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_000536.4(RAG2):c.48G>T (p.Gln16His)

Version: 1.0

CA380145022

2301303 (ClinVar)

Gene: RAG2 (HGNC:5897)
Condition: recombinase activating gene 2 deficiency (MONDO:0000573)
Inheritance Mode: Autosomal recessive inheritance
UUID: 0fd3f27d-0443-47f8-9c91-8a551cbaeb64
Approved on: 2024/5/1
Published on: 2024/5/1

HGVS expressions

NM_000536.4(RAG2):c.48G>T (p.Gln16His)
NM_000536.4:c.48G>T
NC_000011.10:g.36594121C>A
CM000673.2:g.36594121C>A
NC_000011.9:g.36615671C>A
CM000673.1:g.36615671C>A
NC_000011.8:g.36572247C>A
NG_007573.1:g.9116G>T
NG_033154.1:g.4629C>A
ENST00000527033.6:c.48G>T
ENST00000529083.2:c.48G>T
ENST00000532616.2:c.48G>T
ENST00000311485.8:c.48G>T
ENST00000311485.7:c.48G>T
ENST00000524423.1:n.131+3981G>T
ENST00000527033.5:c.48G>T
ENST00000529083.1:c.48G>T
ENST00000618712.4:c.48G>T
NM_000536.3:c.48G>T
NM_001243785.1:c.48G>T
NM_001243786.1:c.48G>T
NM_001243785.2:c.48G>T
NM_001243786.2:c.48G>T

Uncertain Significance

Met criteria codes 2

PM1_Supporting 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 RAG2 Version 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

NM_000536.4(RAG2):c.48G>T is a missense variant predicted to cause substitution of Glutamine by Histidine at amino acid 16 (p.Gln16His).This missense variant is located in the core domain (amino acids 1-383) (PM1_supporting). The variant is absent in gnomAD v4 (PM2_supporting). To our knowledge, this variant has not been reported in the literature in individuals affected with RAG2 related conditions or in functional studies. In summary, this variant meets the criteria to be classified as a variant of uncertain significance for autosomal recessive severe combined immunodeficiency due to RAG2 deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: PM1_supporting, PM2_supporting (VCEP specifications version 1).

Met criteria codes

PM1_Supporting	✔	This missense variant is located in the core domain (amino acids 1-383) (PM1_supporting).
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000536.4(RAG2):c.48G>T (p.Gln16…	Uncertain Significance	Recombinase Activating Gene 2 Deficiency	2024-05-01	1.0	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RAG2 Version 1.0.0	RAG2

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