

Variant: *NM_004086.3(COCH):c.260G>C (p.Gly87Ala)*

Version: 1.0

CA389344544 [↗](#)

517362 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b17ae703-89d3-413b-b547-8e1701d4e055

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HGVS expressions

NM_004086.3(COCH):c.260G>C (p.Gly87Ala)

NM_004086.3:c.260G>C

NC_000014.9:g.30878831G>C

CM000676.2:g.30878831G>C

NC_000014.8:g.31348037G>C

CM000676.1:g.31348037G>C

NC_000014.7:g.30417788G>C

NG_008211.2:g.9297G>C

ENST00000216361.9:c.455G>C

ENST00000396618.9:c.260G>C

ENST00000555117.2:c.260G>C

ENST00000643575.1:c.260G>C

ENST00000643697.1:n.505G>C

ENST00000644874.2:c.260G>C

ENST00000216361.8:c.260G>C

ENST00000396618.7:c.260G>C

ENST00000460581.6:c.-77G>C

ENST00000475087.5:c.260G>C

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

ENST00000553833.5:n.414G>C

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

ENST00000556908.5:c.212G>C

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

NM_001135058.1:c.260G>C

NM_004086.2:c.260G>C

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

NM_001347720.1:c.455G>C

NM_001135058.2:c.260G>C

NM_001347720.2:c.455G>C

Likely Pathogenic

Met criteria codes **5**

PP1 PP3 PP4 PM5_Strong

PM2_Supporting

Not Met criteria codes **1**

Expert Panel

Hearing Loss VCEP [↗](#)

Criteria Specification Information

PS4

Evidence Links 0

- [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>C in COCH is a missense variant predicted to cause substitution of Glycine to Alanine at amino acid 87 (p.Gly87Ala). This variant was not found in gnomAD v4.1.1 (PM2_Supporting met). The in silico predictor REVEL yields a score of 0.793 (PP3 met). This variant was identified in a family (Laboratory for Molecular Medicine internal data, SCV000731610.2), in which it segregated with autosomal dominant, progressive, late-onset HL and concomitant vestibular dysfunction in two affected family members (PP1 met). Given the highly specific phenotype of autosomal dominant, progressive HL with vestibular dysfunction, the HL-VCEP decided to apply PP4 evidence criteria. Two different missense variant c.259G>T (p.Gly87Trp) and c.260G>T (p.Gly87Val) at the same amino acid position have been classified Likely-Pathogenic by the ClinGen HL-VCEP (PM5_Strong met). In summary, based on the current evidence (PM5_Strong, PP1, PP4, PP3 & PM2_Supporting), the variant NM_004086.3(COCH):c.260G>C (p.Gly87Ala) 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			This variant was identified in a family (Laboratory for Molecular Medicine internal data, SCV000731610.2), in which it segregated with autosomal dominant, progressive, late-onset HL and concomitant vestibular dysfunction in two affected family members.
PP3			The in silico predictor REVEL yields a score of 0.793.
PP4			Given the highly specific phenotype of autosomal dominant, progressive HL with vestibular dysfunction, the HL-VCEP decided to apply PP4 evidence criteria.
PM5_Strong			Two different missense variant c.259G>T (p.Gly87Trp) and c.260G>T (p.Gly87Val) at the same amino acid position have been classified Likely-Pathogenic by the ClinGen HL-VCEP.
PM2_Supporting			This variant was not found in gnomAD v4.1.1.

Not Met criteria codes

PS4			This variant was identified in only one family (Laboratory for Molecular Medicine internal data, SCV000731610.2), in which it segregated with autosomal dominant, progressive, late-onset HL and concomitant vestibular dysfunction.
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