

Variant: NM_000022.4(ADA):c.402C>T (p.Gly134=)

Version: 1.0

CA9871679 [↗](#)

536186 (ClinVar) [↗](#)

Gene: ADA ([HGNC:100](#))
Condition: adenosine deaminase deficiency ([MONDO:0007064](#))
Inheritance Mode: Autosomal recessive inheritance
UUID: 2cc1057b-9ac7-4d27-a0c7-0eab74eb6052
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HGVS expressions

NM_000022.4:c.402C>T
NM_000022.4(ADA):c.402C>T (p.Gly134=)
NC_000020.11:g.44625645G>A
CM000682.2:g.44625645G>A
NC_000020.10:g.43254286G>A
CM000682.1:g.43254286G>A
NC_000020.9:g.42687700G>A
NG_007385.1:g.31091C>T
ENST00000492931.6:n.493C>T
ENST00000536076.2:c.249C>T
ENST00000536532.6:c.402C>T
ENST00000537820.2:c.402C>T
ENST00000539235.6:c.219-2567C>T
ENST00000695889.1:c.219-2715C>T
ENST00000695890.1:n.2205C>T
ENST00000695891.1:c.219-2715C>T
ENST00000695927.1:c.480C>T
ENST00000695949.1:c.399C>T
ENST00000695957.1:c.362+811C>T
ENST00000695991.1:c.217-2715C>T
ENST00000695992.1:c.402C>T
ENST00000695993.1:c.402C>T
ENST00000695994.1:c.402C>T
ENST00000695995.1:c.217-2567C>T
ENST00000695996.1:n.473C>T
ENST00000695997.1:n.433+811C>T
ENST00000696003.1:n.494C>T
ENST00000696004.1:n.494C>T
ENST00000696006.1:c.402C>T
ENST00000696007.1:c.329+811C>T
ENST00000696008.1:n.318C>T
ENST00000696009.1:n.513C>T
ENST00000696017.1:c.399C>T
ENST00000696034.1:c.402C>T
ENST00000696035.1:n.512C>T
ENST00000696036.1:n.1092C>T
ENST00000696037.1:n.2079C>T

	ENST00000696038.1:c.*148C>T ENST00000696039.1:n.690C>T ENST00000696058.1:c.402C>T ENST00000696059.1:c.*347C>T ENST00000696060.1:c.402C>T ENST00000696061.1:c.399C>T ENST00000696062.1:c.465C>T ENST00000696063.1:c.477C>T ENST00000696064.1:c.249C>T ENST00000696065.1:c.66-2715C>T ENST00000696075.1:c.*372C>T ENST00000696076.1:c.402C>T ENST00000696077.1:c.399C>T ENST00000696078.1:c.402C>T ENST00000696079.1:c.402C>T ENST00000696080.1:c.402C>T ENST00000696082.1:c.480C>T ENST00000696083.1:n.44C>T ENST00000696084.1:n.503C>T ENST00000696104.1:c.362+811C>T ENST00000696105.1:c.402C>T ENST00000372874.9:c.402C>T ENST00000372874.8:c.402C>T ENST00000464097.5:n.76C>T ENST00000492931.5:n.486C>T ENST00000536532.5:c.402C>T ENST00000537820.1:c.402C>T ENST00000539235.5:c.219-2567C>T NM_000022.2:c.402C>T NM_000022.3:c.402C>T NM_001322050.1:c.73+811C>T NM_001322051.1:c.402C>T NR_136160.1:n.553C>T NM_001322050.2:c.73+811C>T NM_001322051.2:c.402C>T NR_136160.2:n.494C>T
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Likely Benign

Met criteria codes 3

BS1

BS2_Supporting

BP7

Evidence Links 0

Expert Panel

Severe Combined Immunodeficiency Disease VCEP

Criteria Specification Information

Criteria Specification: ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ADA Version 1.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Severe Combined Immunodeficiency Disease VCEP

The c.402C>T (p.Gly134=) variant (NM_000022.4) is a synonymous (silent) variant that is not predicted by SpliceAI and varSEAK to impact splicing (BP7). The highest population minor allele frequency in gnomAD v2.1.1 is 0.003095 (89/19874 alleles) in African/African American population, which is higher than the ClinGen SCID VCEP threshold (>0.00161) for BS1, and therefore meets this criterion (BS1). In the gnomAD v2.1.1 database, this variant has been observed in 1 homozygous individual with no features of SCID, a condition with full penetrance at an early age. BS2_supporting is met. In summary, this variant is classified as a Likely Benign for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP (specification version 1.0): BS1, BP7, and BS2_Supporting.

Met criteria codes

BS1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.003095 (89/19874 alleles) in African/African American population, which is higher than the ClinGen SCID VCEP threshold (>0.00161) for BS1, and therefore meets this criterion (BS1).
BS2_Supporting			In the gnomAD v2.1.1 database, this variant has been observed in 1 homozygous individual with no features of SCID, a condition with full penetrance at an early age. BS2_supporting is met.
BP7			The c.402C>T (p.Gly134=) variant (NM_000022.4) is a synonymous (silent) variant that is not predicted by SpliceAI and varSEAK to impact splicing (BP7).

Curation History



 

 

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