



MEFV (NM_000243.3) - cDNA + Protein - 2026-06-27

CTACCAGAAG CCAGACAGCT GGCTCGAGCC TCTCCTGCTC AGCACCATGG 4 chr16:g.3292027_3306633dup chr16:g.3242027_3256633del -12C>G
MetA 2

CTAAGACCCC TAGTGACCAT CTGCTGTCCA CCCTGGAGGA GCTGGTGCCC 54 L9L T12I
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TATGACTTCG AGAAGTTCA GTTCAAGCTG CAGAACACCA GTGTGCA GAA 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

AGATGCCAC TCTGCTGTC ACCTACTATG GGGAAGATA CGCCGTGCAG 204 A56S L57L Y61C G62W p.(Glu64_Tyr65delinsAspPhe) Y65Y V67E
ysMetAlaTh rLeuLeuVal ThrTyrTyrG lyGluGluTy rAlaValGln 68

CTCACCTGC AGGTCCTGC GGCCATCAAC CAGCGCCTGC TGGCCGAGGA 254 R75Q R84K
LeuThrLeuG lnValLeuAr gAlaIleAsn GlnArgLeuL euAlaGluGl 85

GCTCCACAGG GCAGCCATTC AGGAATATTC CACA CAAGAA AA CGGCACAG 304 H87R A89T Q97K Q97X Q97R N99N
uLeuHisArg AlaAlaIleG lnGluTyrSe rThrGlnGlu AsnGlyThrA 102

ATGATTCCGC AGCGTCCAGC TCCCTGGGGG AGAACAAGCC CAGGAGCCTG 354 D102D D103D S104C A105E S108R S108G L110P L110L G111R G111E G111G 334_335insG P115T

P115R R116S

spAspSerAl aAlaSerSer SerLeuGlyG luAsnLysPr oArgSerLeu 118

AAGACTCCAG ACCACCCCGA GGGGAACGAG GGGAA~~CGGCC~~ CTCGGCCGTA 404 T120I H123O P124P E125E E128_N130del 390_391insGAGGGGAAC 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

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yArgSerPro GlyProCysA rgAlaLeuGl uGlyGlyGln AlaGluValA 202

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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

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GGGAAAGATG CGACCTAGAA GCCTTGAGGT CACCA~~TTTCT~~ ACAGGGGAGA 754 G236V M238I p.Arg239Leu R239R R241K S242G S242S S242R_C>G S242R_C>A E244K I247V T249A

G250A E251K

rGlyLysMet ArgProArgS erLeuGluVa lThrIleSer ThrGlyGluL 252

AGGCGCCCGC AAATCCAGAA ATTCTCTCTGA CTCTAGAGGA AAAGACAGCT 804 [761_764dup](#) [P257L](#) [I259V](#) [K266E](#) [T267I](#) [A268V](#)
ysAlaProAl aAsnProGlu IleLeuLeuT hrLeuGluGl uLysThrAla 268

GCGAATCTGG ACTCGGCAAC AGAACCCCGG GCAAGGCCCA CTCGGGATGG 854 [N270D](#) [S273L](#) [P277T](#) [R278P](#) [P283R](#) [P283L](#)
AlaAsnLeuA spSerAlaTh rGluProArg AlaArgProT hrProAspGl 285

AGGGGCATCT GCGGACCTGA AGGAAGGCC TGGAAATCCA GAACATTCCG 904 [S288Y](#) [A289V](#) [A289E](#) [E299G](#)
yGlyAlaSer AlaAspLeuL ysGluGlyPr oGlyAsnPro GluHisSerV 302

TCACCGGAAG GCCACCAAGAC ACGGCTGCGA GTCCCCGCTG CCACGCCAG 954 [G304R](#) [P307P](#) [T309M](#) [P313H](#) [R314C](#) [R314H](#) [R314R](#) [A317T](#)
alThrGlyAr gProProAsp ThrAlaAlaS erProArgCy sHisAlaGln 318

GAAGGAGACC CAGTTGACGG TACCTGTGTG CGTGATTCCT GCAGCTTCCC 1004 [E319K](#) [V328A](#) [R329H](#)
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GCCCCCGGTG CCAGGACTCC CATGAAAGGA AGAGCCCGGG AAGCCTAAGC 1104 [C352C](#) [P353A](#) [R354W](#) [Q356R](#) [R361T](#) [S363N](#) [S363S](#) [P364L](#) [P364P](#) [L367V](#)
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ProGlnProL euProGlnCy sLysArgHis LeuLysGlnV alGlnLeuLe 385

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uPheCysGlu AspHisAspG luProIleCy sLeuIleCys SerLeuSerG 402

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CAATGTTCCG GAGCTGATTG GCGCTCAGGC ACATGCTGTT AATGTGATTG 1804 P588P I591T I591M G592G A595V c.1792G>A N599D

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CCTCTGACTA TCTGTCCAGT GGGTGGTCAG GGGCCTGACT GAATGCCCAA *8 P769A I772V Q778Sfs*4 Q778L G779G P780T
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ACACACCATG GATTTCAGAG GAGGAAGTAC GGAGTCGTTG CATAATCCGC *258 c.*245G>A
CCCTGGTGGG TGGCACTCTC AGGTGCTCCT GAACAGAAGA TTTGGCCCTC *308 c.*267G>A
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GCTTCTGTCT TGAGGATATG GGAAGCCTAG AGAAACGCAA GCAGACTGGA *408
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