

Variant: *NM_004004.6(GJB2):c.109G>T (p.Val37Phe)*

Version: 1.0

CA273605 [↗](#)

179256 (ClinVar) [↗](#)

Gene: GJB2 ([HGNC:2706](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8ce5aea1-5ed1-4071-bd6b-1a3d6b2f1821

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HGVS expressions

NM_004004.6(GJB2):c.109G>T (p.Val37Phe)

NM_004004.6:c.109G>T

NC_000013.11:g.20189473C>A

CM000675.2:g.20189473C>A

NC_000013.10:g.20763612C>A

CM000675.1:g.20763612C>A

NC_000013.9:g.19661612C>A

NG_008358.1:g.8503G>T

ENST00000382844.2:c.109G>T

ENST00000382848.5:c.109G>T

ENST00000382844.1:c.109G>T

ENST00000382848.4:c.109G>T

NM_004004.5:c.109G>T

Likely Pathogenic

Met criteria codes **4**

PP3 PM3 PM5 PM2_Supporting

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 NM_004004.6(GJB2):c.109G>T variant in GJB2 is a missense variant predicted to cause substitution of Valine to Phenylalanine at amino acid 37 (p.Val37Phe). This variant is absent from gnomAD v4.1.1 (PM2_Supporting met). The computational predictor REVEL yields a score of 0.828 (PP3 met).This variant has been detected in one individual with hearing loss in trans with a pathogenic c.167del

(p.Leu56Argfs*26) (Laboratory for Molecular Medicine internal data, Clinvar ID 179256) (1.0 point, PM3 met). One missense variant, p.Val37Ile (ClinVar ID 17023), in the same codon has been classified as pathogenic for autosomal recessive non syndromic hearing loss by the ClinGen HL-VCEP (PM5 met). In summary, based on the current evidence (PM2_Supporting, PM3, PM5, PP3), the variant NM_004004.6(GJB2):c.109G>T (p.Val37Phe) is classified as likely pathogenic for autosomal recessive non-syndromic HL based on the ACMG/AMP criteria applied, as specified by the ClinGen Hearing Loss VCEP. (ClinGen Hearing Loss VCEP specifications version 2; 8/19/2026).

Met criteria codes

PP3			The computational predictor REVEL yields a score of 0.828.
PM3			This variant has been detected in 1 individual with hearing loss, compound heterozygous for this variant and a loss of function variant (Laboratory for molecular medicine, Clinvar ID 179256) (PM3).
PM5			One missense variant, p.Val37Ile (ClinVar ID 17023), in the same codon has been classified as pathogenic for autosomal recessive non syndromic hearing loss by the ClinGen HL-VCEP.
PM2_Supporting			This variant is absent from gnomAD v4.1.1.

Curation History



 

 

Showing 1 to 1 of 1 rows

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