

Variant: *NM_206933.4(USH2A):c.14419G>A (p.Ala4807Thr)*

Version: 1.2

[CA246980](#)

[198366 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UID: c1d605e5-67f2-43d5-8643-a70536da96b7

Approved on: 2024-02-21

Published on: 2024-04-01

HGVS expressions

NM_206933.4:c.14419G>A

NM_206933.4(USH2A):c.14419G>A (p.Ala4807Thr)

NC_000001.11:g.215648691C>T

CM000663.2:g.215648691C>T

NC_000001.10:g.215822033C>T

CM000663.1:g.215822033C>T

NC_000001.9:g.213888656C>T

NG_009497.1:g.779706G>A

NG_009497.2:g.779758G>A

ENST00000307340.8:c.14419G>A

ENST00000674083.1:c.14419G>A

ENST00000307340.7:c.14419G>A

NM_206933.2:c.14419G>A

NM_206933.3:c.14419G>A

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

PM6 PVS1 PM1 PM3 PM5
PM4 BS1 BS4 BS3 BS2
BP5 BP7 BP4 BP3 BP1 BP2
PS1 PS2 PS3 PS4 BA1 PP1
PP2 PP3 PP4

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

Evidence submitted by expert panel

Hearing Loss VCEP



The variant **NM_206933.4:c.14419G>A** in **USH2A** is a missense variant predicted to cause substitution of alanine by threonine at amino acid 4807 (p.Ala4807Thr). The filtering allele frequency (the upper threshold of the 95% CI of 5/30782) of the c.14419G>A variant in USH2A is



0.00006365 for South Asian chromosomes by gnomAD v2.1.1, which is lower than the ClinGen Hearing Loss VCEP threshold (<0.00007) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting). In summary, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Hearing Loss Expert Panel: PM2_Supporting (ClinGen Hearing Loss VCEP specifications version 2; 2/21/2024).



Met criteria codes



PM2_Supporting   ExAC MAF of 0.03% in South Asian. 0.016% in gnomAD. <http://gnomad.broadinstitute.org/variant/1-215822033-C-T>



Not Met criteria codes



PM6   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PVS1   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline



PM1   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline


PM3   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline



PM5   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline



PM4   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline


BS1   ExAC MAF of 0.03% in South Asian. 0.016% in gnomAD. <http://gnomad.broadinstitute.org/variant/1-215822033-C-T>

BS4   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

























BS3  No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS2   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP5   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP7  

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			ExAC MAF of 0.03% in South Asian. 0.016% in gnomAD. http://gnomad.broadinstitute.org/variant/1-215822033-C-T
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

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