

Variant: *NM_004086.3(COCH):c.260G>T (p.Gly87Val)*

Version: 1.0

CA389344543 [↗](#)

4532319 (ClinVar) [↗](#)

Gene: COCH ([HGNC:1690](#))

Condition: nonsyndromic genetic deafness ([MONDO:0019497](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 6420bd38-2259-427e-9383-048b8d482bbd

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HGVS expressions

NM_004086.3:c.260G>T

NM_004086.3(COCH):c.260G>T (p.Gly87Val)

NC_000014.9:g.30878831G>T

CM000676.2:g.30878831G>T

NC_000014.8:g.31348037G>T

CM000676.1:g.31348037G>T

NC_000014.7:g.30417788G>T

NG_008211.2:g.9297G>T

ENST00000216361.9:c.455G>T

ENST00000396618.9:c.260G>T

ENST00000555117.2:c.260G>T

ENST00000643575.1:c.260G>T

ENST00000643697.1:n.505G>T

ENST00000644874.2:c.260G>T

ENST00000216361.8:c.260G>T

ENST00000396618.7:c.260G>T

ENST00000460581.6:c.-77G>T

ENST00000475087.5:c.260G>T

ENST00000553772.5:c.239+1103G>T

ENST00000553833.5:n.414G>T

ENST00000555881.5:c.83-1621G>T

ENST00000556908.5:c.212G>T

ENST00000557065.1:c.156-592G>T

NM_001135058.1:c.260G>T

NM_004086.2:c.260G>T

NR_038356.1:n.1618-2279C>A

NM_001347720.1:c.455G>T

NM_001135058.2:c.260G>T

NM_001347720.2:c.455G>T

Likely Pathogenic

Met criteria codes **4**

PP1_Moderate

PM2_Supporting

PP3

PP4

Evidence Links **0**

Expert Panel

Hearing Loss VCEP [↗](#)

Criteria Specification Information

- [Criteria Specification: ClinGen Hearing Loss Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for CDH23, COCH, GJB2, KCNQ4, MYO6, MYO7A, SLC26A4, TECTA and USH2A Version 2.0](#)
- [PDF](#)
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Hearing Loss VCEP

The variant NM_004086.3:c.260G>T in COCH is a missense variant predicted to cause substitution of Glycine to Valine at amino acid 87 (p.Gly87Val). This variant was not found in gnomAD v4.1.1 (PM2_Supporting met). The in silico predictor REVEL yields a score of 0.867 (PP3 met). This variant was identified in two probands (PS4_Supporting met), including one from an East Asian family reported in two publications (HL-VCEP internal data, PMID:23993205, 23767834), in which it segregated with autosomal dominant, progressive, late-onset HL and concomitant vestibular dysfunction in 4 affected family members (PP1_Moderate met). Given the highly specific phenotype of autosomal dominant progressive HL with vestibular dysfunction, the HL-VCEP decided to apply PP4 evidence criteria. A different missense variant c.259G>T (p.Gly87Trp) at the same amino acid position has been classified as pathogenic by the ClinGen HL-VCEP (PM5 met). In summary, based on the current evidence (PM5, PP1_Moderate, PS4_Supporting, PP4, PP3 & PM2_Supporting), the variant NM_004086.3(COCH):c.260G>T (p.Gly87Val) is classified as likely pathogenic for autosomal dominant non-syndromic SNHL based on the ACMG/AMP criteria applied, as specified by the ClinGen HL-VCEP (ClinGen Hearing Loss VCEP specifications version 2; 09/16/2026).

Met criteria codes

PP1_Moderate			East Asian family reported in two publications (PMID:23993205, 23767834), in which variant segregated with autosomal dominant, progressive, late-onset HL and concomitant vestibular dysfunction in 4 affected family members
PM2_Supporting			This variant was not found in gnomAD v4.1.1.
PP3			The in silico predictor REVEL yields a score of 0.867.
PP4			Given the highly specific phenotype of autosomal dominant progressive HL with vestibular dysfunction, the HL-VCEP decided to apply PP4 evidence criteria.

Curation History [↗](#)

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