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Variant: NM_000206.3(IL2RG):c.181C>T (p.Gln61Ter)

Version: 1.0

CA413497169

633275 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: f9102fac-7abe-4c90-9e15-a6c4c8c21ab5
Approved on: 2024/6/12
Published on: 2024/6/12

HGVS expressions

NM_000206.3:c.181C>T
NM_000206.3(IL2RG):c.181C>T (p.Gln61Ter)
NC_000023.11:g.71110985G>A
CM000685.2:g.71110985G>A
NC_000023.10:g.70330835G>A
CM000685.1:g.70330835G>A
NC_000023.9:g.70247560G>A
NG_009088.1:g.5569C>T
NG_021141.1:g.804C>T
ENST00000482750.6:c.181C>T
ENST00000696903.1:n.232C>T
ENST00000374202.7:c.181C>T
ENST00000642473.1:n.545C>T
ENST00000644022.1:n.587C>T
ENST00000644708.1:n.587C>T
ENST00000644911.1:n.587C>T
ENST00000645266.1:c.181C>T
ENST00000645518.1:c.181C>T
ENST00000646106.1:c.181C>T
ENST00000646505.1:c.181C>T
ENST00000647492.1:c.181C>T
ENST00000276110.6:n.566C>T
ENST00000374188.7:c.-536C>T
ENST00000374202.6:c.181C>T
ENST00000456850.6:c.24+440C>T
ENST00000464642.5:c.49C>T
ENST00000473378.1:c.118C>T
ENST00000487883.1:c.145C>T
ENST00000512747.3:n.248C>T
NM_000206.2:c.181C>T

Likely Pathogenic

Met criteria codes 2

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 c.181C>T (p.Gln61Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1_Met). The variant is absent in gnomAD v4 (PM2_supporting). This variant is reported in a patient in PMID: 35874699 with no details provided. The patient doesn't meet PP4 criteria. In summary, this variant meets the criteria to be classified as a Likely Pathogenic variant for X-linked severe combined immunodeficiency due to IL2RG deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: PVS1_Met, PM2_supporting (VCEP specifications version 1).

Met criteria codes

PVS1	✔	The c.181C>T (p.Gln61Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

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