

Variant: NM_000448.3(RAG1):c.1346G>A (p.Arg449Lys)

Version: 1.0

CA5950130 [↗](#)

256188 (ClinVar) [↗](#)

Gene: RAG1 ([HGNC:5896](#))

Condition: recombinaase activating gene 1 deficiency ([MONDO:0000572](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 9a1a1479-2183-4e3d-bff7-3898c6865d3a

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HGVS expressions

NM_000448.3:c.1346G>A

NM_000448.3(RAG1):c.1346G>A (p.Arg449Lys)

NC_000011.10:g.36574650G>A

CM000673.2:g.36574650G>A

NC_000011.9:g.36596200G>A

CM000673.1:g.36596200G>A

NC_000011.8:g.36552776G>A

NG_007528.1:g.11638G>A

ENST00000697713.1:c.1346G>A

ENST00000697714.1:c.1346G>A

ENST00000697715.1:c.1346G>A

ENST00000299440.6:c.1346G>A

ENST00000299440.5:c.1346G>A

ENST00000534663.1:c.1346G>A

NM_000448.2:c.1346G>A

NM_001377277.1:c.1346G>A

NM_001377278.1:c.1346G>A

NM_001377279.1:c.1346G>A

NM_001377280.1:c.1346G>A

Benign

Met criteria codes **2**

BS2_Supporting

BA1

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 RAG1 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_000448.3:c.1346G>A variant in RAG1 is a missense variant predicted to cause the substitution of Arginine by Lysine at amino acid 449 (p.Arg449Lys). The filtering allele frequency (the lower threshold of the 95% CI of 21803/1180030) of the c.1346G>A variant in RAG1 is 0.01837 for European (non-Finnish) chromosomes by gnomAD v.4, which is higher than the ClinGen SCID VCEP threshold (>0.00872) for BA1 and, therefore, meets this criterion (BA1). Additionally, 207 homozygous adults are reported on gnomAD v.4 (BS2_Supporting). In summary, this variant meets the criteria to be classified as Benign for autosomal recessive SCID. ACMG/AMP criteria applied, as specified by the ClinGen SCID-VCEP: BA1 and BS2_Supporting. (VCEP specifications version 1).

Met criteria codes

BS2_Supporting			207 homozygous adults are reported on GnomAD v.4, BS2_Supporting is Met.
BA1			The filtering allele frequency (the lower threshold of the 95% CI of 21803/1180030) of the c.1346G>A variant in RAG1 is 0.01837 for European (non-Finnish) chromosomes by gnomAD v.4, which is higher than the ClinGen SCID VCEP threshold (>0.00872) for BA1, and therefore meets this criterion (BA1).

Curation History



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