

Variant: *NM_000215.4(JAK3):c.452C>G (p.Pro151Arg)*

Version: 1.0

CA160252 [↗](#)

36422 (ClinVar) [↗](#)

Gene: JAK3 ([HGNC:3718](#))

Condition: T-B+ severe combined immunodeficiency due to JAK3 deficiency ([MONDO:0010938](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 1b8a4b35-8c5e-42d1-a036-34d2454bce11

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HGVS expressions

NM_000215.4(JAK3):c.452C>G (p.Pro151Arg)

NM_000215.4:c.452C>G

NC_000019.10:g.17843141G>C

CM000681.2:g.17843141G>C

NC_000019.9:g.17953950G>C

CM000681.1:g.17953950G>C

NC_000019.8:g.17814950G>C

NG_007273.1:g.9851C>G

ENST00000526008.6:c.452C>G

ENST00000458235.7:c.452C>G

ENST00000458235.5:c.452C>G

ENST00000526008.5:n.552C>G

ENST00000527031.5:n.542C>G

ENST00000527670.5:c.452C>G

ENST00000528293.1:n.467C>G

ENST00000534444.1:c.452C>G

NM_000215.3:c.452C>G

Benign

Met criteria codes **2**

BA1

BS2_Supporting

Not Met criteria codes **1**

BS1

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 JAK3 Version 1.0.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The NM_000215.4(JAK3):c.452C>G (p.Pro151Arg) variant in JAK3 is a missense variant predicted to cause the substitution of Proline by Arginine at amino acid 151 (p.Pro151Arg). The Popmax Filtering allele frequency (95% CI) of the variant is 0.008679 in gnomAD v.4 for European (non-Finnish) population 10486/1179548 alleles, which is higher than the ClinGen SCID VCEP threshold (>0.00447) for BA1, therefore, meets this criterion (BA1). Also, 59 homozygous adults are reported on GnomAD v2.1.1 (BS2_Supporting). In summary, this variant meets the criteria to be classified as Benign for autosomal recessive T-B+ severe combined immunodeficiency due to JAK3 deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP. Criteria applied: BA1 and BS2_Supporting (VCEP specifications version 1.0).

Met criteria codes

BA1			The Popmax Filtering allele frequency (95% CI) of the variant is 0.008679 in gnomAD v.4 for European (non-Finnish) population 10486/1179548 alleles, which is higher than the ClinGen SCID VCEP threshold (>0.00447) for BA1, therefore, meets this criterion (BA1).
BS2_Supporting			59 homozygous adults are reported on GnomAD v.4 (BS2_Supporting).

Not Met criteria codes

BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History



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