

Variant: *NM_004086.2(COCH):c.429A>G (p.Pro143=)*

Version: 1.0

CA7143032 [↗](#)

227258 (ClinVar) [↗](#)

Gene: COCH ([HGNC:1690](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a9f35732-d723-466d-a748-67465ee0c69d

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HGVS expressions

NM_004086.2:c.429A>G

NM_004086.2(COCH):c.429A>G (p.Pro143=)

NC_000014.9:g.30879478A>G

CM000676.2:g.30879478A>G

NC_000014.8:g.31348684A>G

CM000676.1:g.31348684A>G

NC_000014.7:g.30418435A>G

NG_008211.2:g.9944A>G

ENST00000216361.9:c.624A>G

ENST00000396618.9:c.429A>G

ENST00000555117.2:c.429A>G

ENST00000643575.1:c.429A>G

ENST00000643697.1:n.674A>G

ENST00000644874.2:c.429A>G

ENST00000216361.8:c.429A>G

ENST00000396618.7:c.429A>G

ENST00000460581.6:c.93A>G

ENST00000475087.5:c.429A>G

ENST00000553772.5:c.240-974A>G

ENST00000553833.5:n.583A>G

ENST00000555881.5:c.83-974A>G

ENST00000556908.5:c.381A>G

ENST00000557065.1:c.211A>G

NM_001135058.1:c.429A>G

NR_038356.1:n.1618-2926T>C

NM_001347720.1:c.624A>G

NM_004086.3:c.429A>G

NM_001135058.2:c.429A>G

NM_001347720.2:c.624A>G

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

Expert Panel

Hearing Loss VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

Hearing Loss VCEP

The filtering allele frequency (the lower threshold of the 95% CI of 44/24026) of the c.429A>G (p.Pro143=) variant in the COCH gene is 0.14% for African chromosomes by gnomAD, 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			filtering allele frequency 0.14% (44/24026) of African chromosomes in gnomad, over BA1 cutoff of 0.1% for AD hearing loss
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Curation History [↗](#)



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