The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000206.3(IL2RG):c.184T>A (p.Cys62Ser)

Version: 1.0

CA413497161

636917 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: 56f53605-6436-4776-a982-efe087c916a9
Approved on: 2024/5/28
Published on: 2024/5/28

HGVS expressions

NM_000206.3(IL2RG):c.184T>A (p.Cys62Ser)
NM_000206.3:c.184T>A
NC_000023.11:g.71110982A>T
CM000685.2:g.71110982A>T
NC_000023.10:g.70330832A>T
CM000685.1:g.70330832A>T
NC_000023.9:g.70247557A>T
NG_009088.1:g.5572T>A
NG_021141.1:g.807T>A
ENST00000482750.6:c.184T>A
ENST00000696903.1:n.235T>A
ENST00000374202.7:c.184T>A
ENST00000642473.1:n.548T>A
ENST00000644022.1:n.590T>A
ENST00000644708.1:n.590T>A
ENST00000644911.1:n.590T>A
ENST00000645266.1:c.184T>A
ENST00000645518.1:c.184T>A
ENST00000646106.1:c.184T>A
ENST00000646505.1:c.184T>A
ENST00000647492.1:c.184T>A
ENST00000276110.6:n.569T>A
ENST00000374188.7:c.-533T>A
ENST00000374202.6:c.184T>A
ENST00000456850.6:c.24+443T>A
ENST00000464642.5:c.52T>A
ENST00000473378.1:c.121T>A
ENST00000487883.1:c.148T>A
ENST00000512747.3:n.251T>A
NM_000206.2:c.184T>A

Likely Pathogenic

Met criteria codes 3

PM1_Strong PP4_Moderate PM2_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

NM_000206.3(IL2RG):c.184T>A is a missense variant predicted to cause substitution of Cysteine by Serine at amino acid 62 (p.Cys62Ser). This variant affects a conserved cysteine residue Cys62 (PM1_strong).The variant is absent in gnomAD v4 (PM2_supporting). Male patient (0.5 pt.) with SCID (0.5 pt.), genome sequencing conducted (1 pt.) ,total :2 pts. (PMID : 35874699) (PP4_Moderate). 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: PM1_strong, PM2_supporting, PP4_moderate (VCEP specifications version 1).

Met criteria codes

PM1_Strong	✔	This variant affects a conserved cysteine residue Cys62 (PM1_strong).
PP4_Moderate	✔	Male patient (0.5 pt.) with SCID (0.5 pt.), genome sequencing conducted (1 pt.) ,total :2 pts. (PMID : 35874699) (PP4_Moderate)
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

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