



MEFV (NM_000243.3) - cDNA - 2024-05-20

CTACCA <u>G</u> AAG CCAGACAGCT GGCTCGAGCC TCTC <u>C</u> TGCTC AGCACC <u>A</u> T <u>G</u> G	4	chr16:g.3320350_3256171dup -12C>G
CTAAGACCCC TAGTGACCAT <u>C</u> TGCTGTCCA <u>C</u> CCTGGAGGA GCTGGTGCCC	54	L9L T12I
T <u>A</u> TGACTTCG AGAAGTTCA <u>A</u> GTTCAAG <u>C</u> TG CAGAACACCA GTGTGC <u>A</u> GA <u>A</u>	104	Y19C K25R L28V Q34P K35R
GGAGCACTCC <u>A</u> GGATCCCC <u>C</u> GGAGCCAGAT CCAGAGAGCC AGGC <u>C</u> GGTGA	154	R39G R42W P50L
AGATGGCCAC TCTGCT <u>G</u> GTC ACCTACTAT <u>G</u> GGAAGAGTA <u>C</u> GCCG <u>T</u> GCAG	204	L57L G62W Y65Y V67E
CTCACCCTGC AGGTCCTGC <u>G</u> GGCCATCAAC CAGCGCCTGC TGGCC <u>G</u> AGGA	254	R75Q E84K
GCTCC <u>A</u> CAGG <u>G</u> CAGCCATTC AGGAATATTC CACA <u>C</u> AAGAA AA <u>C</u> GGCACAG	304	H87R A89T Q97K Q97X Q97R N99N
A <u>T</u> GAT <u>T</u> C <u>G</u> C <u>A</u> GCCTCC <u>A</u> GC TCCC <u>T</u> GGGGG AGAACAAAG <u>C</u> C CAG <u>G</u> AGCCTG	354	D102D D103D S104C A105E S108R S108G L110P L110L G111R G111E G111G 334_335insG P115T
P115R R116S		
AAGAC <u>T</u> CCAG ACCA <u>C</u> CC <u>C</u> GA <u>G</u> GGGAAC <u>G</u> AG GGGAA <u>C</u> GGCC CTCGGCCG <u>T</u> A	404	T120I H123Q P124P E125E E128_N130del 390_391insGAGGGGAAC N130N Y135H
<u>C</u> GGGGGC <u>G</u> GA GCTGCCA <u>G</u> CC TGC <u>G</u> TGC <u>A</u> G CCAGCC <u>C</u> GAG GCC <u>G</u> G <u>A</u> G <u>G</u> G	454	G136R G136W G136E G136G G138G S141I R143P S145G E148RfsX5 E148Q E148V E148D G150G
R151S G152R		
GGCTG <u>T</u> CGA <u>G</u> GAAGCCCCTG <u>A</u> GCAAACGCA GAG <u>A</u> GAAG <u>G</u> C <u>C</u> T <u>C</u> GGAG <u>G</u> GGC	504	S154P R155I S159N E163A E163D A165I A165A S166L S166S E167D
<u>C</u> TGGACGCGC <u>A</u> GGGCAAG <u>C</u> C TCGGA <u>C</u> CCGG <u>A</u> GCC <u>C</u> G <u>G</u> CCC T <u>G</u> <u>C</u> <u>C</u> G <u>G</u> GCGG	554	L169V Q172P P175H T177I S179I S179N P180R P180P P183I P183P
GAGAA <u>G</u> CCCC GGCCCCTGCA GGG <u>C</u> GCTAG <u>A</u> <u>G</u> GGGGGCCAG GCCGAGGTCC	604	S187N P188P A193V L194P E195E G196W G196R G197R
<u>G</u> GCT <u>G</u> CGCAG AA <u>A</u> CGCCA <u>G</u> C TCCGC <u>G</u> GGGA GGCTGCAGGG GCTGGCGG <u>G</u> G	654	R202Q L203P R204H N206S N206K 606_621dup S208C S208T S209S G211G G218A
GGC <u>G</u> CCCCGG GGCAG <u>A</u> AGGA <u>G</u> TGCAGGCC TT <u>C</u> GAAGT <u>G</u> T ACC <u>T</u> GCC <u>C</u> T <u>C</u>	704	G219G P221P G222R K224del E225G E225D E230K E230Q Y232H c.698_700dupTGC P234P S235L
GGGAAAGAT <u>G</u> CGA <u>C</u> CTAGAA <u>G</u> CCTT <u>G</u> AGGT CACC <u>A</u> TTTCT <u>A</u> CAGGGGAGA	754	G236V M238I R239R R241K S242G S242S S242R_C>G S242R_C>A E244K I247V T249A G250A E251K
AGGCGC <u>C</u> CGC AAATC <u>C</u> AGAA <u>A</u> TTCTCCTGA CTCTAGAGGA AA <u>A</u> GACAG <u>C</u> T	804	761_764dup P257L I259V K266E T267I A268V
GCG <u>A</u> ATCTGG ACT <u>C</u> GGCAAC AGAA <u>C</u> CCCCG GCAAGGCCCA CTC <u>C</u> GGATGG	854	N270D S273L P277I R278P P283R P283L
AGGGGCAT <u>C</u> T <u>G</u> CGGACCTGA AGGAAGGCC TGGAAATCCA G <u>A</u> ACATTCCG	904	S288Y A289V A289E E299G
TCACC <u>G</u> GAAG GCCACC <u>A</u> GAC <u>A</u> C <u>G</u> GCTGCGA GTCC <u>C</u> C <u>G</u> CTG CCAC <u>G</u> CCAG	954	G304R P307P T309M P313H R314C R314H R314R A317T

