ADA2 (NM_001282225.2) - cDNA + Protein - 2025-06-02

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AGTTGGTGAG CTTTTCCGGT GCTCTGCACA GATGCTGGGG CGCTGAGCAA -77
ACAGCCCTCA GTTTCTGGAG CTGTTCCGAG TCCCGTGGAG TCTCCATCTG -27   -43C>T c.-42G>A -31A>G
AGCCCTTTCC TAGTCCAGGC ATCCCGATGT TGGTGGATGG CCCATCTGAG 24   c.-14G>T M1T P6L
                           MetL euValAspGl yProSerGlu 8

CGGCCAGCCC TGTGCTTCTT GCTGTTGGCT GTGGCAATGT CTTTCTTCGG 74   R9W G25C
ArgProAlaL euCysPheLe uLeuLeuAla ValAlaMetS erPhePheGl 25

CTCAGCTCTA TCCATAGATG AAACACGGGC GCATCTGTTG TTGAAAGAAA 124  I30T I33Nfs*29 R34W
ySerAlaLeu SerIleAspG luThrArgAl aHisLeuLeu LeuLysGluL 42

AGATGATGCG GCTGGGGGGG CGGCTGGTGC TGAACACCAA GGAGGAGCTG 174  R45W c.138G>C G47R G47W G47R_GC G47A G47V G48R c.144delG R49Afs*13 R49W c.158del N53N

K13del
ysMetMetAr gLeuGlyGly ArgLeuValL euAsnThrLy sGluGluLeu 58

GCCAATGAGA GGCTCATGAC GCTCAAAATC GCTGAGATGA AGGAGGCCAT 224
AlaAsnGluA rgLeuMetTh rLeuLysIle AlaGluMetL ysGluAlaMe 75

GAGGACCCTG ATATTCCCAC CCAGCATGCA CTTTTTCCAG GCCAAGCATC 274  c.232_322+105delinsATG H91R H91Lfs
tArgThrLeu IleProProP roSerMetHi sPhePheGln AlaLysHisL 92
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TCATTGAGAG AAGTCAAGTG TTTAATATTC TAAGGATGAT GCCAAAAGGG 324
euIleGluAr gSerGlnVal PheAsnIleL euArgMetMe tProLysGly 108

GCTGCCTTGC ACCTCCATGA CATTGGCATC GTGACTATGG ACTGGCTGGT 374
AlaAlaLeuH isLeuHisAs pIleGlyIle ValThrMetA spTrpLeuVa 125

GAGGAATGTC ACCTACAGGC CTCACTGCCA CATCTGTTTC ACCCCAAGGG 424
lArgAsnVal ThrTyrArgP roHisCysHi sIleCysPhe ThrProArgG 142

GGATCATGCA GTTCAGATTT GCTCACCCAA CTCCCCGTCC ATCAGAAAAA 474
lyIleMetGl nPheArgPhe AlaHisProT hrProArgPr oSerGluLys 158

TGTTCCAAGT GGATTCTGCT GGAGGATTAT CGGAAGCGGG TGCAGAACGT 524
CysSerLysT rpIleLeuLe uGluAspTyr ArgLysArgV alGlnAsnVa 175

CACTGAGTTT GATGACAGCT TGCTGAGGAA TTTCACTCTG GTGACCCAGC 574
lThrGluPhe AspAspSerL euLeuArgAs nPheThrLeu ValThrGlnH 192

ACCCGGAGGT GATTTACACA AACCAAAATG TTGTCTGGTC GAAATTTGAA 624
isProGluVa lIleTyrThr AsnGlnAsnV alValTrpSe rLysPheGlu 208

ACCATCTTCT TCACCATCTC TGGTCTCATC CATTACGCAC CAGTGTTCAG 674
ThrIlePheP heThrIleSe rGlyLeuIle HisTyrAlaP roValPheAr 225

AGACTATGTC TTCCGGAGCA TGCAGGAGTT CTACGAGGAC AACGTGCTCT 724
gAspTyrVal PheArgSerM etGlnGluPh eTyrGluAsp AsnValLeuT 242

ACATGGAGAT CAGAGCCAGG CTGCTGCCGG TGTATGAGCT CAGTGGAGAG 774
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I93T P106S p.A109Lfs*11

A109D H112Y H112Q 116Lfs*4

T129P p.(Y130Sfs*48) R131Sfs c.396_397del C134Y

I143Sfs*41 P151QfsX P155Hfs*29

C159Y W162R R169G R169Q

F178S L183P T187P L188V L188P

P193L V203A W204C G>T W204C F207S

I210Tfs F212del Y220X

Y227fs M232T Y236del c.709delC (p.Glu237fs) D238N A247Qfs*16

M243R E244A A247V L249P P251L P251P

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yrMetGluIl eArgAlaArg LeuLeuProV alTyrGluLe uSerGlyGlu 258

CACCATGACG AAGAGTGGTC AGTGAAGACT TACCAGGAAG TAGCTCAGAA 824  D261Pfs*2 W264S W264Ter S265X
HisHisAspG luGluTrpSe rValLysThr TyrGlnGluV alAlaGlnLy 275

GTTTGTGGAA ACTCACCCTG AGTTTATTGG AATCAAAATC ATTTATTCGG 874  F283L S291L
sPheValGlu ThrHisProG luPheIleGl yIleLysIle IleTyrSerA 292

ATCACAGATC CAAAGATGTG GCTGTCATCG CAGAATCCAT CCGAATGGCC 924  R306*
spHisArgSe rLysAspVal AlaValIleA laGluSerIl eArgMetAla 308

