

Variant: NM_001033855.3(DCLRE1C):c.973-1801T>A

Version: 1.0

CA203388902 [↗](#)

1679474 (ClinVar) [↗](#)

Gene: DCLRE1C (HGNC:64421)

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

Inheritance Mode: Autosomal recessive inheritance

UUID: c5f15080-5d5f-4040-9c29-844b3b06be47

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HGVS expressions

NM_001033855.3:c.973-1801T>A

NM_001033855.3(DCLRE1C):c.973-1801T>A

NC_000010.11:g.14924870A>T

CM000672.2:g.14924870A>T

NC_000010.10:g.14966869A>T

CM000672.1:g.14966869A>T

NC_000010.9:g.15006875A>T

NG_007276.1:g.34226T>A

ENST00000378241.6:c.*1020-1801T>A

ENST00000456122.2:c.*1159-1801T>A

ENST00000489161.2:c.*696-1801T>A

ENST00000492201.6:c.973-1801T>A

ENST00000697047.1:c.973-1801T>A

ENST00000697070.1:c.973-1801T>A

ENST00000697071.1:c.*893-1801T>A

ENST00000697072.1:c.918-1801T>A

ENST00000697073.1:c.*751-1801T>A

ENST00000697074.1:c.*751-1801T>A

ENST00000697075.1:c.973-1801T>A

ENST00000697076.1:c.918-1801T>A

ENST00000697077.1:c.*684-1801T>A

ENST00000697078.1:c.*680-1801T>A

ENST00000697079.1:n.677-1801T>A

ENST00000697080.1:c.*837-1801T>A

ENST00000697081.1:c.*590-1801T>A

ENST00000697082.1:c.*1002-1801T>A

ENST00000697083.1:c.*778-1801T>A

ENST00000697084.1:c.1029+276T>A

ENST00000697085.1:c.*740-1801T>A

ENST00000697086.1:n.3410-1801T>A

ENST00000697087.1:c.*893-1801T>A

ENST00000697088.1:c.*590-1801T>A

ENST00000697089.1:c.*893-1801T>A

ENST00000378278.7:c.973-1801T>A

ENST00000357717.6:c.628-1801T>A

ENST00000378246.6:c.628-1801T>A

ENST00000378249.5:c.628-1801T>A

ENST00000378254.5:c.613-1801T>A
ENST00000378255.5:c.613-1801T>A
ENST00000378258.5:c.613-1801T>A
ENST00000378278.6:c.973-1801T>A
ENST00000378289.8:c.973-1801T>A
ENST00000396817.6:c.613-1801T>A
ENST00000489161.1:n.48-1801T>A
ENST00000492201.5:n.123-1801T>A
NM_001033855.2:c.973-1801T>A
NM_001033857.2:c.613-1801T>A
NM_001033858.2:c.613-1801T>A
NM_001289076.1:c.628-1801T>A
NM_001289077.1:c.613-1801T>A
NM_001289078.1:c.628-1801T>A
NM_001289079.1:c.613-1801T>A
NM_022487.3:c.628-1801T>A
NR_110297.1:n.1607-1801T>A
NM_001350965.1:c.973-1801T>A
NM_001350966.1:c.628-1801T>A
NM_001350967.1:c.613-1801T>A
NR_146960.1:n.1340-1801T>A
NR_146961.1:n.1424-1801T>A
NR_146962.1:n.1395-1801T>A
NM_001033857.3:c.613-1801T>A
NM_001033858.3:c.613-1801T>A
NM_001289076.2:c.628-1801T>A
NM_001289077.2:c.613-1801T>A
NM_001289078.2:c.628-1801T>A
NM_001289079.2:c.613-1801T>A
NM_001350965.2:c.973-1801T>A
NM_001350966.2:c.628-1801T>A
NM_001350967.2:c.613-1801T>A
NM_022487.4:c.628-1801T>A
NR_110297.2:n.1271-1801T>A
NR_146961.2:n.1088-1801T>A

Uncertain Significance

Met criteria codes **1**

BP7

Not Met criteria codes **3**

BS1 BA1 PM2

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](#)



[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel






Severe Combined Immunodeficiency Disease VCEP

The c.973-1801T>A (NM_001033855.3) variant in DCLRE1C is an intronic variant which locates in the deep intronic region in the intron 11. It is not predicted to impact splicing by SpliceAI, varSEAK, and NNSplice. BP7 is met. The highest population minor allele frequency in gnomAD v2.1.1 is 0.0004608 (4/8180 alleles) in African/African American population, which is lower than the SCID-VCEP threshold for BS1 (>0.00078) and BA1 (>0.00346) but higher than the threshold (<0.00003266) for PM2_Supporting (BS1 not met, BA1 not met, PM2_Supporting not met). To our knowledge, this variant has not been reported in the literature in individuals affected with DCLRE1C/SCID conditions or in functional studies. Due to insufficient evidence, this variant is classified as a variant of uncertain significance for autosomal recessive SCID based on ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP (specification version 1.0).

Met criteria codes

| | | | |
|------------|---|---|---|
| BP7 |  |  | The c.973-1801T>A (NM_001033855.3) variant in DCLRE1C is an intronic variant which locates in the deep intronic region in the intron 11. It is not predicted to impact splicing by SpliceAI, varSEAK, and NNSplice. BP7 is met. |
|------------|---|---|---|

Not Met criteria codes

| | | | |
|------------|---|---|---|
| BS1 |  |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| BA1 |  |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| PM2 | |  | The highest population minor allele frequency in gnomAD v2.1.1 is 0.0004608 (4/8180 alleles) in African/African American population, which is lower than the SCID-VCEP threshold for BS1 (>0.00078) and BA1 (>0.00346) but higher than the threshold (<0.00003266) for PM2_Supporting (BS1 not met, BA1 not met, PM2_Supporting not met). |

Curation History

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