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.202G>A (p.Glu68Lys)

Version: 1.0

CA16608976

379561 (ClinVar)

Gene: IL2RG (HGNC:3561)
Condition: T-B+ severe combined immunodeficiency due to gamma chain deficiency (MONDO:0010315)
Inheritance Mode: X-linked inheritance
UUID: 2777e3d3-64d9-471a-a3d8-c36d5f50889d
Approved on: 2024/1/31
Published on: 2024/1/31

HGVS expressions

NM_000206.3(IL2RG):c.202G>A (p.Glu68Lys)
NM_000206.3:c.202G>A
NC_000023.11:g.71110964C>T
CM000685.2:g.71110964C>T
NC_000023.10:g.70330814C>T
CM000685.1:g.70330814C>T
NC_000023.9:g.70247539C>T
NG_009088.1:g.5590G>A
NG_021141.1:g.825G>A
ENST00000482750.6:c.202G>A
ENST00000696903.1:n.253G>A
ENST00000374202.7:c.202G>A
ENST00000642473.1:n.566G>A
ENST00000644022.1:n.608G>A
ENST00000644708.1:n.608G>A
ENST00000644911.1:n.608G>A
ENST00000645266.1:c.202G>A
ENST00000645518.1:c.202G>A
ENST00000646106.1:c.202G>A
ENST00000646505.1:c.202G>A
ENST00000647492.1:c.202G>A
ENST00000276110.6:n.587G>A
ENST00000374188.7:c.-515G>A
ENST00000374202.6:c.202G>A
ENST00000456850.6:c.24+461G>A
ENST00000464642.5:c.70G>A
ENST00000473378.1:c.139G>A
ENST00000487883.1:c.166G>A
ENST00000512747.3:n.269G>A
NM_000206.2:c.202G>A

Pathogenic

Met criteria codes 3

PS4 PM2_Supporting PP4_Moderate

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.202G>A is a missense variant predicted to cause substitution of Glutamic Acid by Lysine at amino acid 68 (p.Glu68Lys). The variant is absent in gnomAD v4 (PM2_supporting). Male patient (0.5 pt.) with SCID (0.5 pt.), genome sequencing conducted (1 pt.), Absent CD132 expression (1 pt.), T-B+NK- lymphocyte subset profile (0.5 pt.); total :3.5 pts PMID: 30778343 (PP4_moderate). This variant is found in multiple unrelated affected probands (total pts. >16) (PS4_very strong) (PMIDs: 9058718, 24534054,11129345, 32477911,31456805, 27484032,31031743,30778343). As per the SCID VCEP specifications and the Bayesian interpretation of the ACMG/AMP combining rules, 1 Very Strong,1 Moderate and 1 Supporting criteria results in a Pathogenic classification. In summary, this variant meets the criteria to be classified as Pathogenic for X-linked severe combined immunodeficiency due to IL2RG deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: PM2_supporting,PP4_moderate,PS4_very strong (VCEP specifications version 1).

Met criteria codes

PS4	✔	This variant is found in multiple unrelated affected probands (total pts. >16) (PS4_very strong) (PMIDs: 9058718, 24534054,11129345, 32477911,31456805, 27484032,31031743,30778343)
PM2_Supporting	✔	The variant is absent in gnomAD v4 (PM2_supporting).
PP4_Moderate	✔	Male patient (0.5 pt.) with SCID (0.5 pt.), genome sequencing conducted (1 pt.), Absent CD132 expression (1 pt.), T-B+NK- lymphocyte subset profile (0.5 pt.); total :3.5 pts PMID: 30778343 (PP4_moderate).

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.