

Variant: NM_000206.3(IL2RG):c.100G>T (p.Glu34Ter)

Version: 1.0

CA413497595 [↗](#)

463378 (ClinVar) [↗](#)

Gene: IL2RG ([HGNC:3561](#))

Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency ([MONDO:0010315](#))

Inheritance Mode: X-linked inheritance

UUID: 0d780336-80c9-40b0-98e1-faf71cd2dda8

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HGVS expressions

NM_000206.3:c.100G>T

NM_000206.3(IL2RG):c.100G>T (p.Glu34Ter)

NC_000023.11:g.71111440C>A

CM000685.2:g.71111440C>A

NC_000023.10:g.70331290C>A

CM000685.1:g.70331290C>A

NC_000023.9:g.70248015C>A

NG_009088.1:g.5114G>T

NG_021141.1:g.349G>T

ENST00000482750.6:c.100G>T

ENST00000696903.1:n.151G>T

ENST00000374202.7:c.100G>T

ENST00000642473.1:n.90G>T

ENST00000644022.1:n.132G>T

ENST00000644708.1:n.132G>T

ENST00000644911.1:n.132G>T

ENST00000645266.1:c.100G>T

ENST00000645518.1:c.100G>T

ENST00000646106.1:c.100G>T

ENST00000646505.1:c.100G>T

ENST00000647492.1:c.100G>T

ENST00000276110.6:n.111G>T

ENST00000374188.7:c.-617G>T

ENST00000374202.6:c.100G>T

ENST00000456850.6:c.9G>T

ENST00000464642.5:c.-33G>T

ENST00000473378.1:c.9G>T

ENST00000487883.1:c.-311G>T

ENST00000512747.3:n.167G>T

NM_000206.2:c.100G>T

Likely Pathogenic

Met criteria codes 2

PVS1PM2_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.100G>T (p.Glu34Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 1/8 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). There are no publications for this variant in the literature. 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.100G>T (p.Glu34Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 1/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).

PM2_Supporting

The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

Showing 1 to 1 of 1 rows

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