

Variant: NM_000448.3(RAG1):c.15C>G (p.Phe5Leu)

Version: 1.0

CA5949888 [↗](#)

971195 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 6a3f0ede-f2af-4357-8d97-0ef9e2d69281

Approved on: 2024/4/23

Published on: 2024/4/23

HGVS expressions

NM_000448.3:c.15C>G

NM_000448.3(RAG1):c.15C>G (p.Phe5Leu)

NC_000011.10:g.36573319C>G

CM000673.2:g.36573319C>G

NC_000011.9:g.36594869C>G

CM000673.1:g.36594869C>G

NC_000011.8:g.36551445C>G

NG_007528.1:g.10307C>G

ENST00000697713.1:c.15C>G

ENST00000697714.1:c.15C>G

ENST00000697715.1:c.15C>G

ENST00000299440.6:c.15C>G

ENST00000299440.5:c.15C>G

ENST00000534663.1:c.15C>G

NM_000448.2:c.15C>G

NM_001377277.1:c.15C>G

NM_001377278.1:c.15C>G

NM_001377279.1:c.15C>G

NM_001377280.1:c.15C>G

Uncertain Significance

Met criteria codes **1**

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 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.15C>G variant in RAG1 is a missense variant predicted to cause a substitution of phenylalanine by leucine at amino acid 5 (p.Phe5Leu). The Popmax filtering allele frequency of this variant in gnomAD v2.1.1 is 0.00000292, which is lower than the SCID-VCEP’s threshold for PM2 (<0.000102). No homozygous individual has been observed in the gnomAD v2.1.1 (PM2_Supporting). This variant has not been reported in the literature in individuals with SCID. In ClinVar, the variant was reported in one affected individual who didn't have a second RAG1 variant, and the variant was classified as a Variant of Uncertain Significance (Invitae, SCV001420312.4). There is no functional evidence for this variant. Due to insufficient evidence, this variant is classified as a variant of uncertain significance for SCID. ACMG/AMP criteria applied, as specified by the ClinGen SCID-VCEP: PM2_supporting (SCID VCEP specifications version 1.0).

Met criteria codes

PM2_Supporting



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Curation History



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