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Variant: NM_000206.3(IL2RG):c.710G>A (p.Trp237Ter)

Version: 1.0

CA260419

36388 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: 102e89ce-4550-481f-b12a-83c240ae02b2
Approved on: 2024/6/13
Published on: 2024/6/13

HGVS expressions

NM_000206.3:c.710G>A
NM_000206.3(IL2RG):c.710G>A (p.Trp237Ter)
NC_000023.11:g.71109275C>T
CM000685.2:g.71109275C>T
NC_000023.10:g.70329125C>T
CM000685.1:g.70329125C>T
NC_000023.9:g.70245850C>T
NG_009088.1:g.7279G>A
NG_021141.1:g.2514G>A
ENST00000482750.6:c.710G>A
ENST00000696903.1:n.761G>A
ENST00000374202.7:c.710G>A
ENST00000642473.1:n.1074G>A
ENST00000644022.1:n.976G>A
ENST00000644708.1:n.1116G>A
ENST00000644911.1:n.1116G>A
ENST00000645266.1:c.710G>A
ENST00000645518.1:c.710G>A
ENST00000646106.1:c.710G>A
ENST00000646505.1:c.710G>A
ENST00000647492.1:c.710G>A
ENST00000276110.6:n.1303G>A
ENST00000374188.7:c.-7G>A
ENST00000374202.6:c.710G>A
ENST00000456850.6:c.140G>A
ENST00000464642.5:c.578G>A
ENST00000482750.5:c.123G>A
ENST00000512747.3:n.637G>A
NM_000206.2:c.710G>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.710G>A (p.Trp237Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 5/8, leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met). This variant is absent from gnomAD v4 (PM2_Supporting). At least one proband in the literature presents: Diagnostic criteria for SCID/Leaky SCID/Omenn syndrome met (0.5 pts) + XY male sex (0.5 pts), total is 1 point, PP4 is met (PMID: 28702026). 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 (specification version 1.0): PM2_Supporting, PVS1, and PP4.

Met criteria codes

PVS1	✔	The c.710G>A (p.Trp237Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 5/8, leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).
PM2_Supporting	✔	This variant is absent from gnomAD v4 (PM2_Supporting).
PP4	✔	At least one proband in the literature presents: Diagnostic criteria for SCID/Leaky SCID/Omenn syndrome met (0.5 pts) + XY male sex (0.5 pts), total is 1 point, PP4 is met (PMID: 28702026).

Curation History

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