

Variant: *NM\_000206.3(IL2RG):c.1061A>G (p.His354Arg)*

Version: 2.0

[CA10443761](#)

[1199323 \(ClinVar\)](#)

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

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

**Inheritance Mode:** X-linked inheritance

**UUID:** c507f13c-43cd-4cfe-b95f-fa1e3960702c

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### *HGVS expressions*

**NM\_000206.3:c.1061A>G**

NM\_000206.3(IL2RG):c.1061A>G (p.His354Arg)

NC\_000023.11:g.71107785T>C

CM000685.2:g.71107785T>C

NC\_000023.10:g.70327635T>C

CM000685.1:g.70327635T>C

NC\_000023.9:g.70244360T>C

NG\_009088.1:g.8769A>G

NG\_021141.1:g.4004A>G

ENST00000482750.6:c.\*181A>G

ENST00000696903.1:n.1364A>G

ENST00000374202.7:c.1061A>G

ENST00000642473.1:n.1288+492A>G

ENST00000644022.1:n.1190+492A>G

ENST00000644708.1:n.1302+68A>G

ENST00000644911.1:n.1399+68A>G

ENST00000645266.1:c.924+492A>G

ENST00000645518.1:c.924+492A>G

ENST00000646106.1:c.993+68A>G

ENST00000646505.1:c.924+492A>G

ENST00000647492.1:c.924+492A>G

ENST00000276110.6:n.1654A>G

ENST00000374188.7:c.248A>G

ENST00000374202.6:c.1061A>G

ENST00000456850.6:c.491A>G

ENST00000482750.5:c.377A>G

ENST00000512747.3:n.1595A>G

NM\_000206.2:c.1061A>G

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **1**

PM2

Expert Panel

Severe Combined Immunodeficiency Disease VCEP [↗](#)

Criteria Specification Information

## Evidence submitted by expert panel

**Severe Combined Immunodeficiency Disease VCEP**

**NM\_000206.3(IL2RG):c.1061A>G** is a missense variant predicted to cause substitution of Histidine by Arginine at amino acid 354 (p.His354Arg). The filtering allele frequency (the upper threshold of the 95% CI of 7/30447) of the c.1061A>G variant in IL2RG is 0.0001216 for East Asian chromosomes by gnomAD v4, which is lower than the ClinGen SCID VCEP threshold (<0.000124 ) for PM2\_Supporting. However, 4 hemizygotes were reported (PM2 not met).The variant is observed in 4 hemizygotes in gnomAD v4 (BS2\_Strong). To our knowledge, this variant has not been reported in the literature in individuals affected with IL2RG related conditions or in functional studies. In summary, this variant meets the criteria to be classified as likely benign 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: BS2 (VCEP specifications version 1).

**Met criteria codes****BS2**

The variant is observed in 4 hemizygotes in gnomAD v4 (BS2\_Strong).

**Not Met criteria codes****PM2**

The filtering allele frequency (the upper threshold of the 95% CI of 7/30447) of the c.1061A&gt;G variant in IL2RG is 0.0001216 for East Asian chromosomes by gnomAD v4, which is lower than the ClinGen SCID VCEP threshold (&lt;0.000124 ) for PM2\_Supporting. However, 4 hemizygotes were reported (PM2 not met).

Curation History [↗](#)

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