

*Variant: NM_001033855.3(DCLRE1C):c.181T>C
(p.Cys61Arg)*

Version: 1.0

CA376063513 [↗](#)

648095 (ClinVar) [↗](#)

Gene: DCLRE1C ([HGNC:64421](#))

Condition: severe combined immunodeficiency due to DCLRE1C deficiency
([MONDO:0011225](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 0c4780cf-802d-4844-b6f4-4edffd11937d

Approved on: 2024/2/15

Published on: 2024/2/15

HGVS expressions

NM_001033855.3(DCLRE1C):c.181T>C (p.Cys61Arg)

NM_001033855.3:c.181T>C

NC_000010.11:g.14945170A>G

CM000672.2:g.14945170A>G

NC_000010.10:g.14987169A>G

CM000672.1:g.14987169A>G

NC_000010.9:g.15027175A>G

NG_007276.1:g.13926T>C

ENST00000378241.6:c.*101T>C

ENST00000456122.2:c.*101T>C

ENST00000489161.2:c.*15T>C

ENST00000492201.6:c.181T>C

ENST00000697047.1:c.181T>C

ENST00000697070.1:c.181T>C

ENST00000697071.1:c.*101T>C

ENST00000697072.1:c.181T>C

ENST00000697073.1:c.*15T>C

ENST00000697074.1:c.*15T>C

ENST00000697075.1:c.181T>C

ENST00000697076.1:c.181T>C

ENST00000697077.1:c.181T>C

ENST00000697078.1:c.181T>C

ENST00000697079.1:n.1T>C

ENST00000697080.1:c.*101T>C

ENST00000697081.1:c.181T>C

ENST00000697082.1:c.*101T>C

ENST00000697083.1:c.*101T>C

ENST00000697084.1:c.181T>C

ENST00000697085.1:c.181T>C

ENST00000697086.1:n.2618T>C

ENST00000697087.1:c.*101T>C

ENST00000697088.1:c.181T>C

ENST00000697089.1:c.*101T>C

ENST00000697090.1:n.189T>C

ENST00000697091.1:n.242T>C

	ENST00000378278.7:c.181T>C ENST00000357717.6:c.-44+3866T>C ENST00000378241.5:c.-307T>C ENST00000378246.6:c.-109T>C ENST00000378249.5:c.-109T>C ENST00000378254.5:c.-180T>C ENST00000378255.5:c.-502T>C ENST00000378258.5:c.-180T>C ENST00000378278.6:c.181T>C ENST00000378289.8:c.181T>C ENST00000396817.6:c.-502T>C ENST00000418843.5:c.-217T>C ENST00000456122.1:c.-431T>C NM_001033855.2:c.181T>C NM_001033857.2:c.-180T>C NM_001033858.2:c.-502T>C NM_001289076.1:c.-44+3866T>C NM_001289077.1:c.-180T>C NM_001289078.1:c.-109T>C NM_001289079.1:c.-502T>C NM_022487.3:c.-109T>C NR_110297.1:n.688T>C NM_001350965.1:c.181T>C NM_001350966.1:c.-109T>C NM_001350967.1:c.-180T>C NR_146960.1:n.603T>C NR_146961.1:n.688T>C NR_146962.1:n.603T>C NM_001033857.3:c.-180T>C NM_001033858.3:c.-502T>C NM_001289076.2:c.-44+3866T>C NM_001289077.2:c.-180T>C NM_001289078.2:c.-109T>C NM_001289079.2:c.-502T>C NM_001350965.2:c.181T>C NM_001350966.2:c.-109T>C NM_001350967.2:c.-180T>C NM_022487.4:c.-109T>C NR_110297.2:n.352T>C NR_146961.2:n.352T>C
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Uncertain Significance

Met criteria codes 1

PM2_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 DCLRE1C Version 1.0.0

Criteria Specification Approval History

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The **c.181T>C (NM_001033855.3)** variant in **DCLRE1C** is a missense variant predicted to cause substitution of Cysteine by Arginine at amino acid **61** (p.Cys61Arg). This variant is absent from gnomAD v4 (PM2_Supporting). To our knowledge, this variant has not been reported in the literature in individuals affected with DCLRE1C-related conditions or in functional studies. In summary, this variant meets the criteria to be classified as a variant of uncertain significance for autosomal recessive severe combined immunodeficiency due to DCLRE1C deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP. Criteria applied: PM2_Supporting (VCEP specifications version 1.0).

Met criteria codes

PM2_Supporting   This variant is absent from gnomAD v4 (PM2_Supporting).

Curation History [↗](#)



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