CGACCTAGAA GCCTTGAGGT CACCAATTCT ACAGGGGAGA 754 G236V M238I R239R R241K S242G S242S S242R C>G S242R C>A E244K I247V T249A G250A E251K  
rGlyLysMet ArgProArgS erLeuGluVa lThrIleSer ThrGlyGluL 252

AGGCGCCCGC AAATCCAGAA ATTCTCCTGA CTCTAGAGGA AAAGACAGCT 804 761\_764dup P257L I259V K266E T267I A268V  
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GCGAATCTGG ACTCGGCAAC AGAACCCCGG GCAAGGCCA CTCGGATGG 854 N270D S273L P277T R278P P283R P283L  
AlaAsnLeuA spSerAlaTh rGluProArg AlaArgProT hrProAspG1 285

AGGGGCATCT CGGGACCTGA AGGAAGGCC CGAACATTGG 904 S288Y A289V A289E E299G  
yGlyAlaSer AlaAspLeuL ysGluGlyPr oGlyAsnPro GluHisSerV 302

TCACCGGAAG GCCACCAGAC ACGGCTGCGA GTCCCGCTG CCACGCCAG 954 G304R P307P T309M P313H R314C R314H R314R A317T  
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GAAGGAGACC CAGTTGACGG TACCTGTGTG CGTGATTCCCT GCAGCTTCCC 1004 E319K V328A R329H  
GluGlyAspP roValAspG1 yThrCysVal ArgAspSerC ysSerPhePr 335

CGAGGCAGTT TCTGGCACC CCCAGGCCTC AGGCAGCCGC TCACTGGCT 1054 E336K V338L S339F R348H P350S P350R  
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GCCCCGGTG CCAGGACTCC CATGAAAAGGA AGAGCCCGGG AAGCCTAACG 1104 C352C P353A R354W R361T S363N S363S P364L P364P L367V  
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CCCCAGCCCC TGCCACAGTG TAAGCGCCAC CTGAAGCAGG TCCAGCTGCT 1154 P369S L372P P373L K376R Q383K L384P  
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CTTCTGTGAG GATCACGATG AGCCCATCTG CCTCATCTGC AGTCTGAGTC 1204 D389V D391N P393P L396F  
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AGGAGCAACCA AGGCCACCGG GTGCGCCCA TTGAGGAGGT CGCCCTGAA 1254 E403K H404R R408Q c.1229G>A V415V E418\*

lnGluHisG1 nGlyHisArg ValArgProI leGluGluVa lAlaLeuGlu 418

CACAAGAAGA AAATTCAGAA GAGCTGGAG CATCTGAAGA AGCTGAGAAA 1304 I423V I423T Q426R

HisLysLysL ysIleGlnLy sGlnLeuGlu HisLeuLysL ysLeuArgLy 435

ATCAGGGGAG GAGCAGCGAT CCTATGGGA GGAGAGGCA GTGAGCTTC 1354 Q440E S442T K447M

sSerGlyGlu GluGlnArgS erTyrGlyG1 uGluLysAla ValSerPheL 452

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uAspValGly GlnMetValG lyGlnIleAr gLysAlaTyr AspThrArgV 502

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uHisArgAla LysThrValP roValProG1 uLysTrpThr ThrProGlnG 552

AGATAAAACA AAAGATCCAA CTCCTCCACC AGAACGTCAGA GTTTGTGGAG 1704 E552D L559F

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AAGAGCACAA AGTACTTCTC AGAAACCCTG CGTCAGAAA TGGAAATGTT 1754 T577SAT T577A T577N T577SCG M582I E583A  
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CAATGTTCCG GAGCTGATTG GCGCTCAGGC ACATGCTGTT AATGTGATTC 1804 P588P I591T I591M G592G A595V c.1792G>A N599D  
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TGATGCAGA AACCGCTTAC CCCAACCTCA TCTTCTCTGA TGATCTGAAG 1854 L602L T606T P609P N610D  
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SerValArgL euGlyAsnLy sTrpGluArg LeuProAspG lyProGlnAr 635

ATTTGACAGC TGTATCATTG TTCTGGGCTC TCCGAGTTTC CTCTCTGGCC 1954 F636Y D637G I640M I641F V642A P646L P646P L649F L649P S650Y G651S R652C  
gPheAspSer CysIleIleV alLeuGlySe rProSerPhe LeuSerGlyA 652

GCCGTTACTG GGAGGTGGAG GTTGGAACA AGACAGCATG GATCCTGGA 2004 R652H R652R R653S R653H E656A V659F D661N D661Y K662Dfs\*36 I666V G668R  
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GCCTGCAAGA CATCCATAAG CAGGAAGGG AACATGACTC TGTCGCCAGA 2054 K671M S675N G678R G678E N679H M680L M680V M680IGC M680IGA M680IGT T681I S683S E685K  
AlaCysLysT hrSerIleSe rArgLysGly AsnMetThrL euSerProGl 685

