

Variant: *NM_005214.5(CTLA4):c.615C>G (p.Pro205=)*

Version: 1.0

CA2067158 [↗](#)

741101 (ClinVar) [↗](#)

Gene: CTLA4 ([HGNC:1493](#))

Condition: autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency ([MONDO:0014493](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 0c9d5cdf-c9d0-4199-857c-8b46e4dc779a

Approved on: 2025/11/24

Published on: 2025/12/2

HGVS expressions

NM_005214.5:c.615C>G

NM_005214.5(CTLA4):c.615C>G (p.Pro205=)

NC_000002.12:g.203872755C>G

CM000664.2:g.203872755C>G

NC_000002.11:g.204737478C>G

CM000664.1:g.204737478C>G

NC_000002.10:g.204445723C>G

NG_011502.1:g.9970C>G

ENST00000696049.1:c.555C>G

ENST00000696479.1:c.687C>G

ENST00000427473.3:n.539C>G

ENST00000648405.2:c.615C>G

ENST00000650075.1:n.639C>G

ENST00000295854.10:c.505C>G

ENST00000302823.7:c.615C>G

ENST00000427473.2:c.394C>G

ENST00000472206.1:c.220C>G

ENST00000487393.1:n.157C>G

NM_001037631.2:c.505C>G

NM_005214.4:c.615C>G

NM_001037631.3:c.505C>G

Benign

Met criteria codes 3

BP4 BP7 BA1

Not Met criteria codes 1

PP3

Evidence Links 0

Expert Panel

Antibody Deficiencies VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Antibody Deficiencies Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for CTLA4 Version 1.0.0

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

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

Antibody Deficiencies VCEP

NM_005214.5(CTLA4):c.615C>G (p.Pro205=) is a synonymous variant in exon 2 that does not have a predicted impact at splicing sites (BP7). The splicing impact predictor SpliceAI gives a delta score of 0.03 for acceptor gain, which is below the ClinGen Antibody Deficiencies VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4). This variant is present in gnomAD v4.1.0 at a GrpMax allele frequency of 0.0004811, with 30 alleles / 44,874 total alleles in the East Asian population, which is higher than the ClinGen Antibody Deficiencies VCEP BA1 threshold of >0.0000111 (BA1). In summary, this variant meets the criteria to be classified as benign for autosomal dominant autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen Antibody Deficiencies VCEP: BA1, BP4, and BP7. (VCEP specifications version 1.0.0; date of approval 09/18/2025).

Met criteria codes

BP4			The splicing impact predictor SpliceAI gives a delta score of 0.03 for acceptor gain, which is below the ClinGen Antibody Deficiencies VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4).
BP7			The synonymous variant NM_005214.5(CTLA4):c.615C>G (p.Pro205=) does not have a predicted impact at splicing sites (BP7).
BA1			This variant is present in gnomAD v4.1.0 at a GrpMax allele frequency of 0.0004811, with 30 alleles / 44,874 total alleles in the East Asian population, which is higher than the ClinGen Antibody Deficiencies VCEP BA1 threshold of >0.0000111 (BA1).

Not Met criteria codes

PP3			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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