



MEFV (NM_000243.3) - cDNA + Protein - 2026-05-01

CTACCAGAAG CCAGACAGCT GGCTCGAGCC TCTCCTGCTC AGCACCATGG 4 [chr16:g.3320350_3256171dup](#) [-12C>G](#)
Meta 2

CTAAGACCCC TAGTGACCAT CTGCTGTCCA CCCTGGAGGA GCTGGTGCCC 54 [L9L](#) [T12I](#)
laLysThrPr oSerAspHis LeuLeuSerT hrLeuGluGl uLeuValPro 18

TATGACTTCG AGAAGTTCAA GTTCAAGCTG 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 GGGGAAGGTA CGCCGTGCAG 204 [A56S](#) [L57L](#) [Y61C](#) [G62W](#) [p.\(Glu64_Tyr65delinsAspPhe\)](#) [Y65Y](#) [V67E](#)
ysMetAlaTh rLeuLeuVal ThrTyrTyrG lyGluGluTy rAlaValGln 68

CTCACCTGC AGGTCCTGCG GGCCATCAAC CAGCGCCTGC TGGCCGAGGA 254 [R75Q](#) [E84K](#)
LeuThrLeuG lnValLeuAr gAlaIleAsn GlnArgLeuL euAlaGluGl 85

GCTCCACAGG GCAGCCATTC AGGAATATTC CACACAAGAA AACGGCACAG 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 ACCA~~CCC~~CGA GGGGAACGAG GGGAA~~CGG~~CC CTCGGCCGTA 404 T120I H123Q 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

GGCTGTTCGAG GAAGCCCCTG AGCAAACGCA GAGAGAAGGC CTCGGAGGGC 504 S154P R155T S159N E163A E163D A165T A165A S166L S166S E167D

lyLeuSerAr gLysProLeu SerLysArgA rgGluLysAl aSerGluGly 168

CTGGACGCGC AGGGCAAGCC TCGGACCCGG AGCCCGGCC TGCCGGGCGG 554 L169V Q172P P175H T177I S179I S179N P180R P180P P183T P183P

LeuAspAlaG lnGlyLysPr oArgThrArg SerProAlaL euProGlyGl 185

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yArgSerPro GlyProCysA rgAlaLeuGl uGlyGlyGln AlaGluValA 202

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rgLeuArgAr gAsnAlaSer SerAlaGlyA rgLeuGlnGl yLeuAlaGly 218

GG~~CGCCCC~~GG GGCAG~~AAGGA~~ G~~TGC~~AGGCC TTCGAAGTGT ACC~~TGCC~~CTC 704 G219G P221P G222R K224del E225G E225D E230K E230Q Y232H c.698_700dupTGC P234P S235L

GlyAlaProG lyGlnLysGl uCysArgPro PheGluValT yrLeuProSe 235

GGGAAAGATG CGACCTAGAA GCCTTGAGGT CACCA~~TTT~~CT A~~CAG~~GGGAGA 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

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AlaAsnLeuA spSerAlaTh rGluProArg AlaArgProT hrProAspGl 285

AGGGGCATCT GCGGACCTGA AGGAAGGCC TGGAAATCCA GAACATTCGG 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](#)
GluGlyAspP roValAspGl yThrCysVal ArgAspSerC ysSerPhePr 335

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ProGlnProL euProGlnCy sLysArgHis LeuLysGlnV alGlnLeuLe 385

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uPheCysGlu AspHisAspG luProIleCy sLeuIleCys SerLeuSerG 402

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lnGluHisGl nGlyHisArg ValArgProI leGluGluVa lAlaLeuGlu 418

CACAAGAAGA AAATTCAGAA GCAGCTGGAG CATCTGAAGA AGCTGAGAAA 1304 I423V I423T Q426R
HisLysLysL ysIleGlnLy sGlnLeuGlu HisLeuLysL ysLeuArgLy 435

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sSerGlyGlu GluGlnArgS erTyrGlyGl uGluLysAla ValSerPheL 452

TGAAACAAAC TGAAGCGCTG AAGCAGCGGG TGCAGAGGAA GCTGGAGCAG 1404 E456D A457V A457A R461Q Q468Q
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ValTyrTyrP heLeuGluGl nGlnGluHis PhePheValA laSerLeuGl 485

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

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gPheAspSer CysIleIleV alLeuGlySe rProSerPhe LeuSerGlyA 652

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TAACAAAATA GTTACTGTGC CCACGGAGCC TACCCGATTA TAGCAGAGGT *158 c.*133G>A
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ACACACCATG GATTTTCAGAG GAGGAAGTAC GGAGTCGTTG CATAATCCGC *258 c.*245G>A
CCCTGGTGGG TGGCACTCTC AGGTGCTCCT GAACAGAAGA TTTGGCCCTC *308 c.*267G>A
ATTTTCCCTC AGAACCCAC GGCAAGGATA TATGTCCCCT TGTTCCTCTCT *358
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