

Variant: *NM_206933.3(USH2A):c.7167C>T (p.Ser2389=)*

Version: 1.0

CA1394924 [↗](#)

681822 (ClinVar) [↗](#)

Gene: USH2A ([HGNC:7399](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 0f97b706-2a00-4fce-8bbe-6744fe0a63bc

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HGVS expressions

NM_206933.3(USH2A):c.7167C>T (p.Ser2389=)

NM_206933.3:c.7167C>T

NC_000001.11:g.215934749G>A

CM000663.2:g.215934749G>A

NC_000001.10:g.216108091G>A

CM000663.1:g.216108091G>A

NC_000001.9:g.214174714G>A

NG_009497.1:g.493648C>T

NG_009497.2:g.493700C>T

ENST00000307340.8:c.7167C>T

ENST00000674083.1:c.7167C>T

ENST00000307340.7:c.7167C>T

NM_206933.2:c.7167C>T

NM_206933.4:c.7167C>T

Likely Benign

Met criteria codes **3**

BS1_Supporting

BP4

BP7

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 filtering allele frequency (the lower threshold of the 95% CI of 57/19928 of the c.7167C>T (p.Ser2389=) variant in the USH2A gene is 0.22% for East Asian chromosomes by gnomAD v2.1.1, which is a higher frequency than would be expected for an autosomal recessive pathogenic variant based on the thresholds defined by the ClinGen Hearing Loss Expert Panel (BS1_Supporting). The silent c.7167C>T variant in USH2A is not predicted by the computational prediction analysis using MaxEntScan to impact splicing (BP4, BP7). 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_P, BP4, BP7.**

Met criteria codes

BS1_Supporting			The filtering AF (95% CI) is 0.22% (57/19928) of East Asian chromosomes in gnomAD v2.1.1 and 0.22% (12/3114) of South Asian chromosomes in gnomAD v3.
BP4			Residue is not highly conserved and no splicing impact predicted by MaxEntScan and other splicing predictors in Alamut.
BP7			no splicing impact predicted by MaxEntScan and other splicing predictors in Alamut.

Curation History



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