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Variant: NM_005422.2(TECTA):c.5836T>C (p.Tyr1946His)

Version: 1.0

CA178113

165370 (ClinVar)

Gene: TECTA (HGNC:7007)
Condition: nonsyndromic genetic deafness (MONDO:0019497)
UUID: 4b0e8b50-666a-4b08-ba95-8bb5daecfb6d
Approved on: 2019-01-28
Published on: 2019-07-17

HGVS expressions

NM_005422.2(TECTA):c.5836T>C (p.Tyr1946His)
NM_005422.2:c.5836T>C
NM_005422.2:n.5836T>C
ENST00000264037.2:n.5836T>C
ENST00000392793.5:c.5836T>C
NC_000011.10:g.121168762T>C
CM000673.2:g.121168762T>C
NC_000011.9:g.121039471T>C
CM000673.1:g.121039471T>C
NC_000011.8:g.120544681T>C
NG_011633.1:g.71097T>C

Likely Benign

The Expert Panel has overridden the computationally generated classification - "*Uncertain Significance - Conflicting Evidence*"

Met criteria codes 2

BS1 PP3

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 c.5836T>C (p.Tyr1946His) variant in the TECTA gene is 0.495% for Ashkenazi Jewish chromosomes by gnomAD (64/10370 with 95% CI), which is a high enough frequency to be classified as likely benign based on the thresholds defined by the ClinGen Hearing Loss Expert Panel (HL EP) for autosomal recessive hearing loss variants (BS1). The REVEL computational prediction analysis tool produced a score of 0.8, which is above the threshold necessary to apply PP3. The HL EP allows classification of variants as likely benign with only BS1 if no other criteria are in conflict. The HL EP reviewed the conflicting evidence (PP3) and felt it did not override the Likely Benign classification in this case since computational scores are error prone, especially when predicting pathogenicity. In summary, the HL EP classified this variant as likely benign. ACMG/AMP criteria applied, as specified by the Hearing Loss Expert Panel: BS1, PP3.

Met criteria codes

BS1		✔	Filtering allele frequency 0.49% in AJ population in gnomAD
PP3		✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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