

Variant: NM_000022.2(ADA):c.22G>A (p.Asp8Asn)

Version: 1.2

CA115289 [↗](#)

1973 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 89074206-c4a1-4f51-9071-c651840148ce

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HGVS expressions

NM_000022.2(ADA):c.22G>A (p.Asp8Asn)

NC_000020.11:g.44651586C>T
CM000682.2:g.44651586C>T
NC_000020.10:g.43280227C>T
CM000682.1:g.43280227C>T
NC_000020.9:g.42713641C>T
NG_007385.1:g.5150G>A
ENST00000492931.6:n.113G>A
ENST00000536076.2:c.-121+316G>A
ENST00000536532.6:c.22G>A
ENST00000537820.2:c.22G>A
ENST00000539235.6:c.22G>A
ENST00000695889.1:c.22G>A
ENST00000695891.1:c.22G>A
ENST00000695949.1:c.22G>A
ENST00000695957.1:c.22G>A
ENST00000695991.1:c.22G>A
ENST00000695992.1:c.22G>A
ENST00000695993.1:c.22G>A
ENST00000695994.1:c.22G>A
ENST00000695995.1:c.22G>A
ENST00000695996.1:n.93G>A
ENST00000695997.1:n.93G>A
ENST00000696003.1:n.114G>A
ENST00000696004.1:n.114G>A
ENST00000696006.1:c.22G>A
ENST00000696009.1:n.133G>A
ENST00000696010.1:n.135G>A
ENST00000696017.1:c.22G>A
ENST00000696034.1:c.22G>A
ENST00000696038.1:c.22G>A
ENST00000696039.1:n.321+316G>A
ENST00000696058.1:c.22G>A
ENST00000696059.1:c.22G>A
ENST00000696060.1:c.22G>A
ENST00000696061.1:c.22G>A
ENST00000696062.1:c.96+514G>A

	ENST00000696063.1:c.22G>A ENST00000696064.1:c.-118+316G>A ENST00000696065.1:c.-121+316G>A ENST00000696075.1:c.22G>A ENST00000696076.1:c.22G>A ENST00000696077.1:c.22G>A ENST00000696078.1:c.22G>A ENST00000696079.1:c.22G>A ENST00000696080.1:c.22G>A ENST00000696084.1:n.123G>A ENST00000696104.1:c.22G>A ENST00000696105.1:c.22G>A ENST00000372874.9:c.22G>A ENST00000372874.8:c.22G>A ENST00000492931.5:n.106G>A ENST00000535573.1:n.332+316G>A ENST00000536076.1:n.213+316G>A ENST00000536532.5:c.22G>A ENST00000537820.1:c.22G>A ENST00000539235.5:c.22G>A ENST00000545776.5:n.76G>A NM_000022.2:c.22G>A NM_000022.3:c.22G>A NM_001322050.1:c.-268G>A NM_001322051.1:c.22G>A NR_136160.1:n.173G>A NM_000022.4:c.22G>A NM_001322050.2:c.-268G>A NM_001322051.2:c.22G>A NR_136160.2:n.114G>A
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Benign	Met criteria codes 2 BS2_SupportingBA1 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 NM_000022.2:c.22G>A variant in ADA is a missense variant predicted to cause substitution of aspartic acid by asparagine at amino acid 8 (p.Asp8Asn). This variant is present at an overall minor allele frequency of 0.05863 (10050/171418 alleles) with the highest population minor allele frequency of 0.1300 (3033/23322) in the South Asian population in gnomAD v2.1.1. This frequency is higher than

the ClinGen SCID VCEP's threshold for BA1 (>0.00721); therefore, BA1 is met. This variant has been observed in 434 homozygous individuals in gnomAD, a condition with full penetrance at an early age (BS2_Supporting). In summary, this variant meets the criteria to be classified as benign for SCID. ACMG/AMP criteria applied, as specified by the ClinGen SCID-VCEP: BA1 and BS2_Supporting (SCID VCEP specifications version 1.0).

Met criteria codes

BS2_Supporting			This variant has been observed in 434 homozygous individuals in gnomAD, a condition with full penetrance at an early age (BS2_Supporting).
BA1			Overall frequency in gnomAD is 0.05863 (10050/171418 alleles) and highest frequency is 0.1300 (3033/23322) in South Asian population. https://gnomad.broadinstitute.org/variant/20-43280227-C-T This is higher than the cutoff set for BA1 in the SCID VCEP's ADA specifications (gnomAD popmax filtering allele frequency > 0.00721. There are also 434 homozygotes in gnomAD. Therefore, BA1 applies.

Curation History

  

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