



ADA2 (NM_001282225.2) - cDNA - 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 CCATCTGAG 24 c.-14G>T M1T P6L
CGGCCAGCCC TGTGCTTCTT GCTGTTGGCT GTGGCAATGT CTTTCTTCGG 74 R9W G25C
CTCAGCTCTA TCCATAGATG AAACA CGGGC GCATCTGTTG TTGAAAGAAA 124 I30T T33Nfs*29 R34W
AGATGATGCG GCTGGGGGGG CGGCTGGTGC TGAACACCAA GGAGGAGCTG 174 R45W c.138G>C G47R G47W G47R_GC G47A G47V G48R R49Afs*13 c.144delG R49W c.158del N53N

K13del

GCCAATGAGA GGCTCATGAC GCTCAAAATC GCTGAGATGA AGGAGGCCAT 224
GAGGACCCTG ATATTCCCAC CCAGCATGCA CTTTTTCCAG GCCAAGCATC 274 c.232_322+105delinsATG H91R H91Lfs
TCATTGAGAG AAGTCAAGTG TTTAATATTC TAAGGATGAT GCCAAAAGGG 324 I93T P106S p.A109Lfs*11
GCTGCCTTGC ACCCTCCATGA CATTGGCATC GTGACTATGG ACTGGCTGGT 374 A109D H112Y H112Q 116Lfs*4
GAGGAATGTC ACCTACAGGC CTCACTGCCA CATCTGTTTC ACCCAAGGG 424 T129P p.(Y130Sfs*48) R131Sfs c.396_397del C134Y
GGATCATGCA GTTCAGATTT GCTCACC~~CAA~~ CTCCCCGTC ATCAGAAAA 474 I143Sfs*41 P151OfsX P155Hfs*29
TGTTC~~CAAGT~~ GGATTCTGCT GGAGGATTAT CGGAAGCGGG TGCAGAACGT 524 C159Y W162R R169G R169Q
CACTGAGTIT GATGACAGCT TGCIGAGGAA TTTC~~ACTCTG~~ GTGACCCAGC 574 F178S L183P T187P L188V L188P
ACCCGGAGGT GATTTACACA AACCAAAATG TTGICTGGTC GAAATITGAA 624 P193L Y203A W204C_G>T W204C P207S
ACCATCTTCT TCACCATCTC TGGTCTCATC CATTACGCAC CAGTGTTTCTG 674 I210Tfs F212del Y217D Y220X
AGACTATGTC TTCCGGAGCA TGCAGGAGTT CTACGAGGAC AACGTGCTCT 724 Y227fs M232T Y236del c.709delC (p.Glu237fs) D238N A247Ofs*16
ACATGGAGAT CAGAGCCAGG CTGCTGCCGG TGTATGAGCT CAGTGGAGAG 774 M243R E244A A247V L249P P251L P251P
CACCATGACG AAGAGTGGTC AGTGAAGACT TACCAGGAAG TAGCTCAGAA 824 D261Pfs*2 W264S W264Ter S265X
GTTTGTGGAA ACTCACCTG AGTTIATTGG AATCAAAATC ATTTATT~~CGG~~ 874 F283L S291L

ATCACAGATC CAAAGATGTG GCTGTCATCG CAGAATCCAT CCGAATGGCC 924 R306*
 ATGGGGCTCC GAATCAAGTT CCCCACGGTG GTGGCAGGGT TTGACCTGGT 974 M309I L311R R312ter T317M G321E G321A delEx7 Dup_Exon7
GGGGCATGAG GACACTGGCC ACTCCTTGCA TGACTACAAG GAAGCTCTGA 1024 G326V E328K E328D D329N H335R
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 TTCATGGGCA TTGGGGGGAT GAAGGCTGAC CTGAGGACCC TCAAACAGCT 1424 p.(M460K) M465fsX K466Tfs*2
 GGCCATGAAC TCTATCAAGT ACAGTACCCT GTTGGAGAGT GAGAAAAATA 1474 S479P Y482C S483Pfs* E489Q E489D
 CTTTCATGGA AATCTGGAAG AAGAGATGGG ATAAGTTCAT AGCAGATGTG 1524 W501*
 GCTACAAAGT GAGGAGAAGC TAGCCAGCCC TCTACAAGCT GTCTTCTTGC *38
 ACACGCTGTC ACTTCTCTC ACTCGTCTT GAATCAGCTC CATGTGCCCA *88
 TGAAATCAAT GGCCCTGTGA TGGAGCGACC CTGTGAGAAG CACTTGGCTG *138
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 TGCATCAGCC TCGCGAGTAG TTGGAATTAT AGGCATGTGC CACCACACCC *488
 AGCTGATTTT TGCATTTTTA GTAGAGACAG GTTTTCACCA TGTGGCGAG *538
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 TCTTTAACTC TCTCCTCTT ATTTCTCTT TCTCTCTGTA CACTTTTCCT *688
 GGGTGGTCTC ATCCATTCCT TTGCTTTTTT ATACCATTTA TTTGTTAATG *738

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GTCTCAGAGA GGTTCCTCTG TGCTCGCCCT GTTTCTCTCA GGAAGCCTTG *1988
CTCTTTTCCA TCATGCCTCT AATCACAGCT TATAATCGGA TATTTATTTT *2038
TGTGCTTACA GTCTTGCCCT GCCAGACTGT ATGCCCATG TGGGCAGGCG *2088
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CTTGC

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