



ADA2 (NM_001282225.2) - cDNA + Protein - 2024-11-21

AGTTGGTGAG CTTTTCCGGT GCTCTGCACA GATGCTGGGG CGCTGAGCAA -77
ACAGCCCTCA GTTTCTGGAG CTGTTCCGAG TCCCGTGGAG TCTCCATCTG -27 -43C>T c.-42G>A -31A>G
AGCCCTTTCC TAGTCCAGGC ATCCCGATGT TGGTGGATGG CCGCATCTGAG 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 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 R49Afs*13 c.144delG 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 GCCAAGCATC 274 c.232_322+105delinsATG H91R H91Lfs
tArgThrLeu IlePheProp roSerMetHi sPhePheGln AlaLysHisL 92

TCAT**T**GTGAGAG AAGTCAAGTG TTTAATATTC TAAGGATGAT **G**CCAAA**A**GGG 324 [I93T](#) [P106S](#) [p.A109Lfs*11](#)
 euIleGluAr gSerGlnVal PheAsnIleL euArgMetMe tProLysGly 108

GCTGCCT**T**GC **A**CCCTCCATGA **C**ATTGGC**A**T**C** GTGACTATGG ACTGGCTGGT 374 [A109D](#) [H112Y](#) [H112Q](#) [I16Lfs*4](#)
 AlaAlaLeuH isLeuHisAs pIleGlyIle ValThrMetA spTrpLeuVa 125

GAGGAATG**T**C **A**CC**T**A**C**AG**G**C **C**T**C**ACT**G**CC**A** CATCTGTTTC ACCCCAAGGG 424 [T129P](#) [p.\(Y130Sfs*48\)](#) [R131Sfs](#) [c.396_397del](#) [C134Y](#)
 lArgAsnVal ThrTyrArgP roHisCysHi sIleCysPhe ThrProArgG 142

GG**A**TCATGCA GTTCAGATTT GCTCACC**C**AA CTCCCCG**T**CC **A**TCAGAAAAA 474 [I143Sfs*41](#) [P151OfsX](#) [P155Hfs*29](#)
 lyIleMetGl nPheArgPhe AlaHisProT hrProArgPr oSerGluLys 158

TG**T**TC**C**AA**G**T **G**GATTCTGCT GGAGGATTAT **C**GGAAGCGGG TGCAGAACGT 524 [C159Y](#) [W162R](#) [R169G](#) [R169Q](#)
 CysSerLysT rpIleLeuLe uGluAspTyr ArgLysArgV alGlnAsnVa 175

CACTGAG**T**TT **G**ATGACAGCT TGC**T**GAGGAA TTT**C**A**C**T**C**T**G** GTGACCCAGC 574 [F178S](#) [L183P](#) [T187P](#) [L188V](#) [L188P](#)
 lThrGluPhe AspAspSerL euLeuArgAs nPheThrLeu ValThrGlnH 192

ACC**C**GGAGGT GATTTACACA AACCAAAATG TT**G**T**C**T**G**G**T**C GAAAT**T**TGAA 624 [P193L](#) [V203A](#) [W204C](#) [G>T](#) [W204C](#) [P207S](#)
 isProGluVa lIleTyrThr AsnGlnAsnV alValTrpSe rLysPheGlu 208

ACC**A**T**C**T**T**C**T** TCACCATCTC TGG**T**C**T**CATC CATT**A**C**G**CAC CAGTGTTCAG 674 [I210Tfs](#) [F212del](#) [V217D](#) [Y220X](#)
 ThrIlePheP heThrIleSe rGlyLeuIle HisTyrAlaP roValPheAr 225

AGACT**A**TGTC TTCCGGAGCA **T**GCAGGAGTT **C**T**A**C**G**AG**G**AC AACGTGCTCT 724 [Y227fs](#) [M232T](#) [Y236del](#) [c.709delC](#) [\(p.Glu237fs\)](#) [D238N](#) [A247Ofs*16](#)
 gAspTyrVal PheArgSerM etGlnGluPh eTyrGluAsp AsnValLeuT 242

ACA**T**GG**A**GAT CAGAG**C**CAGG **C**T**G**CT**G**CC**G**G 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

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GluThrAspT rpGlnGlyTh rSerIleAsp ArgAsnIleL euAspAlaLe 375

GATGCTGAAC ACTACCAGAA TCGGCCATGG ATTTGCTTTG AGCAAACAAC 1174 [G142S](#) [G383D](#) [H391Q](#)

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 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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CTTTCATGGA AATCTGGAAG AAGAGATGGG ATAAGTTCAT AGCAGATGTG 1524 W501*

hrPheMetGl uIleTrpLys LysArgTrpA spLysPheIl eAlaAspVal 508

GCTACAAAGT GAGGAGAAGC TAGCCAGCCC TCTACAAGCT GTCTTCTTGC *38

AlaThrLysS top

ACACGCTGTC ACTTCCTCTC ACTCGTTCCT GAATCAGCTC CATGTGCCCA *88

TGAAATCAAT GGCCTCTGTA TGGAGCGACC CTGTGAGAAG CACTTGGCTG *138

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

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GCTGGTATCC AACTCTTGAC CTCAGGTGAT CCACTCGCCC CTTGGCTCCC *588
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TTTGTCTTGC CCACTCCTGT TTACCCTTCA AGTTTCAAGT TCATGTCACT *1938
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CTCTTTTCCA TCATGCCTCT AATCACAGCT TATAATCGGA TATTTATTTT *2038
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CTTGC

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