

Variant: NM_000022.4(ADA):c.239A>G (p.Lys80Arg)

Version: 1.0

CA251993 [↗](#)

1954 (ClinVar) [↗](#)

Gene: ADA ([HGNC:100](#))
Condition: adenosine deaminase deficiency ([MONDO:0007064](#))
Inheritance Mode: Autosomal recessive inheritance
UUID: 5adcbc29-0bdb-463d-ae7a-c6fb29411314
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HGVS expressions

NM_000022.4:c.239A>G
NM_000022.4(ADA):c.239A>G (p.Lys80Arg)
NC_000020.11:g.44626579T>C
CM000682.2:g.44626579T>C
NC_000020.10:g.43255220T>C
CM000682.1:g.43255220T>C
NC_000020.9:g.42688634T>C
NG_007385.1:g.30157A>G
ENST00000492931.6:n.330A>G
ENST00000536076.2:c.86A>G
ENST00000536532.6:c.239A>G
ENST00000537820.2:c.239A>G
ENST00000539235.6:c.218+2468A>G
ENST00000695889.1:c.218+2468A>G
ENST00000695890.1:n.2042A>G
ENST00000695891.1:c.218+2468A>G
ENST00000695927.1:c.317A>G
ENST00000695949.1:c.236A>G
ENST00000695957.1:c.239A>G
ENST00000695991.1:c.216+2470A>G
ENST00000695992.1:c.239A>G
ENST00000695993.1:c.239A>G
ENST00000695994.1:c.239A>G
ENST00000695995.1:c.216+2470A>G
ENST00000695996.1:n.310A>G
ENST00000695997.1:n.310A>G
ENST00000696003.1:n.331A>G
ENST00000696004.1:n.331A>G
ENST00000696006.1:c.239A>G
ENST00000696007.1:c.206A>G
ENST00000696009.1:n.350A>G
ENST00000696017.1:c.236A>G
ENST00000696034.1:c.239A>G
ENST00000696035.1:n.349A>G
ENST00000696036.1:n.929A>G
ENST00000696037.1:n.1916A>G
ENST00000696038.1:c.237A>G

	ENST00000696039.1:n.527A>G ENST00000696058.1:c.239A>G ENST00000696059.1:c.*184A>G ENST00000696060.1:c.239A>G ENST00000696061.1:c.236A>G ENST00000696062.1:c.302A>G ENST00000696063.1:c.314A>G ENST00000696064.1:c.86A>G ENST00000696065.1:c.65+2468A>G ENST00000696075.1:c.*209A>G ENST00000696076.1:c.239A>G ENST00000696077.1:c.236A>G ENST00000696078.1:c.239A>G ENST00000696079.1:c.239A>G ENST00000696080.1:c.239A>G ENST00000696082.1:c.317A>G ENST00000696084.1:n.340A>G ENST00000696104.1:c.239A>G ENST00000696105.1:c.239A>G ENST00000372874.9:c.239A>G ENST00000372874.8:c.239A>G ENST00000492931.5:n.323A>G ENST00000536076.1:n.419A>G ENST00000536532.5:c.239A>G ENST00000537820.1:c.239A>G ENST00000539235.5:c.218+2468A>G ENST00000545776.5:n.293A>G NM_000022.2:c.239A>G NM_000022.3:c.239A>G NM_001322051.1:c.239A>G NM_001322050.2:c.-51A>G NM_001322051.2:c.239A>G NR_136160.2:n.331A>G
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Benign	Met criteria codes 2 BA1BS2_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
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Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The NM_000022.4:c.239A>G variant in ADA is a missense variant predicted to cause substitution of lysine by arginine at amino acid 80 (p.Lys80Arg). The Popmax filtering allele frequency of this variant in the gnomAD v2.1.1 database is 0.06430, which is higher than the ClinGen SCID VCEP threshold (>0.00721) for BA1 (BA1). This variant has been observed in 551 homozygous individuals with no feature of SCID, 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. (VCEP specifications version 1).

Met criteria codes

BA1			The Popmax filtering allele frequency of this variant in the gnomAD v2.1.1 database is 0.06430, which is higher than the ClinGen SCID VCEP threshold (>0.00721) for BA1. Therefore BA1 is met.
BS2_Supporting			In the gnomAD v2.1.1 database, this variant has been observed in 551 homozygous individuals with no feature of SCID, a condition with full penetrance at an early age. BS2_Supporting is met.

Curation History



 

 

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