

Variant: *NM_000022.4(ADA):c.192G>A (p.Lys64=)*

Version: 1.0

CA9871743 [↗](#)

719476 (ClinVar) [↗](#)

Gene: ADA ([HGNC:100](#))

Condition: adenosine deaminase deficiency ([MONDO:0007064](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 7ff99571-d16b-4165-bb4e-8b98ebbd25f0

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HGVS expressions

NM_000022.4:c.192G>A

NM_000022.4(ADA):c.192G>A (p.Lys64=)

NC_000020.11:g.44629073C>T

CM000682.2:g.44629073C>T

NC_000020.10:g.43257714C>T

CM000682.1:g.43257714C>T

NC_000020.9:g.42691128C>T

NG_007385.1:g.27663G>A

ENST00000492931.6:n.283G>A

ENST00000536076.2:c.39G>A

ENST00000536532.6:c.192G>A

ENST00000537820.2:c.192G>A

ENST00000539235.6:c.192G>A

ENST00000695889.1:c.192G>A

ENST00000695891.1:c.192G>A

ENST00000695927.1:c.270G>A

ENST00000695949.1:c.189G>A

ENST00000695957.1:c.192G>A

ENST00000695991.1:c.192G>A

ENST00000695992.1:c.192G>A

ENST00000695993.1:c.192G>A

ENST00000695994.1:c.192G>A

ENST00000695995.1:c.192G>A

ENST00000695996.1:n.263G>A

ENST00000695997.1:n.263G>A

ENST00000696003.1:n.284G>A

ENST00000696004.1:n.284G>A

ENST00000696006.1:c.192G>A

ENST00000696007.1:c.159G>A

ENST00000696009.1:n.303G>A

ENST00000696010.1:n.305G>A

ENST00000696017.1:c.189G>A

ENST00000696034.1:c.192G>A

ENST00000696038.1:c.192G>A

ENST00000696039.1:n.480G>A

ENST00000696058.1:c.192G>A

ENST00000696059.1:c.*137G>A

	ENST00000696060.1:c.192G>A ENST00000696061.1:c.189G>A ENST00000696062.1:c.255G>A ENST00000696063.1:c.267G>A ENST00000696064.1:c.39G>A ENST00000696065.1:c.39G>A ENST00000696075.1:c.*162G>A ENST00000696076.1:c.192G>A ENST00000696077.1:c.189G>A ENST00000696078.1:c.192G>A ENST00000696079.1:c.192G>A ENST00000696080.1:c.192G>A ENST00000696082.1:c.270G>A ENST00000696084.1:n.293G>A ENST00000696104.1:c.192G>A ENST00000696105.1:c.192G>A ENST00000372874.9:c.192G>A ENST00000372874.8:c.192G>A ENST00000492931.5:n.276G>A ENST00000536076.1:n.372G>A ENST00000536532.5:c.192G>A ENST00000537820.1:c.192G>A ENST00000539235.5:c.192G>A ENST00000545776.5:n.246G>A NM_000022.2:c.192G>A NM_000022.3:c.192G>A NM_001322050.1:c.-98G>A NM_001322051.1:c.192G>A NR_136160.1:n.343G>A NM_001322050.2:c.-98G>A NM_001322051.2:c.192G>A NR_136160.2:n.284G>A
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Likely Benign	Met criteria codes 2 BS1BP7 Not Met criteria codes 1 BS2 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
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Evidence submitted by expert panel
Severe Combined Immunodeficiency Disease VCEP

The c.192G>A (p.Lys64=) variant (NM_000022.4) in ADA 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 0.002796 (67/19946 alleles) in the East Asian population, which is higher than the ClinGen SCID VCEP threshold (>0.00161) for BS1 and therefore meets this criterion (BS1). In summary, this variant is classified as Likely Benign for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP (specification version 1.0): BP7 and BS1.

Met criteria codes

BS1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.002796 (67/19946 alleles) in East Asian population, which is higher than the ClinGen SCID VCEP threshold (>0.00161) for BS1, and therefore meets this criterion (BS1).
BP7			The c.192G>A (p.Lys64=) variant (NM_000022.4) in ADA is a synonymous (silent) variant that is not predicted by SpliceAI and varSEAK to impact splicing (BP7).

Not Met criteria codes

BS2			No homozygous has been described in gnomAD v2.1.1.
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Curation History



 

 

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