

Variant: NM_000022.4(ADA):c.596A>G (p.Gln199Arg)

Version: 1.0

CA409120756 [↗](#)

2050660 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8afddec5-1eb1-499b-9e91-253b4153875b

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HGVS expressions

NM_000022.4(ADA):c.596A>G (p.Gln199Arg)

NM_000022.4:c.596A>G

NC_000020.11:g.44624212T>C

CM000682.2:g.44624212T>C

NC_000020.10:g.43252853T>C

CM000682.1:g.43252853T>C

NC_000020.9:g.42686267T>C

NG_007385.1:g.32524A>G

ENST00000492931.6:n.687A>G

ENST00000536076.2:c.443A>G

ENST00000536532.6:c.596A>G

ENST00000537820.2:c.596A>G

ENST00000539235.6:c.219-1134A>G

ENST00000695889.1:c.219-1282A>G

ENST00000695890.1:n.2399A>G

ENST00000695891.1:c.219-1282A>G

ENST00000695927.1:c.674A>G

ENST00000695949.1:c.593A>G

ENST00000695957.1:c.*87A>G

ENST00000695991.1:c.217-1282A>G

ENST00000695992.1:c.596A>G

ENST00000695993.1:c.596A>G

ENST00000695994.1:c.596A>G

ENST00000695995.1:c.217-1134A>G

ENST00000695996.1:n.667A>G

ENST00000695997.1:n.551A>G

ENST00000696003.1:n.688A>G

ENST00000696004.1:n.688A>G

ENST00000696005.1:c.118A>G

ENST00000696006.1:c.596A>G

ENST00000696007.1:c.447A>G

ENST00000696008.1:n.1751A>G

ENST00000696009.1:n.1946A>G

ENST00000696017.1:c.593A>G

ENST00000696034.1:c.596A>G

ENST00000696035.1:n.706A>G

ENST00000696036.1:n.1286A>G

	ENST00000696037.1:n.2273A>G ENST00000696038.1:c.*342A>G ENST00000696039.1:n.884A>G ENST00000696058.1:c.596A>G ENST00000696059.1:c.*541A>G ENST00000696060.1:c.596A>G ENST00000696061.1:c.593A>G ENST00000696062.1:c.659A>G ENST00000696063.1:c.671A>G ENST00000696064.1:c.443A>G ENST00000696065.1:c.66-1282A>G ENST00000696074.1:n.212A>G ENST00000696075.1:c.*566A>G ENST00000696076.1:c.596A>G ENST00000696077.1:c.593A>G ENST00000696078.1:c.596A>G ENST00000696079.1:c.596A>G ENST00000696080.1:c.596A>G ENST00000696081.1:n.715A>G ENST00000696082.1:c.674A>G ENST00000696083.1:n.1477A>G ENST00000696084.1:n.697A>G ENST00000696104.1:c.363-1282A>G ENST00000696105.1:c.*137A>G ENST00000372874.9:c.596A>G ENST00000372874.8:c.596A>G ENST00000372887.5:c.*236T>C ENST00000464097.5:n.270A>G ENST00000492931.5:n.680A>G ENST00000536532.5:c.596A>G ENST00000537820.1:c.596A>G ENST00000539235.5:c.219-1134A>G NM_000022.2:c.596A>G NM_000022.3:c.596A>G NM_001322050.1:c.191A>G NM_001322051.1:c.596A>G NR_136160.1:n.747A>G NM_001322050.2:c.191A>G NM_001322051.2:c.596A>G NR_136160.2:n.688A>G
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Uncertain Significance

Met criteria codes 1

PM2_Supporting

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

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

NM_000022.4:c.596A>G is a missense variant predicted to cause substitution of Glutamine by Arginine at amino acid 199 (p.Gln199Arg). The highest population minor allele frequency in gnomAD v4 is 0.00001440 (1/69426 alleles) in South Asian population, which is lower than the ClinGen SCID VCEP threshold (<0.0001742) for PM2_Supporting, meeting this criterion (PM2_Supporting). There are no publications for this variant in the literature. Based on insufficient evidence, this variant may be classified as variant of uncertain significance for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP(specification version 1.0): PM2_supporting.

Met criteria codes

PM2_Supporting			The highest population minor allele frequency in gnomAD v4 is 0.00001440 (1/69426 alleles) in South Asian population, which is lower than the ClinGen SCID VCEP threshold (<0.0001742) for PM2_Supporting, meeting this criterion (PM2_Supporting).
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Curation History [↗](#)



 

 

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