

Variant: NM_000260.3(MYO7A):c.324C>T (p.Tyr108=)

CA132282 [↗](#)

43201 (ClinVar) [↗](#)

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

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

UUID: 8f1b935c-2f47-463c-987b-93081430a581

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HGVS expressions

NM_000260.3:c.324C>T

NM_000260.3(MYO7A):c.324C>T (p.Tyr108=)

NC_000011.10:g.77155945C>T

CM000673.2:g.77155945C>T

NC_000011.9:g.76866991C>T

CM000673.1:g.76866991C>T

NC_000011.8:g.76544639C>T

NG_009086.1:g.32682C>T

NM_001127179.2:c.324C>T

NM_001127180.1:c.324C>T

NM_000260.4:c.324C>T

ENST00000409619.6:c.291C>T

ENST00000409709.7:c.324C>T

ENST00000409893.5:c.324C>T

ENST00000458637.6:c.324C>T

ENST00000620575.4:c.324C>T

Benign

Met criteria codes 1

BA1

Not Met criteria codes 21

BS4 BS1 BS2 BP5 BP2 BP3
BP4 PVS1 PS2 PS4 PS3
PS1 PP4 PP1 PP3 PM3
PM1 PM4 PM5 PM6 PM2

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 of the p.Tyr108= variant in the MYO7A gene is 1.45% (300/18658) of East Asian chromosomes by the Genome Aggregation Database (<http://gnomad.broadinstitute.org>; calculated by using inverse allele frequency at

https://www.cardiodb.org/allelefrequencyapp/), which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen Hearing Loss Expert Panel for autosomal dominant hearing loss variants (BA1).

Met criteria codes

BA1			ExAC filtered allele frequency 1.76% from East Asian population
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Not Met criteria codes

BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			ExAC filtered allele frequency 1.76% from East Asian population
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
BP2			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
BP4			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
PS2			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
PS3			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

PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			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
PM3			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
PM4			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
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			ExAC filtered allele frequency 1.76% from East Asian population

Curation History 		
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