



ADA2 (NM_001282225.2) - cDNA + Protein - 2024-05-17

AGTTGGTGAG CTTTTCGGT GCTCTGCACA GATGCTGGGG CGCTGAGCAA -77

ACAGCCCTCA GTTCTGGAG 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 TCCATIAGATG AAACACGGGC GCATCTGTTG TTGAAAGAAA 124 I30I I33Nfs*29 R34W

ySerAlaLeu SerIleAspG luThrArgAl aHisLeuLeu LeuLysGluL 42

AGATGATGCG GCTGGGGGGGG CGGCTGGTGC TGAACCAAA GGAGGAGCTG 174 R45W c.138G>C G47R G47W G47R_GC G47A G47V G48R R49Afs*13 c.144delG 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 IlePheProP roSerMetHi sPhePheGln AlaLysHisL 92

TCAITGAGAG AAGTCAAGTG TTTAATATTC TAAGGATGAT GCCAAAAGGG 324 I93I P106S p.A109Lfs*11
 euIleGluAr gSerGlnVal PheAsnIleL euArgMetMe tProLysGly 108

GCTGCCTTGC AACTCCATGA CATTGGCATC GTGACTATGG ACTGGCTGGT 374 A109D H112Y H112Q 116Lfs*4
 AlaAlaLeuH isLeuHisAs pIleGlyIle ValThrMetA spTrpLeuVa 125

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

GGATCATGCA GTTCAGATTT GCTCACCCAA CTCCCCGTCC ATCAGAAAAA 474 I143Sfs*41 P151QfsX P155Hfs*29
 lyIleMetGl nPheArgPhe AlaHisProT hrProArgPr oSerGluLys 158

TGTTCCAAGT GGATTCTGCT GGAGGATTAT CGGAAGCGGG TGCAGAACGT 524 C159Y W162R R169G R169Q
 CysSerLysT rpIleLeuLe uGluAspTyr ArgLysArgV alGlnAsnVa 175

CACTGAGTIT GATGACAGCT TGCIGAGGAA TTTCACTCTG GTGACCCAGC 574 F178S L183P T187P L188V L188P
 lThrGluPhe AspAspSerL euLeuArgAs nPheThrLeu ValThrGlnH 192

ACCCGGAGGT GATTACACA AACCAAAATG TTGTCTGGTC GAAATITGAA 624 P193L V203A W204C G>T W204C P207S
 isProGluVa lIleTyrThr AsnGlnAsnV alValTrpSe rLysPheGlu 208

ACCAICTTCT TCACCATCTC TGGTCICATC CATTACGCAC CAGTGTTCAG 674 I210Tfs F212del V217D Y220X
 ThrIlePheP heThrIleSe rGlyLeuIle HisTyrAlaP roValPheAr 225

AGACTATGTC TTCCGGAGCA TGCAGGAGTT CTACGAGGAC AACGTGCTCT 724 Y227fs M232T Y236del c.709delC (p.Glu237fs) D238N A247Qfs*16
 gAspTyrVal PheArgSerM etGlnGluPh eTyrGluAsp AsnValLeuT 242

ACAITGGAGAT CAGAGCCAGG CTGCTGCCGG TGTATGAGCT CAGTGGAGAG 774 M243R E244A A247V L249P P251L P251P

yrMetGluI1 eArgAlaArg LeuLeuProV alTyrGluLe uSerGlyGlu 258

CACCATGACG AAGAGTGGTC AGTGAAGACT TACCAGGAAG TAGCTCAGAA 824 [D261Pfs*2](#) [W264S](#) [W264Ter](#) [S265X](#)
HisHisAspG luGluTrpSe rValLysThr TyrGlnGluV alAlaGlnLy 275

GTTTGTGGAA ACTCACCTG AGTTIATTGG AATCAAAATC ATTTATTGG 874 [F283L](#) [S291L](#)
sPheValGlu ThrHisProG luPheIleGl yIleLysIle IleTyrSerA 292

ATCACAGATC CAAAGATGTG GCTGTCATCG CAGAATCCAT CCGAATGGCC 924 [R306*](#)
spHisArgSe rLysAspVal AlaValIleA laGluSerI1 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 TGACTIONAG GAAGCTCTGA 1024 [G326V](#) [E328K](#) [E328D](#) [D329N](#) [H335R](#)
lGlyHisGlu AspThrGlyH isSerLeuHi sAspTyrLys GluAlaLeuM 342

TGATCCCCGC CAAGGATGGC GTTAAGCTIGC CTIACTTCTT CCACGCCGGA 1074 [P344L](#) [V349I](#) [L351Q](#) [Y353H](#) [F355L](#) [A357T](#) [G358R](#)
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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

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spProAlaMe tPheGlyAla LysGlyLeuS erTyrAspPh eTyrGluVal 458

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hrPheMetGl uIleTrpLys LysArgTrpA spLysPheIl eAlaAspVal 508

GCTACAAAGT GAGGAGAAGC TAGCCAGCCC TCTACAAGCT GTCTTCTTGC *38

AlaThrLysS top

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CTCTTTTCCA TCATGCCTCT AATCACAGCT TATAATCGGA TATTTATTTT *2038
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CTTGC

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