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Variant: NM_000206.3(IL2RG):c.207C>A (p.Tyr69Ter)

Version: 1.0

CA413497107

941327 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: d8fabf90-19f4-4693-87f9-0d08b2826ba4
Approved on: 2024/2/1
Published on: 2024/2/1

HGVS expressions

NM_000206.3(IL2RG):c.207C>A (p.Tyr69Ter)
NM_000206.3:c.207C>A
NC_000023.11:g.71110959G>T
CM000685.2:g.71110959G>T
NC_000023.10:g.70330809G>T
CM000685.1:g.70330809G>T
NC_000023.9:g.70247534G>T
NG_009088.1:g.5595C>A
NG_021141.1:g.830C>A
ENST00000482750.6:c.207C>A
ENST00000696903.1:n.258C>A
ENST00000374202.7:c.207C>A
ENST00000642473.1:n.571C>A
ENST00000644022.1:n.613C>A
ENST00000644708.1:n.613C>A
ENST00000644911.1:n.613C>A
ENST00000645266.1:c.207C>A
ENST00000645518.1:c.207C>A
ENST00000646106.1:c.207C>A
ENST00000646505.1:c.207C>A
ENST00000647492.1:c.207C>A
ENST00000276110.6:n.592C>A
ENST00000374188.7:c.-510C>A
ENST00000374202.6:c.207C>A
ENST00000456850.6:c.24+466C>A
ENST00000464642.5:c.75C>A
ENST00000473378.1:c.144C>A
ENST00000487883.1:c.171C>A
ENST00000512747.3:n.274C>A
NM_000206.2:c.207C>A

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PP4

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.207C>A (p.Tyr69Ter)(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).The variant is absent in gnomAD v4 (PM2_supporting). Male patient (0.5 pt.) with SCID (0.5 pt.); total: 1 pt. (PP4) (PMID: 19398866). In summary, this variant meets the criteria to be classified as a 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,PM2_supporting,PP4 (VCEP specifications version 1).

Met criteria codes

PVS1	✔	The c.207C>A (p.Tyr69Ter)(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).
PP4	✔	Male patient (0.5 pt.) with SCID (0.5 pt.); total: 1 pt. (PP4) (PMID: 19398866).

Curation History

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