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Variant: NM_000022.4(ADA):c.736C>T (p.Gln246Ter)

Version: 1.0

CA409120239

549915 (ClinVar)

Gene: ADA (HGNC:100)
Condition: adenosine deaminase deficiency (MONDO:0007064)
Inheritance Mode: Autosomal recessive inheritance
UUID: ea0a6057-1873-4413-9acd-7ccbc79ce6fa
Approved on: 2024/1/24
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HGVS expressions

NM_000022.4(ADA):c.736C>T (p.Gln246Ter)
NM_000022.4:c.736C>T
NC_000020.11:g.44622873G>A
CM000682.2:g.44622873G>A
NC_000020.10:g.43251514G>A
CM000682.1:g.43251514G>A
NC_000020.9:g.42684928G>A
NG_007385.1:g.33863C>T
ENST00000492931.6:n.903C>T
ENST00000536076.2:c.583C>T
ENST00000536532.6:c.736C>T
ENST00000537820.2:c.664C>T
ENST00000539235.6:c.*120C>T
ENST00000695889.1:c.276C>T
ENST00000695890.1:n.2615C>T
ENST00000695891.1:c.276C>T
ENST00000695927.1:c.814C>T
ENST00000695949.1:c.661C>T
ENST00000695957.1:c.*227C>T
ENST00000695991.1:c.274C>T
ENST00000695992.1:c.736C>T
ENST00000695993.1:c.736C>T
ENST00000695994.1:c.709C>T
ENST00000695995.1:c.346C>T
ENST00000695996.1:n.883C>T
ENST00000696003.1:n.904C>T
ENST00000696004.1:n.904C>T
ENST00000696005.1:c.186C>T
ENST00000696006.1:c.664C>T
ENST00000696007.1:c.663C>T
ENST00000696008.1:n.3090C>T
ENST00000696017.1:c.733C>T
ENST00000696034.1:c.736C>T
ENST00000696035.1:n.922C>T
ENST00000696036.1:n.1502C>T
ENST00000696037.1:n.2413C>T
ENST00000696038.1:c.*558C>T

ENST00000696039.1:n.1100C>T
ENST00000696058.1:c.733C>T
ENST00000696059.1:c.*681C>T
ENST00000696060.1:c.805C>T
ENST00000696061.1:c.733C>T
ENST00000696062.1:c.799C>T
ENST00000696063.1:c.811C>T
ENST00000696064.1:c.583C>T
ENST00000696065.1:c.123C>T
ENST00000696073.1:n.1047C>T
ENST00000696074.1:n.352C>T
ENST00000696075.1:c.*706C>T
ENST00000696076.1:c.805C>T
ENST00000696077.1:c.730C>T
ENST00000696078.1:c.733C>T
ENST00000696079.1:c.733C>T
ENST00000696080.1:c.736C>T
ENST00000696081.1:n.855C>T
ENST00000696082.1:c.811C>T
ENST00000696083.1:n.1693C>T
ENST00000696084.1:n.913C>T
ENST00000696104.1:c.420C>T
ENST00000372874.9:c.736C>T
ENST00000372874.8:c.736C>T
ENST00000372887.5:c.152-1060G>A
ENST00000464097.5:n.486C>T
ENST00000492931.5:n.896C>T
ENST00000536532.5:c.736C>T
ENST00000537820.1:c.664C>T
ENST00000539235.5:c.*120C>T
NM_000022.2:c.736C>T
NM_000022.3:c.736C>T
NM_001322050.1:c.331C>T
NM_001322051.1:c.664C>T
NR_136160.1:n.887C>T
NM_001322050.2:c.331C>T
NM_001322051.2:c.664C>T
NR_136160.2:n.828C>T

Pathogenic

Met criteria codes 3

PVS1 PM3_Supporting 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

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The c.736C>T (p.Gln246Ter)(NM_000022.4) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 8/12 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). One patient (U444) was found homozygous for this mutation (PMID: 17185467,PM3_Supporting).Based on the above evidence, this variant can be classified as pathogenic for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP(specification version 1.0): PVS1_Met,PM2_Supporting,PM3_Supporting.

Met criteria codes

PVS1	✔	The c.736C>T (p.Gln246Ter)(NM_000022.4) variant in ADA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 8/12 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1_met).
PM3_Supporting	✔	One patient (U444) was found homozygous for this mutation (PMID: 17185467,PM3_Supporting)
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_Supporting)

Curation History

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