The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000022.4(ADA):c.201C>G (p.Tyr67Ter)

Version: 1.0

CA409121739

1075328 (ClinVar)

Gene: ADA (HGNC:100)
Condition: adenosine deaminase deficiency (MONDO:0007064)
Inheritance Mode: Autosomal recessive inheritance
UUID: 22a9fb83-de28-42e2-9a6a-8823273ab3b6
Approved on: 2024/1/16
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HGVS expressions

NM_000022.4(ADA):c.201C>G (p.Tyr67Ter)
NM_000022.4:c.201C>G
NC_000020.11:g.44629064G>C
CM000682.2:g.44629064G>C
NC_000020.10:g.43257705G>C
CM000682.1:g.43257705G>C
NC_000020.9:g.42691119G>C
NG_007385.1:g.27672C>G
ENST00000492931.6:n.292C>G
ENST00000536076.2:c.48C>G
ENST00000536532.6:c.201C>G
ENST00000537820.2:c.201C>G
ENST00000539235.6:c.201C>G
ENST00000695889.1:c.201C>G
ENST00000695891.1:c.201C>G
ENST00000695927.1:c.279C>G
ENST00000695949.1:c.198C>G
ENST00000695957.1:c.201C>G
ENST00000695991.1:c.201C>G
ENST00000695992.1:c.201C>G
ENST00000695993.1:c.201C>G
ENST00000695994.1:c.201C>G
ENST00000695995.1:c.201C>G
ENST00000695996.1:n.272C>G
ENST00000695997.1:n.272C>G
ENST00000696003.1:n.293C>G
ENST00000696004.1:n.293C>G
ENST00000696006.1:c.201C>G
ENST00000696007.1:c.168C>G
ENST00000696009.1:n.312C>G
ENST00000696010.1:n.314C>G
ENST00000696017.1:c.198C>G
ENST00000696034.1:c.201C>G
ENST00000696038.1:c.201C>G
ENST00000696039.1:n.489C>G
ENST00000696058.1:c.201C>G
ENST00000696059.1:c.*146C>G

ENST00000696060.1:c.201C>G
ENST00000696061.1:c.198C>G
ENST00000696062.1:c.264C>G
ENST00000696063.1:c.276C>G
ENST00000696064.1:c.48C>G
ENST00000696065.1:c.48C>G
ENST00000696075.1:c.*171C>G
ENST00000696076.1:c.201C>G
ENST00000696077.1:c.198C>G
ENST00000696078.1:c.201C>G
ENST00000696079.1:c.201C>G
ENST00000696080.1:c.201C>G
ENST00000696082.1:c.279C>G
ENST00000696084.1:n.302C>G
ENST00000696104.1:c.201C>G
ENST00000696105.1:c.201C>G
ENST00000372874.9:c.201C>G
ENST00000372874.8:c.201C>G
ENST00000492931.5:n.285C>G
ENST00000536076.1:n.381C>G
ENST00000536532.5:c.201C>G
ENST00000537820.1:c.201C>G
ENST00000539235.5:c.201C>G
ENST00000545776.5:n.255C>G
NM_000022.2:c.201C>G
NM_000022.3:c.201C>G
NM_001322050.1:c.-89C>G
NM_001322051.1:c.201C>G
NR_136160.1:n.352C>G
NM_001322050.2:c.-89C>G
NM_001322051.2:c.201C>G
NR_136160.2:n.293C>G

Likely Pathogenic

Met criteria codes 2

PM2_Supporting PVS1

Not Met criteria codes 1

PP4

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**

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The NM_000022.4(ADA):c.201C>G (p.Tyr67Ter) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 3/12 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). This variant has been reported in ClinVar without patient information; however, it has not been reported in the literature to our knowledge. In summary, this variant meets the criteria to be classified as Likely Pathogenic for SCID. ACMG/AMP criteria applied, as specified by the ClinGen SCID-VCEP: PVS1 and PM2_Supporting (VCEP specifications version 1).

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1	✔	The c.201C>G (p.Tyr67Ter)(NM_000022.4) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 3/12 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).

Not Met criteria codes

PP4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 1 of 1 rows

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