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Variant: NM_000448.3(RAG1):c.906C>A (p.Asp302Glu)

Version: 1.0

CA214206

36715 (ClinVar)

Gene: RAG1 (HGNC:5896)
Condition: recombinase activating gene 1 deficiency (MONDO:0000572)
Inheritance Mode: Autosomal recessive inheritance
UUID: 81a9682b-1815-4c02-ac77-a69b610f573b
Approved on: 2024/1/17
Published on: 2024/1/17

HGVS expressions

NM_000448.3(RAG1):c.906C>A (p.Asp302Glu)
NM_000448.3:c.906C>A
NC_000011.10:g.36574210C>A
CM000673.2:g.36574210C>A
NC_000011.9:g.36595760C>A
CM000673.1:g.36595760C>A
NC_000011.8:g.36552336C>A
NG_007528.1:g.11198C>A
ENST00000697713.1:c.906C>A
ENST00000697714.1:c.906C>A
ENST00000697715.1:c.906C>A
ENST00000299440.6:c.906C>A
ENST00000299440.5:c.906C>A
ENST00000534663.1:c.906C>A
NM_000448.2:c.906C>A
NM_001377277.1:c.906C>A
NM_001377278.1:c.906C>A
NM_001377279.1:c.906C>A
NM_001377280.1:c.906C>A

Benign

Met criteria codes 1

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 c.906C>A (NM_000448.3) variant in RAG1 is a missense variant predicted to cause substitution of Aspartic Acid by Glutamic Acid at amino acid 302 (p.Asp302Glu). The filtering allele frequency (the lower threshold of the 95% CI of 1968/24958) of the c.906C>A variant in RAG1 is 0.07684 for African/African American 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). In summary, this variant meets the criteria to be classified as Benign for autosomal recessive SCID based on the ACMG/AMP criteria applied, BA1, as specified by the ClinGen SCID VCEP (VCEP specifications version 1).

Met criteria codes

BA1	✔	The filtering allele frequency (the lower threshold of the 95% CI of 1968/24958) of the c.906C>A variant in RAG1 is 0.07684 for African/African American 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).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000448.3(RAG1):c.906C>A (p.Asp...	Benign	Recombinase Activating Gene 1 Deficiency	2024-01-17	1.0	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RAG1 Version 1.0.0	RAG1

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