

Variant: *NM_000448.3(RAG1):c.303G>A (p.Ala101=)*

Version: 1.0

CA293046 [↗](#)

138882 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: d3b4ef76-02a8-4269-9cfa-7e8c9ddd34a5

Approved on: 2024/1/17

Published on: 2024/1/17

HGVS expressions

NM_000448.3(RAG1):c.303G>A (p.Ala101=)

NM_000448.3:c.303G>A

NC_000011.10:g.36573607G>A

CM000673.2:g.36573607G>A

NC_000011.9:g.36595157G>A

CM000673.1:g.36595157G>A

NC_000011.8:g.36551733G>A

NG_007528.1:g.10595G>A

ENST00000697713.1:c.303G>A

ENST00000697714.1:c.303G>A

ENST00000697715.1:c.303G>A

ENST00000299440.6:c.303G>A

ENST00000299440.5:c.303G>A

ENST00000534663.1:c.303G>A

NM_000448.2:c.303G>A

NM_001377277.1:c.303G>A

NM_001377278.1:c.303G>A

NM_001377279.1:c.303G>A

NM_001377280.1:c.303G>A

Benign

Met criteria codes **3**

BP7

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**

Severe Combined Immunodeficiency Disease VCEP

The c.303G>A (p.Ala101=) variant (NM_000022.4) is a synonymous (silent) variant that is not predicted by SpliceAI and varSEAK to impact splicing (BP7). The filtering allele frequency (the lower threshold of the 95% CI of 799/30614) of the c.303G>A variant in RAG1 is 0.02460 for South Asian chromosomes by gnomAD v2.1.1 which is higher than the ClinGen SCID VCEP threshold (>0.00872) for BA1 and therefore meets this criterion (BA1). Additionally, 76 homozygous individuals were reported (BS2_Supporting is met). In summary, this variant is classified as a Benign for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP (specification version 1.0): BA1, BS2_Supporting, and BP7.

Met criteria codes

BP7			The c.303G>A (p.Ala101=) variant (NM_000022.4) is a synonymous (silent) variant that is not predicted by SpliceAI and varSEAK to impact splicing (BP7).
BS2_Supporting			There are 76 homozygous individuals reported (gnomAD v.2.1.1).
BA1			The filtering allele frequency (the lower threshold of the 95% CI of 799/30614) of the c.303G>A variant in RAG1 is 0.02460 for South Asian chromosomes by gnomAD v2.1.1 which is higher than the ClinGen SCID VCEP threshold (>0.00872) for BA1 and therefore meets this criterion (BA1). Additionally, 76 homozygous individuals were reported.

Curation History



 

 

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