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_000448.3(RAG1):c.2459A>G (p.Lys820Arg)

Version: 1.0

CA5950267

256189 (ClinVar)

Gene: RAG1 (HGNC:5896)
Condition: recombinase activating gene 1 deficiency (MONDO:0000572)
Inheritance Mode: Autosomal recessive inheritance
UUID: 6cf7f377-1194-41d6-bf55-408745e22428
Approved on: 2024/1/23
Published on: 2024/1/23

HGVS expressions

NM_000448.3(RAG1):c.2459A>G (p.Lys820Arg)
NM_000448.3:c.2459A>G
NC_000011.10:g.36575763A>G
CM000673.2:g.36575763A>G
NC_000011.9:g.36597313A>G
CM000673.1:g.36597313A>G
NC_000011.8:g.36553889A>G
NG_007528.1:g.12751A>G
ENST00000697713.1:c.2459A>G
ENST00000697714.1:c.2459A>G
ENST00000697715.1:c.2459A>G
ENST00000299440.6:c.2459A>G
ENST00000299440.5:c.2459A>G
ENST00000524423.1:n.340T>C
ENST00000534663.1:c.2459A>G
NM_000448.2:c.2459A>G
NM_001377277.1:c.2459A>G
NM_001377278.1:c.2459A>G
NM_001377279.1:c.2459A>G
NM_001377280.1:c.2459A>G

Benign

Met criteria codes 2

BA1 BS2_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 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.2459A>G variant in RAG1 is a missense variant predicted to cause the substitution of Lysine by Arginine at amino acid 820 (p.Lys820Arg). The filtering allele frequency (the lower threshold of the 95% CI of 26338/44862) of the c.2459A>G variant in RAG1 is 0.5818 for East Asian 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, 22786 homozygous adults are reported on gnomAD v.4, BS2_Supporting is Met. 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

BA1	✔	The filtering allele frequency (the lower threshold of the 95% CI of 26338/44862) of the c.2459A>G variant in RAG1 is 0.5818 for East Asian chromosomes by gnomAD v.4, which is higher than the ClinGen SCID VCEP threshold (>0.00872) for BA1, and therefore meets this criterion (BA1).
BS2_Supporting	✔	22786 homozygous adults are reported on gnomAD v.4, BS2_Supporting is Met.

Curation History

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