GAAGGAGACC CAGTTGACGG TACCTGTGTG CGTGATTCCT GCAGCTTCCC 1004 E319K V328A R329H
CGAGGCAGTT TCTGGGCACC CCCAGGCCTC AGGCAGCCGC TCACCTGGCT 1054 E336K V338L S339E R348H P350S P350R
GCCCCCGGTG CCAGGACTCC CATGAAAGGA AGAGCCCGGG AAGCCTAAGC 1104 C352C P353A R354W R361T S363N S363S P364L P364P L367V
CCCCAGCCCC TGCACAGTG TAAGCGCCAC CTGAAGCAGG TCAGCTGCT 1154 P369S L372P P373L K376R Q383K L384P
CTTCTGTGAG GATCACGATG AGCCCATCTG CCTCATCTGC AGTCTGAGTC 1204 D389V D391N P393P L396F
AGGAGCACC AAGGCCACC GG GTGC GCCCA TTGAGGAGGT CGCCCTGGA 1254 E403K H404R R408Q c.1229G>A V415V E418*
CACAAGAAGA AAATTCAGAA GCAGCTGGAG CATCTGAAGA AGCTGAGAAA 1304 I423V I423T Q426R
ATCAGGGGAG GAGCAGCGAT CCTATGGGGA GGAGAAGGCA GTGAGCTTTC 1354 Q440E S442T K447M
TGAAACAAAC TGAAGCGCTG AAGCAGCGGG TGCAGAGGAA GCTGGAGCAG 1404 E456D A457V A457A R461Q Q468Q
GTGTACTACT TCCTGGAGCA GCAAGAGCAT TTCTTTGTGG CCTCACTGGA 1454 V469L Y471X E474K E474E Q476Q H478Y F479L L484M
GGACGTGGGC CAGATGGTTG GGCAGATCAG GAAGGCATAT GACACC CGCG 1504 V487M V487L Q489Q R495K R501G R501C R501H R501R
TATCCCAGGA CATCGCCCTG CTCGATGCGC TGAATGGGGA ACTGGAGGCC 1554 S503C D505G I506V I506I L508Q D510N D510D A511V A511E I513T G514E
AAGGAGTGCC AGTCAGAATG GGAAC TTCTG CAGGACATTG GAGACATCTT 1604 E520V
GCACAGGGCT AAGACAGTGC CTGTCCCTGA AAAGTGGACC ACTCCTCAAG 1654 K539N P550A
AGATAAAACA AAAGATCCAA CTCCTCCACC AGAAGTCAGA GTTTGTGGAG 1704 E552D L559E
AAGAGCACA AAGTACTTCTC AGAAACCCTG CGTTCAGAAA TGGAAATGTT 1754 T577SAT T577A T577N T577SCG M582L E583A
CAATGTTCCG GAGCTGATTG GCGCTCAGGC ACATGCTGTT AATGTGATTC 1804 P588P I591T I591M G592G A595V c.1792G>A N599D
TGATGCAGA AACCGCTTAC CCAACCTCA TCTTCTCTGA TGATCTGAAG 1854 L602L T606T P609P N610D
AGTGTTAGAC TTGGAACAA GTGGGAGAGG CTGCCTGATG GCCCGCAAAG 1904 W626R R628K P630AfsX2 G632S G632A P633L
ATTGACAGC TGTATCATTG TTCTGGGCTC TCCGAGTTTC CTCTCTGGCC 1954 F636Y D637G I640M I641F V642A P646L P646P L649F L649P S650Y G651S R652C
GCCGTTACTG GAGAGGTGGAG GTTGGAGACA AGACAGCATG GATCCTGGA 2004 R652H R652R R653S R653H E656A V659F D661N D661Y K662Dfs*36 I666V G668R
GCCTGCAAGA CATCCATAAG CAGGAAAGGG AACATGACTC TGTCGCCAGA 2054 K671M S675N G678R G678E N679H M680L M680V M680IGC M680IGA M680IGT T681I S683S E685K
GAATGGCTAC TGGGTGGTGA TAATGATGAA GAAAAATGAG TACCAGGCCT 2104 G687D Y688C Y688F Y688X V690L V690G V691G V691A I692DEL M693K M693I M694V M694L
M694DEL M694K M694I K695R K695M K695N E698D A701A
CCAGCGTTCC CCCGACC CGC CTGCTAATAA AGGAGCCTCC CAAGCGTGTG 2154 S702C S703S V704I P705S P706P R708C L709R R717S R717H R717L
GGCATCTTCG TGGACTACAG AGTTGGAAGC ATCTCCTTTT ACAATGTGAC 2204 I720M F721L F721F V722M D723D V726A I729V I729M S730C N733D N733S
AGCCAATCC CACATCTATA CATTCGCCAG CTGCTCTTTC TCTGGGCCCC 2254 R737K F743Y F743L F743F A744S A744T A744D S747C S749C
TTCAACCTAT CTTCAGCCCT GGGACCGTG ATGGAGGGA GAACACAGCT 2304 Q753Q Q753H P754R I755V P758S R761C R761H G764G N766H

