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Variant: NM_000206.3(IL2RG):c.43C>T (p.Gln15Ter)

Version: 1.0

CA16043285

372386 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance (recessive (HP:0001419))
UUID: 75db25fb-ac24-4642-9772-ac7b424784e3
Approved on: 2024/1/17
Published on: 2024/1/17

HGVS expressions

NM_000206.3(IL2RG):c.43C>T (p.Gln15Ter)
NM_000206.3:c.43C>T
NC_000023.11:g.71111497G>A
CM000685.2:g.71111497G>A
NC_000023.10:g.70331347G>A
CM000685.1:g.70331347G>A
NC_000023.9:g.70248072G>A
NG_009088.1:g.5057C>T
NG_021141.1:g.292C>T
ENST00000482750.6:c.43C>T
ENST00000696903.1:n.94C>T
ENST00000374202.7:c.43C>T
ENST00000642473.1:n.33C>T
ENST00000644022.1:n.75C>T
ENST00000644708.1:n.75C>T
ENST00000644911.1:n.75C>T
ENST00000645266.1:c.43C>T
ENST00000645518.1:c.43C>T
ENST00000646106.1:c.43C>T
ENST00000646505.1:c.43C>T
ENST00000647492.1:c.43C>T
ENST00000276110.6:n.54C>T
ENST00000374188.7:c.-674C>T
ENST00000374202.6:c.43C>T
ENST00000464642.5:c.-87-3C>T
ENST00000473378.1:c.-49C>T
ENST00000487883.1:c.-368C>T
ENST00000512747.3:n.110C>T
NM_000206.2:c.43C>T

Pathogenic

Met criteria codes 3

PP4 PVS1 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 IL2RG 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_000206.3(IL2RG):c.43C>T (p.Gln15Ter) nonsense variant occurs in exon 1 of 8 and, therefore, may result in NMD (PVS1). Male (0.5pt) X-SCID patient with T-B+NK+ subset (ALC of 66/mm3 with 3% CD3+, 39% CD19+, and 36% CD56+); Diagnostic criteria for SCID met (very low CD3+ T cells and this variant is Likely Pathogenic without considering PP4) 0.5pt. Total is 1pt; PP4 met (PMID: 18615703). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as pathogenic for X-linked T-B+ severe combined immunodeficiency due to gamma chain deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP. Criteria applied: PVS1, PM2_supporting, PP4. (VCEP specifications version 1).

Met criteria codes

PP4	✔	Male (0.5pt) X-SCID patient with T-B+NK+ subset (ALC of 66/mm3 with 3% CD3+, 39% CD19+, and 36% CD56+); Diagnostic criteria for SCID met (very low CD3+ T cells and this variant is Likely Pathogenic without considering PP4) 0.5pt. Total is 1pt; PP4 met (PMID: 18615703).
PVS1	✔	The NM_000206.3(IL2RG):c.43C>T (p.Gln15Ter) nonsense variant occurs in exon 1 of 8 and therefore may result in NMD (PVS1).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History

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