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Variant: NM_000022.4(ADA):c.511A>T (p.Lys171Ter)

Version: 1.0

CA409120933

983782 (ClinVar)

Gene: ADA (HGNC:100)
Condition: adenosine deaminase deficiency (MONDO:0007064)
Inheritance Mode: Autosomal recessive inheritance
UUID: d06c2b91-9539-41e6-a028-f22d6f42d49a
Approved on: 2024/5/28
Published on: 2024/5/28

HGVS expressions

NM_000022.4(ADA):c.511A>T (p.Lys171Ter)
NM_000022.4:c.511A>T
NC_000020.11:g.44624297T>A
CM000682.2:g.44624297T>A
NC_000020.10:g.43252938T>A
CM000682.1:g.43252938T>A
NC_000020.9:g.42686352T>A
NG_007385.1:g.32439A>T
ENST00000492931.6:n.602A>T
ENST00000536076.2:c.358A>T
ENST00000536532.6:c.511A>T
ENST00000537820.2:c.511A>T
ENST00000539235.6:c.219-1219A>T
ENST00000695889.1:c.219-1367A>T
ENST00000695890.1:n.2314A>T
ENST00000695891.1:c.219-1367A>T
ENST00000695927.1:c.589A>T
ENST00000695949.1:c.508A>T
ENST00000695957.1:c.*2A>T
ENST00000695991.1:c.217-1367A>T
ENST00000695992.1:c.511A>T
ENST00000695993.1:c.511A>T
ENST00000695994.1:c.511A>T
ENST00000695995.1:c.217-1219A>T
ENST00000695996.1:n.582A>T
ENST00000695997.1:n.466A>T
ENST00000696003.1:n.603A>T
ENST00000696004.1:n.603A>T
ENST00000696005.1:c.33A>T
ENST00000696006.1:c.511A>T
ENST00000696007.1:c.362A>T
ENST00000696008.1:n.1666A>T
ENST00000696009.1:n.1861A>T
ENST00000696017.1:c.508A>T
ENST00000696034.1:c.511A>T
ENST00000696035.1:n.621A>T
ENST00000696036.1:n.1201A>T

ENST00000696037.1:n.2188A>T
ENST00000696038.1:c.*257A>T
ENST00000696039.1:n.799A>T
ENST00000696058.1:c.511A>T
ENST00000696059.1:c.*456A>T
ENST00000696060.1:c.511A>T
ENST00000696061.1:c.508A>T
ENST00000696062.1:c.574A>T
ENST00000696063.1:c.586A>T
ENST00000696064.1:c.358A>T
ENST00000696065.1:c.66-1367A>T
ENST00000696074.1:n.127A>T
ENST00000696075.1:c.*481A>T
ENST00000696076.1:c.511A>T
ENST00000696077.1:c.508A>T
ENST00000696078.1:c.511A>T
ENST00000696079.1:c.511A>T
ENST00000696080.1:c.511A>T
ENST00000696081.1:n.630A>T
ENST00000696082.1:c.589A>T
ENST00000696083.1:n.1392A>T
ENST00000696084.1:n.612A>T
ENST00000696104.1:c.363-1367A>T
ENST00000696105.1:c.*52A>T
ENST00000372874.9:c.511A>T
ENST00000372874.8:c.511A>T
ENST00000464097.5:n.185A>T
ENST00000492931.5:n.595A>T
ENST00000536532.5:c.511A>T
ENST00000537820.1:c.511A>T
ENST00000539235.5:c.219-1219A>T
NM_000022.2:c.511A>T
NM_000022.3:c.511A>T
NM_001322050.1:c.106A>T
NM_001322051.1:c.511A>T
NR_136160.1:n.662A>T
NM_001322050.2:c.106A>T
NM_001322051.2:c.511A>T
NR_136160.2:n.603A>T

Likely Pathogenic

Met criteria codes 2

PM2_Supporting PVS1

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 c.511A>T (p.Lys171Ter) (NM_000022.4) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 6/12 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1_Met). The variant is absent in gnomAD v4 (PM2_supporting). There are no publications for this variant in the literature. As per the SCID VCEP specifications and the Bayesian interpretation of the ACMG/AMP combining rules, 1 very strong and 1 supporting criteria results in a Likely Pathogenic classification. In summary, this variant meets the criteria to be classified as a Likely Pathogenic variant for autosomal recessive severe combined immunodeficiency due to ADA deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP (VCEP specifications version 1): PVS1,PM2_supporting.

Met criteria codes

PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).
PVS1	✔	The c.511A>T (p.Lys171Ter) (NM_000022.4) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 6/12 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).

Curation History

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