GAATGGCTAC TGGGTGGTGA TAATGATGAA GGAAAATGAG TACCAGGGGT 2104 G687D Y688C Y688F Y688X V690L V690G V691G V691A I692DEL M693K M693I M694V M694L  
M694DEL M694K M694I K695R K695M K695N E698D A701A  
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GGCATCTTCG TGGACTACAG AGTGGAAGC ATCTCTTTT ACAATGTGAC 2204 I720M F721L F721F V722M D723D V726A I729V I729M S730C N733D N733S  
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AGCCAGATCC CACATCTATA CATTCGCCAG CTGCTCTTTTC TCTGGGCC~~CC~~ 2254 R737K F743Y E743L F743F A744S A744T A744D S747C S749C  
rAlaArgSer HisIleTyrT hrPheAlaSe rCysSerPhe SerGlyProL 752

TTCAACCTAT CTTCAGCCCT GGGACACGTG ATGGAGGGAA GAACACAGCT 2304 Q753Q Q753H P754R I755V P758S R761C R761H G764G N766H  
euGlnProII ePheSerPro GlyThrArgA spGlyGlyLy sAsnThrAla 768

CCTCTGACTA TCTGTCCAGT GGGTGGTCAG GGGCCTGACT GAATGCCAA \*8 P769A I772V Q778Sfs\*4 Q778L G779G P780T  
ProLeuThrI leCysProVa lGlyGlyGln GlyProAspS top

CACTTGCATCT CTCTTCCTGC TTCTGGCCTT GTATCTGCA TTCACACTCA \*58 \*9C>T \*12T>C \*21C>G  
ATAGTCACGG AATGCCGACT AGGTGCTAGC TGCTATGGGA AATGCAAAA \*108  
TAACAAAATA GTTACTGTGC CCACGGGAGCC TACCCGATTA TAGCAGAGGT \*158 c.\*133G>A  
AAGTTAGGAA CGAACATGTT AGTCAATCCG GGTGAAGACA TGTACTGATG \*208  
ACACACCATG GATTTCAGAG GAGGAAGTAC GGAGTCTTG CATAATCCGC \*258 c.\*245G>A  
CCCTGGGG TGGCACTCTC AGGTGCTCCT GAACAGAAGA TTTGGCCCTC \*308 c.\*267G>A  
ATTTTCCCTC AGAACCCAC GGCAAGGATA TATGCCCCT TGTTCTCTCT \*358  
GCTTCTGTCT TGAGGATATG GGAAGCCTAG AGAAACGCAA GCAGACTGGA \*408  
TTGGGATAGA AGTATTGGT TACCTGGATT AATGAACTAT GATTTTTTT \*458  
TTTTTTTTT GAGACCAAAT CTTGCTCTGT GGCCCCAGGCT GGAGTGCAGT \*508  
GGCACGATCT CAGCTCACTG CAACCTCCAC CTCCCAGGTT CAAGCGATTC \*558  
TCCTGCCTCA GCCTCCTGAG CAGCTGGAT TACAGGTGCG TGCCACCACA \*608  
CCAGGCTGGT TTTCTGTAT TTTTAGTAGA GACGGGGGTT TCACCATGTT \*658  
AGCCAGGCTG GTCTCGAACT CCTGACCTCA GGTGATCCAC CCGCCTCAGC \*708  
CTCCCAAAGT GCTGGGATTA CAGGCATGAG CCACTGTGCC CGGCCTATGA \*758

TTCTTTTTT TTTTTTTTGAGACAAAG TTTGCTCTT GTCACCCAGG \*808  
CTGGAGTGCA GTGGTGCAAT CTTGGCTCGC AACCTCCGCC TCCCAGGTTTC \*858  
AAGAGATTCT CCTGCCTCAG CCTCCGAAGT AGCTGGGATT ACAGGCGCCC \*908  
GCCACCATGC CCGGCTAATT TTTGCATT TTAGTAGACA TGAGGTTTCA \*958  
TCATGTTGGC CAGGCCGGTC TCAAACTCCT GACCTCAGGT GATGCACCCA \*1008  
CCTCAGCCTC CCAAAGTGCA GGGATTACAG GCATGAGCCA CCATGCCGGG \*1058  
CCATGATTCT TAAGAGAATT GACTGGGCCT CATGAATAAA AAAATTAGAA \*1108  
**AATCTG**

**MEFV (NM\_000243.3) - cDNA + Protein - 2024-05-17**

