The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NR_003051.4(RMRP):n.263C>G

Version: 1.3

CA464450136

552081 (ClinVar)

Gene: N/A
Condition: cartilage-hair hypoplasia (MONDO:0009595)
Inheritance Mode: Autosomal recessive inheritance
UUID: 9bca14fb-743e-40ff-aa85-08b132b8634e
Approved on: 2025-06-10
Published on: 2026-02-06

HGVS expressions

NR_003051.4:n.263C>G
NR_003051.4(RMRP):n.263C>G
NC_000009.12:g.35657757G>C
CM000671.2:g.35657757G>C
NC_000009.11:g.35657754G>C
CM000671.1:g.35657754G>C
NC_000009.10:g.35647754G>C
NG_017041.1:g.5262C>G
NG_033120.1:g.4468G>C
NR_003051.3:n.262C>G

Uncertain Significance

Met criteria codes 2

PP4 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 RMRP Version 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Severe Combined Immunodeficiency Disease VCEP

This variant has been found in gnomAD v4 with an allele frequency of 0.000007990 in the African/ African American population. This variant is below the threshold that the SCID VCEP established to use PM2_Supporting (< 0.0000447). Therefore, this criterion is met: PM2_Supporting. At least one patient (P5, PMID: 16244706) has been described with this variant (also known as g.260C>G). The patient presented with metaphyseal dysplasia (+1.0), immune deficiency phenotype (+0.5), and hematological disease (+0.25), reaching a total of 1.75 points. Therefore PP4 is met at default strength. In summary, this variant is classified as uncertain significance - insufficient evidence for Autosomal Recessive Cartilage Hair Hypoplasia based on the ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: PM2_Supporting, PP4 (SCID VCEP specifications version 1).

Met criteria codes

PP4	✔	At least one patient (P5, PMID: 16244706) has been described with this variant (also known as g.260C>G). The patient presented with metaphyseal dysplasia (+1.0), immune deficiency phenotype (+0.5), and hematological disease (+0.25), reaching a total of 1.75 points. Therefore PP4 is met at default strength.
PM2_Supporting	✔	This variant has been found in gnomAD v4 with an allele frequency of 0.000007990 in the African/ African American population. This variant is below the threshold that the SCID VCEP established to use PM2_Supporting (< 0.0000447). Therefore this criterion is met PM2_Supporting

Curation History

Showing 1 to 4 of 4 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NR_003051.4(RMRP):n.263C>G	Uncertain Significance	Cartilage-Hair Hypoplasia	2026-02-06	1.3	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RMRP Version 1.0.0	N/A
View	NR_003051.4(RMRP):n.263C>G	Uncertain Significance	Cartilage-Hair Hypoplasia	2026-02-06	1.2	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RMRP Version 1.0.0	N/A
View	NR_003051.4(RMRP):n.263C>G	Uncertain Significance	Cartilage-Hair Hypoplasia	2025-06-10	1.1	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RMRP Version 1.0.0	RMRP
View	NR_003051.4(RMRP):n.263C>G	Uncertain Significance	Cartilage-Hair Hypoplasia	2025-06-10	1.0	ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RMRP Version 1.0.0	N/A

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