

Variant: *NM_014336.5(AIPL1):c.937G>T (p.Ala313Ser)*

Version: 1.0

CA228922 [↗](#)

100585 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: fa972f40-ef7a-4cf3-85c9-8dd3f11a2a84

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HGVS expressions

NM_014336.5:c.937G>T

NM_014336.5(AIPL1):c.937G>T (p.Ala313Ser)

NC_000017.11:g.6425678C>A

CM000679.2:g.6425678C>A

NC_000017.10:g.6328998C>A

CM000679.1:g.6328998C>A

NC_000017.9:g.6269722C>A

NG_008474.1:g.14522G>T

ENST00000381129.8:c.937G>T

ENST00000250087.9:c.748G>T

ENST00000381128.2:c.*809G>T

ENST00000381129.7:c.937G>T

ENST00000570466.5:c.871G>T

ENST00000570584.5:c.251+8241G>T

ENST00000574506.5:c.901G>T

ENST00000575265.5:c.*908G>T

ENST00000576307.5:c.757G>T

ENST00000576776.5:c.865G>T

ENST00000621374.4:c.936G>T

NM_001033054.2:c.748G>T

NM_001033055.2:c.757G>T

NM_001285399.2:c.901G>T

NM_001285400.2:c.871G>T

NM_001285401.2:c.865G>T

NM_001285402.1:c.820G>T

NM_014336.4:c.937G>T

NM_001033054.3:c.748G>T

NM_001033055.3:c.757G>T

NM_001285399.3:c.901G>T

NM_001285400.3:c.871G>T

NM_001285401.3:c.865G>T

NM_001285402.2:c.820G>T

NM_001285403.3:c.*908G>T

NM_001285403.4:c.*908G>T

Benign

Met criteria codes 3

BS2BP4BA1

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.937G>T (p.Ala313Ser) is a missense variant in exon 6 of 6. The variant is predicted to change the amino acid alanine at position p.313 to serine. This variant is present in gnomAD v.4.1.0 at a GrpMax allele frequency of 0.01581, with 1,244 alleles / 75,040 total alleles in the African/African American population, which is higher than the ClinGen LCA/eoRD VCEP BA1 threshold of >0.0057 (BA1). The variant has been found in the homozygous state in 16 adult individuals in gnomAD which exceeds the LCA/eoRD VCEP threshold of ≥6 (gnomAD version 4.1.0; BS2). The computational predictor REVEL gives a score of 0.069, which is below the ClinGen LCA/eoRD VCEP threshold of ≤0.183 and predicts a non-damaging effect on AIPL1 protein function. In addition, the phyloP score is 1.28882 (suggesting low conservation) and the splicing impact predictor SpliceAI gives a delta score of 0, which is below the ClinGen LCA/eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4_Moderate). In summary, this variant meets the criteria to be classified as Benign for AIPL1-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen LCA/eoRD VCEP: BA1, BS2, and BP4_Moderate. (VCEP specifications version 1.0.0; date of approval 09/24/2025).

Met criteria codes

BS2

i

✓

This variant has been found in the homozygous state in 16 adult individuals in gnomAD which exceeds the LCA/eoRD VCEP threshold of ≥6 (gnomAD version 4.1.0; BS2).

BP4

i

✓

The computational predictor REVEL gives a score of 0.069, which is below the ClinGen LCA/eoRD VCEP threshold of ≤0.183 and predicts a non-damaging effect on AIPL1 protein function. In addition, the phyloP score is 1.28882 suggesting low conservation and the splicing impact predictor SpliceAI gives a delta score of 0, which is below the ClinGen LCA/eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4_Moderate).

BA1

i

✓

This variant is present in gnomAD v.4.1.0 at a GrpMax allele frequency of 0.01581, with 1,244 alleles / 75,040 total alleles in the African/African American population, which is higher than the ClinGen LCA/eoRD VCEP BA1 threshold of >0.0057 (BA1).

Curation History

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