

Variant: *NM_025114.4(CEP290):c.2067G>A (p.Lys689=)*

Version: 1.0

CA481050498 [↗](#)

595539 (ClinVar) [↗](#)

Gene: CEP290 ([HGNC:80184](#))

Condition: CEP290-related ciliopathy ([MONDO:0100451](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 3f6ed27c-ff95-4da1-830e-ae5e736d8ad7

Approved on: 2026/7/20

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HGVS expressions

NM_025114.4(CEP290):c.2067G>A (p.Lys689=)

NM_025114.4:c.2067G>A

NC_000012.12:g.88111844C>T

CM000674.2:g.88111844C>T

NC_000012.11:g.88505621C>T

CM000674.1:g.88505621C>T

NC_000012.10:g.87029752C>T

NG_008417.1:g.35373G>A

NG_008417.2:g.35373G>A

ENST00000309041.12:c.2067G>A

ENST00000547926.7:c.1924G>A

ENST00000552810.6:c.2067G>A

ENST00000671822.2:n.3623G>A

ENST00000672414.2:c.*238G>A

ENST00000673058.2:c.2067G>A

ENST00000674971.1:c.2067G>A

ENST00000675230.1:c.2046G>A

ENST00000675408.1:c.2067G>A

ENST00000675476.1:c.2928G>A

ENST00000675628.1:n.2294G>A

ENST00000675794.1:c.*238G>A

ENST00000675833.1:c.2835G>A

ENST00000676074.1:c.2067G>A

ENST00000676363.1:n.4130G>A

ENST00000676448.1:c.1924G>A

ENST00000309041.11:c.2073G>A

ENST00000397838.7:c.2073G>A

ENST00000547926.6:c.1822G>A

ENST00000552810.5:c.2067G>A

ENST00000604024.5:c.1326G>A

NM_025114.3:c.2067G>A

Met criteria codes **3**

Likely Benign

BP7

BP4

PM2

Expert Panel

Leber Congenital Amaurosis/early onset Retinal Dystrophy VCEP [↗](#)

[Criteria Specification:](#) *ClinGen Leber Congenital Amaurosis/early onset Retinal Dystrophy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for CEP290 Version 1.0*

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Leber Congenital Amaurosis/early onset Retinal Dystrophy VCEP

NM_025114.4(CEP290):c.2067G>A (p.Lys689=) is a synonymous variant in exon 21 that does not have predicted impact on CEP290 splicing (BP7). The splicing impact predictor SpliceAI gives a delta score of 0.03 for acceptor gain, which is below the ClinGen LCA / eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4). This variant is present in gnomAD v4.1.1 at a total allele frequency of 0.00000185 with 6 alleles / 1,592,018 total alleles, which is lower than the ClinGen LCA/eoRD VCEP PM2_Supporting threshold of <0.0006 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Likely Benign for CEP290-related ciliopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen LCA/eoRD VCEP: BP7, BP4, and PM2_Supporting. (LCA/eoRD VCEP Specifications for CEP290 Version 1.0.0)

Met criteria codes

BP7			NM_025114.4(CEP290):c.2067G>A (p.Lys689=) is a synonymous variant in exon 21 that is not predicted to disrupt CEP290 splicing (BP7).
BP4			The splicing impact predictor SpliceAI gives a delta score of 0.03 for acceptor gain, which is below the ClinGen LCA / eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4).
PM2			This variant is present in gnomAD v4.1.0 at a total allele frequency of 0.00000185 with 6 alleles / 1,592,018 total alleles, which is lower than the ClinGen LCA/eoRD VCEP PM2_Supporting threshold of <0.0006 (PM2_Supporting).

Curation History [↗](#)



 

 

Showing 1 to 1 of 1 rows

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