

Variant: *NM_014336.5(AIPL1):c.304C>T (p.Arg102Trp)*

Version: 1.0

CA8328546 [↗](#)

1385342 (ClinVar) [↗](#)

Gene: AIPL1 ([HGNC:23746](#))

Condition: AIPL1-related retinopathy ([MONDO:0100438](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: aabf615f-2f92-4f27-8e52-f9638d7bf7b5

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HGVS expressions

NM_014336.5(AIPL1):c.304C>T (p.Arg102Trp)

NM_014336.5:c.304C>T

NC_000017.11:g.6428479G>A

CM000679.2:g.6428479G>A

NC_000017.10:g.6331799G>A

CM000679.1:g.6331799G>A

NC_000017.9:g.6272523G>A

NG_008474.1:g.11721C>T

ENST00000381129.8:c.304C>T

ENST00000250087.9:c.277-1422C>T

ENST00000381128.2:c.*176C>T

ENST00000381129.7:c.304C>T

ENST00000570466.5:c.238C>T

ENST00000570584.5:c.251+5440C>T

ENST00000571740.5:c.304C>T

ENST00000574506.5:c.268C>T

ENST00000574913.1:c.304C>T

ENST00000575265.5:c.304C>T

ENST00000576307.5:c.124C>T

ENST00000576776.5:c.304C>T

ENST00000621374.4:c.304C>T

NM_001033054.2:c.277-1422C>T

NM_001033055.2:c.124C>T

NM_001285399.2:c.268C>T

NM_001285400.2:c.238C>T

NM_001285401.2:c.304C>T

NM_001285402.1:c.187C>T

NM_001285403.2:c.304C>T

NM_014336.4:c.304C>T

NM_001033054.3:c.277-1422C>T

NM_001033055.3:c.124C>T

NM_001285399.3:c.268C>T

NM_001285400.3:c.238C>T

NM_001285401.3:c.304C>T

NM_001285402.2:c.187C>T

NM_001285403.3:c.304C>T

NM_001285403.4:c.304C>T

Uncertain Significance

Met criteria codes 2

PP3 PM2_Supporting

Evidence Links 0

Expert Panel

Leber Congenital Amaurosis/early onset Retinal Dystrophy VCEP

Criteria Specification Information

- Criteria Specification: ClinGen Leber Congenital Amaurosis/early onset Retinal Dystrophy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for AIPL1 Version 1.0.0
- Criteria Specification Approval History
- Criteria Specifications for this VCEP

Evidence submitted by expert panel

Leber Congenital Amaurosis/early onset Retinal Dystrophy VCEP

NM_014336.5(AIPL1):c.304C>T (p.Arg102Trp) is a missense variant replacing the arginine at position p.102 with tryptophan. This variant is present in gnomAD v.4.1.0 at a total allele frequency of 0.000021, with 34 alleles / 1613846 total alleles, which is lower than the ClinGen LCA/eoRD VCEP PM2_Supporting threshold of <0.0004 (PM2_Supporting). The computational predictor REVEL gives a score of 0.687, which is above the ClinGen LCA/eoRD VCEP threshold of ≥0.644 and predicts a damaging effect on AIPL1 protein function (PP3). In summary, this variant meets the criteria to be classified as a VUS for AIPL1-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen LCA/eoRD VCEP: PM2_Supporting and PP3. (VCEP specifications version 1.0.0; date of approval 09/24/2025).

Met criteria codes

PP3		✓	The computational predictor REVEL gives a score of 0.687, which is above the ClinGen LCA/eoRD VCEP threshold of ≥0.644 and predicts a damaging effect on AIPL1 protein function (PP3).
PM2_Supporting		✓	This variant is present in gnomAD v.4.1.0 at a total allele frequency of 0.000021, with 34 alleles / 1613846 total alleles, which is lower than the ClinGen LCA/eoRD VCEP PM2_Supporting threshold of <0.0004 (PM2_Supporting).

Curation History

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