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Variant: NM_000206.3(IL2RG):c.281C>A (p.Ser94Ter)

Version: 1.0

CA413496915

488696 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: c4092af2-9815-4b28-8afb-597deb955bd3
Approved on: 2024/6/12
Published on: 2024/6/12

HGVS expressions

NM_000206.3(IL2RG):c.281C>A (p.Ser94Ter)
NM_000206.3:c.281C>A
NC_000023.11:g.71110677G>T
CM000685.2:g.71110677G>T
NC_000023.10:g.70330527G>T
CM000685.1:g.70330527G>T
NC_000023.9:g.70247252G>T
NG_009088.1:g.5877C>A
NG_021141.1:g.1112C>A
ENST00000482750.6:c.281C>A
ENST00000696903.1:n.332C>A
ENST00000374202.7:c.281C>A
ENST00000642473.1:n.645C>A
ENST00000644022.1:n.687C>A
ENST00000644708.1:n.687C>A
ENST00000644911.1:n.687C>A
ENST00000645266.1:c.281C>A
ENST00000645518.1:c.281C>A
ENST00000646106.1:c.281C>A
ENST00000646505.1:c.281C>A
ENST00000647492.1:c.281C>A
ENST00000276110.6:n.666C>A
ENST00000374188.7:c.-436C>A
ENST00000374202.6:c.281C>A
ENST00000456850.6:c.24+748C>A
ENST00000464642.5:c.149C>A
ENST00000473378.1:c.218C>A
ENST00000487883.1:c.245C>A
ENST00000512747.3:n.348C>A
NM_000206.2:c.281C>A

Pathogenic

Met criteria codes 3

PM2_Supporting | PP4 | PVS1

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.281C>A (p.Ser94Ter) (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). Male Patient (0.5 pt.) with SCID (0.5 pt.) (Total : 1 pt. , PP4 met, PMID: 23250629). 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

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 met, PMID: 23250629)
PVS1	✔	The c.281C>A (p.Ser94Ter) (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).

Curation History

Showing 1 to 1 of 1 rows

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