



ADA2 (NM_001282225.2) - cDNA + Protein - 2026-05-31

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 TCCA TAGATG AAACACGGGC GCATCTGTTG TTGAAAGAAA 124 I30T T33Nfs*29 R34W
ySerAlaLeu SerIleAspG luThrArgAl aHisLeuLeu LeuLysGluL 42

AGATGATGCG GCTGGGGGGG CCGCTGGTGC 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 GCTCAAATC GCTGAGATGA AGGAGGCCAT 224
AlaAsnGluA rgLeuMetTh rLeuLysIle AlaGluMetL ysGluAlaMe 75

GAGGACCCTG ATATTCCCAC CCAGCATGCA CTTTTTCCAG GCCAAGCAATC 274 c.232_322+105delinsATG H91R H91Lfs
tArgThrLeu IlePheProp roSerMetHi sPhePheGln AlaLysHisL 92

TCATTGAGAG AAGTCAAGTG TTTAATATTC TAAGGATGAT GCCAAAAGGG 324 I93T P106S p.A109Lfs*11
 euIleGluAr gSerGlnVal PheAsnIleL euArgMetMe tProLysGly 108

GCTGCCTTGC ACCTCCATGA CATTGGCATC GTGACTATGG ACTGGCTGGT 374 A109D H112Y H112Q I16Lfs*4 M121T
 AlaAlaLeuH isLeuHisAs pIleGlyIle ValThrMetA spTrpLeuVa 125

GAGGAATGTC ACCTACAGGC CTCACTGCCA CATCTGTTTC ACCCCAAGGG 424 N127I T129P p.(Y130Sfs*48) R131Sfs c.396_397del C134Y
 lArgAsnVal ThrTyrArgP roHisCysHi sIleCysPhe ThrProArgG 142

GGAATCATGCA GTTCAGATTT GCTCACC~~CAA~~ CTCCCCGTCC ATCAGAAAAA 474 I143Sfs*41 P151OfsX P155Hfs*29
 lyIleMetGl nPheArgPhe AlaHisProT hrProArgPr oSerGluLys 158

TGTTC~~CAAGT~~ GGATTCTGCT GGAGGATTAT CGGAAGCGGG TGCAGAACGT 524 C159Y W162R R169G R169Q
 CysSerLysT rpIleLeuLe uGluAspTyr ArgLysArgV alGlnAsnVa 175

CACTGAGT~~TT~~ GATGACAGCT TGC~~T~~GAGGAA TTTCA~~CTCTG~~ GTGACCCAGC 574 F178S L183P T187P L188V L188P
 lThrGluPhe AspAspSerL euLeuArgAs nPheThrLeu ValThrGlnH 192

ACCCGGAGGT GATTTACACA AACCAAAATG TTGTCTGG~~GTC~~ GAAAT~~TTGAA~~ 624 P193L V203A W204C G>T W204C F207S
 isProGluVa lIleTyrThr AsnGlnAsnV alValTrpSe rLysPheGlu 208

ACCA~~TCTTCT~~ TCACCATCTC TGGTCTCATC CATTAC~~GCAC~~ CAGTGTTCAG 674 I210Tfs F212del Y220X
 ThrIlePheP heThrIleSe rGlyLeuIle HisTyrAlaP roValPheAr 225

AGACT~~ATGTC~~ TTCCGGAGCA TGCAGGAGTT CTAC~~GAGGAC~~ AACGTGCTCT 724 Y227fs M232T Y236del c.709delC (p.Glu237fs) D238N A247Ofs*16
 gAspTyrVal PheArgSerM etGlnGluPh eTyrGluAsp AsnValLeuT 242

ACATGG~~AGAT~~ CAGAG~~C~~CAGG CTGCTGCC~~CGG~~ TGTATGAGCT CAGTGGAGAG 774 M243R E244A A247V L249P P251L P251P

yrMetGluIl eArgAlaArg LeuLeuProV alTyrGluLe uSerGlyGlu 258

CACCATGACG AAGAGTGGTC AGTGAAGACT TACCAGGAAG TAGCTCAGAA 824 [D261Pfs*2](#) [W264S](#) [W264Ter](#) [S265X](#)

HisHisAspG luGluTrpSe rValLysThr TyrGlnGluV alAlaGlnLy 275

GTTTGTGGAA ACTCACCCCTG AGTTIATTGG AATCAAAATC ATTTATTGG 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 TGACTIONAAG GAAGCTCTGA 1024 [G326V](#) [E328K](#) [E328D](#) [D329N](#) [H335R](#)

lGlyHisGlu AspThrGlyH isSerLeuHi sAspTyrLys GluAlaLeuM 342

TGATCCCGC CAAGGATGGC GTTAAGCTGC CTIACCTTCTT CCACGCCGGA 1074 [P344L](#) [V349I](#) [L351Q](#) [Y353H](#) [F355L](#) [A357T](#) [G358R](#)

etIleProAl aLysAspGly ValLysLeuP roTyrPhePh eHisAlaGly 358

GAAACAGACT GGCAGGGTAC TTCCATAGAC AGGAACTTC 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](#)

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CCCATCTCTA ACCAGGTGCT GAAACTGGTG TCTGACTTGA GGAACCACCC 1274 P409S P409H V372M N423K P425A

ProIleSerA snGlnValLe uLysLeuVal SerAspLeuA rgAsnHisPr 425

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oValAlaThr LeuMetAlaT hrGlyHisPr oMetValIle SerSerAspA 442

ACCAGCTAT GTTGGTGCC AAAGGCTTGT CCTATGATTT CTATGAGGTC 1374 P443A M445T F404S K449Nfs*2 G450C G450= 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

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hrPheMetGl uIleTrpLys LysArgTrpA spLysPheIl eAlaAspVal 508

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AlaThrLysS top

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TGACCCTCTT GGTTTTCTCC TGTCTCTGGC CATTTCTTCC AGTTTCCCTA *238

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CTTGC

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