ATGGGGCTCC GAATCAAGTT CCCCACGGTG GTGGCAGGGT TTGACCTGGT 974  M309I L311R R312ter T317M G321E G321A delEx7 Dup Exon7
MetGlyLeuA rgIleLysPh eProThrVal ValAlaGlyP heAspLeuVa 325

GGGGCATGAG GACACTGGCC ACTCCTTGCA TGACTACAAG GAAGCTCTGA 1024  G326V E328K E328D D329N H335R
lGlyHisGlu AspThrGlyH isSerLeuHi sAspTyrLys GluAlaLeuM 342

TGATCCCCGC CAAGGATGGC GTTAAGCTGC CTTACTTCTT CCACGCCGGA 1074  P344L V349I L351Q Y353H F355L A357T G358R
etIleProAl aLysAspGly ValLysLeuP roTyrPhePh eHisAlaGly 358

GAAACAGACT GGCAGGGTAC TTCCATAGAC AGGAACATTC TGGATGCTCT 1124  T119A N370K
GluThrAspT rpGlnGlyTh rSerIleAsp ArgAsnIleL euAspAlaLe 375

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uMetLeuAsn ThrThrArgI leGlyHisGl yPheAlaLeu SerLysHisP 392

CCGCAGTCAG GACTTACTCC TGGAAAAAGG ACATCCCCAT AGAAGTCTGT 1224  W399X I405L C408Y
roAlaValAr gThrTyrSer TrpLysLysA spIleProIl eGluValCys 408
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CCCATCTCTA ACCAGGTGCT GAAACTGGTG TCTGACTTGA GGAACCACCC 1274 P409S P409H V372M N423K P425A

ProIleSerA snGlnValLe uLysLeuVal SerAspLeuA rgAsnHisPr 425

TGTAGCCACT CTGATGGCCA CTGGGCACCC CATGGTGATC AGCTCTGATG 1324 P435A M436T

oValAlaThr LeuMetAlaT hrGlyHisPr oMetValIle SerSerAspA 442

ACCCAGCTAT GTTTGGTGCC AAAGGCTTGT CCTATGATTT CTATGAGGTC 1374 P443A M445T F404S K449Nfs*2 G450C L451W L451F Y453C Y453Y D454H Y456C V458D

spProAlaMe tPheGlyAla LysGlyLeuS erTyrAspPh eTyrGluVal 458

TTCATGGGCA TTGGGGGGAT GAAGGCTGAC CTGAGGACCC TCAAACAGCT 1424 p.(M460K) M465fsX K466Tfs*2

PheMetGlyI leGlyGlyMe tLysAlaAsp LeuArgThrL euLysGlnLe 475

GGCCATGAAC TCTATCAAGT ACAGTACCCT GTTGGAGAGT GAGAAAAATA 1474 S479P Y482C S483Pfs* E489Q E489D

uAlaMetAsn SerIleLysT yrSerThrLe uLeuGluSer GluLysAsnT 492

CTTTCATGGA AATCTGGAAG AAGAGATGGG ATAAGTTCAT AGCAGATGTG 1524 W501*

hrPheMetGl uIleTrpLys LysArgTrpA spLysPheIl eAlaAspVal 508

GCTACAAAGT GAGGAGAAGC TAGCCAGCCC TCTACAAGCT GTCTTCTTGC *38

AlaThrLysS top

ACACGCTGTC ACTTCCTCTC ACTCGTTCTT GAATCAGCTC CATGTGCCCA *88

TGAAATCAAT GGCCTCTGTA TGGAGCGACC CTGTGAGAAG CACTTGGCTG *138

GCTGAGCAAA TTCATCCTCT GGAAATATTC TCTCTCAGCC ACAGTGACAT *188 *159G>A

TGACCCTCTT GGTTTTCTCC TGTCTCTGGC CATTTCTTCC AGTTTCCCTA *238

TTTCAGAGTC TTCTCCTCTC TCTGATCTCT GTGCTGTTTC CTCAGGACTC *288

AGTCCTGGGC TCTCTTCTAT TCTGGTCTCT TTATTTTTTT ATTTTTGTAT *338

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GCGATCTCAG CTCAGTGCAA CCTCCGCCAC CCGGGTTCAG GCAATTCTCT *438

TGCATCAGCC TCGCGAGTAG TTGGAATTAT AGGCATGTGC CACCACACCC *488

AGCTGATTTT TGCATTTTTA GTAGAGACAG GTTTTCACCA TGTTGGCGAG *538

GCTGGTATCC AACTCTTGAC CTCAGGTGAT CCACTCGCCC CTTGGCTCCC *588

AAAGTGCTGG AATTACAGGC ATTAGCCACC ATGCCTGGCC TATTCTGGTC *638

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GGTCTCCAAC TCAAGTGATC CTCCTGCCGC AGTCTCCCGA AGTGCTGGGA *938

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CCCAACCCCG GCGGCCCTTT CTTCCTCAAA TGTCCTCGGC TCTATACCGT *1838

GCCTGGGTCT TTTCTCTTTC TCTCTGCCTG GAAGATTCCT TCTTTCCCCT *1888

TTTGTCTTGC CCACTCCTGT TTACCCTTCA AGTTTCAAGT TCATGTCACT *1938

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CTCTTTTCCA TCATGCCTCT AATCACAGCT TATAATCGGA TATTTATTTC *2038

TGTGTCTACA GTCTTGCCCT GCCAGACTGT ATGCCCCATG TGGGCAGGCG *2088

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CGCCCCAAAT AAAAGACCTC ATGTGATCAG AGTCCAGCTC CTCACCTGGC *2288

CTTGGAAGAA GGATGCAAGA AGCCACCTCT GTCCTCACCC AACTCAAGGA *2338

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CTTGC

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