

Variant: *NM_000206.3(IL2RG):c.328G>T (p.Glu110Ter)*

Version: 1.3

CA413496806 [↗](#)

1371376 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 2c8d0838-d84e-4176-b7b5-799ccbad66a4

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HGVS expressions

NM_000206.3:c.328G>T

NM_000206.3(IL2RG):c.328G>T (p.Glu110Ter)

NC_000023.11:g.71110630C>A

CM000685.2:g.71110630C>A

NC_000023.10:g.70330480C>A

CM000685.1:g.70330480C>A

NC_000023.9:g.70247205C>A

NG_009088.1:g.5924G>T

NG_021141.1:g.1159G>T

ENST00000482750.6:c.328G>T

ENST00000696903.1:n.379G>T

ENST00000374202.7:c.328G>T

ENST00000642473.1:n.692G>T

ENST00000644022.1:n.734G>T

ENST00000644708.1:n.734G>T

ENST00000644911.1:n.734G>T

ENST00000645266.1:c.328G>T

ENST00000645518.1:c.328G>T

ENST00000646106.1:c.328G>T

ENST00000646505.1:c.328G>T

ENST00000647492.1:c.328G>T

ENST00000276110.6:n.713G>T

ENST00000374188.7:c.-389G>T

ENST00000374202.6:c.328G>T

ENST00000456850.6:c.24+795G>T

ENST00000464642.5:c.196G>T

ENST00000473378.1:c.265G>T

ENST00000487883.1:c.292G>T

ENST00000512747.3:n.395G>T

NM_000206.2:c.328G>T

Pathogenic

Met criteria codes 3

PM2_SupportingPVS1PP4_Moderate

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.328G>T (p.Glu110Ter) NM_000206.3 variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 3/8, 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). The variant was found in a *Male patient (0.5 pts) with SCID (0.5 pts) and exome sequencing (1 pt). Total is 2 pts, PP4_Moderate. (PMID: 10794430). In summary, this variant meets the criteria to be classified as a Pathogenic variant 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: PVS1, PM2_Supporting, and PP4_Moderate (VCEP specifications version 1).

Met criteria codes

PM2_Supporting	i ✓	The variant is absent in gnomAD v4 (PM2_supporting).
PVS1	i ✓	The c.328G>T (p.Glu110Ter) NM_000206.3 variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 3/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).
PP4_Moderate	i ✓	Male patient (0.5 pt.) with SCID (0.5 pt.),exome sequencing (1 pt.): Total: 2 pts. (PP4_Moderate) PMID: 10794430

Curation History

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