CCTCTGACTA TCTGTCCAGT GGGTGGTCAG GGGCCTGACT GAATGCCCAA *8 P769A I772V Q778Sfs*4 Q778L G779G P780T
CACIGCACTCT CTCTTTCCTGC TTCTGGCCTT GTATCTTGCA TTCACACTCA *58 *9C>T *12T>C *21C>G
ATAGTCACGG AATGCCGACT AGGTGCTAGC TGCTATGGGA AATGCAAAAA *108
TAACAAAATA GTTACTGTGC CCACGGAGCC TACCCGATTA TAGCAGAGGT *158 c.*133G>A
AAGTTAGGAA CGAACATGTT AGTCAATCCG GGTGAAGACA TGTACTGATG *208
ACACACCATG GATTTCAGAG GAGGAAGTAC GGAGTCGTTG CATAATCCGC *258 c.*245G>A
CCCTGGTGGG TGGCACTCTC AGGTGCTCCT GAACAGAAGA TTTGGCCCTC *308 c.*267G>A
ATTTTCCCTC AGAACCCAC GGCAAGGATA TATGTCCCCT TGTCTCTCT *358
GCTTCTGTCT TGAGGATATG GGAAGCCTAG AGAAACGCAA GCAGACTGGA *408
TTGGGATAGA AGTATTTGTG TACCTGGATT AATGAACTAT GATTTTTTTT *458
TTTTTTTTTT GAGACCAAAT CTTGCTCTGT GGCCCAAGGCT GGAGTGCAGT *508
GGCACGATCT CAGCTCACTG CAACCTCCAC CTCCCAGGTT CAAGCGATTC *558
TCCTGCCTCA GCCTCCTGAG CAGCTGGGAT TACAGGTGCG TGCCACCACA *608
CCAGGCTGGT TTTCTTGTAT TTTTAGTAGA GACGGGGGTT TCACCATGTT *658
AGCCAGGCTG GTCTCGAACT CCTGACCTCA GGTGATCCAC CCGCCTCAGC *708
CTCCCAAAGT GCTGGGATTA CAGGCATGAG CCACTGTGCC CGGCCTATGA *758
TTCTTTTTTT TTTTTTTTTT TGAGACAAAG TTTTGCTCTT GTCACCCAGG *808
CTGGAGTGCA GTGGTGCAAT CTTGGCTCGC AACCTCCGCC TCCCAGGTTT *858
AAGAGATTCT CCTGCCTCAG CCTCCGAAGT AGCTGGGATT ACAGGCGCCC *908
GCCACCATGC CCGGCTAATT TTTTGCAATT TTAGTAGACA TGAGGTTTCA *958
TCATGTTGGC CAGGCCGGTC TCAAACCTCT GACCTCAGGT GATGCACCCA *1008
CCTCAGCCTC CCAAAGTGCA GGGATTACAG GCATGAGCCA CCATGCCGGG *1058
CCATGATTCT TAAGAGAATT GACTGGGCCT CATGAATAAA AAAATTAGAA *1108
AATCTG

MEFV (NM_000243.3) - cDNA - 2024-05-20

