

Variant: *NM\_000260.4(MYO7A):c.1997G>A (p.Arg666Gln)*

Version: 1.1

CA6197659 [↗](#)

556881 (ClinVar) [↗](#)

**Gene:** MYO7A ([HGNC:4647](#))

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

**Inheritance Mode:** Autosomal recessive inheritance

**UUID:** 4923dcaa-6018-41a1-b68f-39af89d9e3cf

**Approved on:** 2025/3/12

**Published on:** 2025/6/30

## HGVS expressions

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NC\_000011.10:g.77174817G>A

CM000673.2:g.77174817G>A

NC\_000011.9:g.76885863G>A

CM000673.1:g.76885863G>A

NC\_000011.8:g.76563511G>A

NG\_009086.1:g.51554G>A

NG\_009086.2:g.51572G>A

ENST00000409709.9:c.1997G>A

ENST00000409893.6:c.62G>A

ENST00000409619.6:c.1964G>A

ENST00000409709.7:c.1997G>A

ENST00000409893.5:c.1997G>A

ENST00000458637.6:c.1997G>A

ENST00000620575.4:c.1997G>A

NM\_000260.3:c.1997G>A

NM\_001127179.2:c.1997G>A

NM\_001127180.1:c.1997G>A

NM\_001127180.2:c.1997G>A

NM\_001369365.1:c.1964G>A

Uncertain Significance

Met criteria codes **1**

PP3

Not Met criteria codes **2**

PM2

BS1

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

[↗](#) PDF

[↗](#) Criteria Specification Approval History

[↗](#) Criteria Specifications for this VCEP

**Hearing Loss VCEP**

The c.1997G>A variant in MYO7A is a missense variant predicted to cause substitution of arginine by glycine at amino acid 666. The highest population minor allele frequency in gnomAD v4.1 is 0.0001865 (14/75048 alleles) in the African/African-American population (PM2\_Supporting and BS1\_Supporting are not met). The computational predictor REVEL gives a score of 0.841, which is above the threshold of 0.7, evidence that correlates with impact to MYO7A function (PP3). This variant has been observed in one proband with Usher syndrome, four probands with hearing loss, and two probands with retinal disease. However, they all harbored another variant of uncertain significance c.3527G>A (p.Ser1176Asn) with phase unknown, and two who were found to have an alternate molecular basis for disease (PM3\_Supporting not met; ClinVar ID: 196099; PMIDs: 27460420, 31479088, 33297549, 27344577, LabCorp Genetics (formerly Invitae) internal data ClinVar SCV001498602.2, GeneDx internal data). In summary, this variant has been classified as a variant of uncertain significance for autosomal recessive Usher syndrome based on the ACMG/AMP criteria applied, as specified by the ClinGen Hearing Loss VCEP: PP3 (ClinGen Hearing Loss VCEP specifications version 2; 3/12/2025).

**Met criteria codes**

|            |                                                                                   |                                                                                   |                                                                                                                                                                 |
|------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>PP3</b> |  |  | The computational predictor REVEL gives a score of 0.841, which is above the threshold of 0.7, evidence that correlates with impact to MYO7A function (PM3 met) |
|------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------|

**Not Met criteria codes**

|            |                                                                                     |                                                                                     |                                                                                                                                                                                          |
|------------|-------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>PM2</b> |                                                                                     |  | The highest population minor allele frequency in gnomAD v4.1 is 0.0001865 (14/75048 alleles) in the African/African-American population (PM2_Supporting and BS1_Supporting are not met). |
| <b>BS1</b> |  |  | The highest population minor allele frequency in gnomAD v4.1 is 0.0001865 (14/75048 alleles) in the African/African-American population (PM2_Supporting and BS1_Supporting are not met). |

Curation History 



 

 

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