

Variant: *NM_022124.6(CDH23):c.5131G>A (p.Val1711Ile)*

Version: 1.0

CA245543 [↗](#)

197422 (ClinVar) [↗](#)

Gene: [CDH23](#)

Condition: Usher syndrome ([MONDO:0019501](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: fd061a57-91b5-49ca-b674-206970883363

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HGVS expressions

NM_022124.6:c.5131G>A

NM_022124.6(CDH23):c.5131G>A (p.Val1711Ile)

NC_000010.11:g.71778252G>A

CM000672.2:g.71778252G>A

NC_000010.10:g.73538009G>A

CM000672.1:g.73538009G>A

NC_000010.9:g.73208015G>A

NG_008835.1:g.386306G>A

ENST00000224721.12:c.5131G>A

ENST00000224721.10:c.5146G>A

ENST00000622827.4:c.5131G>A

NM_022124.5:c.5131G>A

Likely Benign

Met criteria codes **2**

BS1 **BP4**

Evidence Links **0**

Expert Panel

[Hearing Loss VCEP](#) [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Hearing Loss VCEP

The c.5131 (p.Val1711Ile) variant has been identified in 0.3% (51/19534 CI 95%) of East Asian alleles in gnomAD (BS1). This variant was observed in 1 patient with Usher syndrome, without a second variant in CDH23 (PMID: 25333064). Computational prediction analysis using the metapredictor tool REVEL produced a score of .17; this combined with the presence of the isoleucine amino acid at position 1711 in 5 organisms including 3 mammals in UCSC suggests that the variant may not impact the protein (BP4). In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Hearing Loss Expert Panel: BS1, BP4.

Met criteria codes

BS1



Present in .3% (51/19534 CI 95%) of South Asian alleles in gnomAD.

BP4



REVEL: .17, which does not meet the cut off for BP4 however the amino acid change is present in 5 organisms in UCSC including 3 mammals. After expert review BP4 was applied.

Curation History [↗](#)

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