

Variant: NR_003051.3(RMRP):n.-17_-15dupTGT

Version: 1.4

CA658821614 [↗](#)

558632 (ClinVar) [↗](#)

Gene: RMRP (HGNC:6023)

Condition: cartilage-hair hypoplasia (MONDO:0009595)

Inheritance Mode: Autosomal recessive inheritance

UID: 50e33583-97fa-4f94-8ef1-bb7d96cdd560

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HGVS expressions

NR_003051.3(RMRP):n.-17_-15dupTGT
NC_000009.12:g.35658033_35658035dup
CM000671.2:g.35658033_35658035dup
NC_000009.11:g.35658030_35658032dup
CM000671.1:g.35658030_35658032dup
NC_000009.10:g.35648030_35648032dup
NG_017041.1:g.4984_4986dup
NG_033120.1:g.4744_4746dup

Uncertain Significance

Met criteria codes **2**

PM2_Supporting PM1_Strong

Not Met criteria codes **1**

PM4

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 RMRP Version 1.0.0

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







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

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

The variant NC_000009.12:g.35658033_35658035dup is absent in population databases such as gnomAD v4. **PM2_Supporting** This variant creates an extension of 3 nucleotides, increasing the distance between the TATA box (spanning n.-32 to n.-24) and the transcription start site (n.1) **PM1_Strong**. However, the insertion is only 3 nucleotides and therefore this criterion is not met. The variant has not been described in patients with cartilage-hair hypoplasia or in trans with any other RMRP variant In summary, this variant is classified as **Uncertain significance: PM2_Supporting, PM1_Strong**.

Met criteria codes

PM2_Supporting			This variant is absent in population databases such as gnomAD v4
PM1_Strong			This variant falls in the region between the TATA box and the initiation of transcription n.1.
Not Met criteria codes			
PM4			This variant creates an extension of 3 nucleotides, increasing the distance between the TATA box (spanning n.-32 to n.-24) and the transcription start site (n.1). However the insertion is only 3 nucleotides and therefore this criterion is not met.

Curation History [↗](#)

Showing 1 to 5 of 5 rows

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