

Variant: NM_000206.3(IL2RG):c.186T>A (p.Cys62Ter)

Version: 1.0

CA254980 [↗](#)

10019 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: da8b976a-ccac-434c-b458-c2cdd9d68446

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HGVS expressions

NM_000206.3:c.186T>A

NM_000206.3(IL2RG):c.186T>A (p.Cys62Ter)

NC_000023.11:g.71110980A>T

CM000685.2:g.71110980A>T

NC_000023.10:g.70330830A>T

CM000685.1:g.70330830A>T

NC_000023.9:g.70247555A>T

NG_009088.1:g.5574T>A

NG_021141.1:g.809T>A

ENST00000482750.6:c.186T>A

ENST00000696903.1:n.237T>A

ENST00000374202.7:c.186T>A

ENST00000642473.1:n.550T>A

ENST00000644022.1:n.592T>A

ENST00000644708.1:n.592T>A

ENST00000644911.1:n.592T>A

ENST00000645266.1:c.186T>A

ENST00000645518.1:c.186T>A

ENST00000646106.1:c.186T>A

ENST00000646505.1:c.186T>A

ENST00000647492.1:c.186T>A

ENST00000276110.6:n.571T>A

ENST00000374188.7:c.-531T>A

ENST00000374202.6:c.186T>A

ENST00000456850.6:c.24+445T>A

ENST00000464642.5:c.54T>A

ENST00000473378.1:c.123T>A

ENST00000487883.1:c.150T>A

ENST00000512747.3:n.253T>A

NM_000206.2:c.186T>A

Pathogenic

Met criteria codes 3

PP4PVS1PM2_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.186T>A (p.Cys62Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2/8 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.), Absent CD132 expression (demonstrated by RT-PCR) (1 pt.) total :1.5 pts (PP4) (PMID: 8462096). 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 Met,PP4,PM2_supporting (VCEP specifications version 1).

Met criteria codes

PP4		✓	Male patient (0.5 pt.), Absent CD132 expression (demonstrated by RT-PCR) (1 pt.) total :1.5 pts (PP4) (PMID: 8462096)
PVS1		✓	The c.186T>A (p.Cys62Ter) (NM_000206.3) variant in IL2RG is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2/8 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1 Met).
PM2_Supporting		✓	The variant is absent in gnomAD v4 (PM2_supporting).

Curation History

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