

Variant: *NM_014336.5(AIPL1):c.111C>T (p.Phe37=)*

Version: 1.0

CA227864 [↗](#)

99789 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 02a43879-4d81-4514-8a4a-c12b5ef8b8aa

Approved on: 2025/9/29

Published on: 2025/9/29

HGVS expressions

NM_014336.5:c.111C>T

NM_014336.5(AIPL1):c.111C>T (p.Phe37=)

NC_000017.11:g.6434084G>A

CM000679.2:g.6434084G>A

NC_000017.10:g.6337404G>A

CM000679.1:g.6337404G>A

NC_000017.9:g.6278128G>A

NG_008474.1:g.6116C>T

ENST00000381129.8:c.111C>T

ENST00000250087.9:c.111C>T

ENST00000381128.2:c.169C>T

ENST00000381129.7:c.111C>T

ENST00000570466.5:c.97-52C>T

ENST00000570584.5:c.86C>T

ENST00000571740.5:c.111C>T

ENST00000574506.5:c.97-22C>T

ENST00000574913.1:c.111C>T

ENST00000575265.5:c.111C>T

ENST00000576307.5:c.96+925C>T

ENST00000576776.5:c.111C>T

ENST00000621374.4:c.111C>T

NM_001033054.2:c.111C>T

NM_001033055.2:c.96+925C>T

NM_001285399.2:c.97-22C>T

NM_001285400.2:c.97-52C>T

NM_001285401.2:c.111C>T

NM_001285402.1:c.-7C>T

NM_001285403.2:c.111C>T

NM_014336.4:c.111C>T

NM_001033054.3:c.111C>T

NM_001033055.3:c.96+925C>T

NM_001285399.3:c.97-22C>T

NM_001285400.3:c.97-52C>T

NM_001285401.3:c.111C>T

NM_001285402.2:c.-7C>T

NM_001285403.3:c.111C>T

NM_001285403.4:c.111C>T

Benign

Met criteria codes 4

BA1 BS2 BP4 BP7

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.111C>T (p.Phe37=) is a synonymous variant in exon 2 of 6. The change from C to T at c.111 is not predicted to change the amino acid phenylalanine at position p.37. This variant is present in gnomAD v.4.1.0 at a Grpmax allele frequency of 0.03109, with 37005 alleles / 1,179,952 total alleles in the European (non-Finnish) population, which is higher than the ClinGen LCA/eoRD VCEP BA1 threshold of >0.0057 (BA1). Additionally, the variant has been found in the homozygous state in 748 adult individuals in gnomAD which exceeds the LCA/eoRD VCEP threshold of ≥6 (gnomAD version 4.1.0; BS2). The splicing impact predictor SpliceAI gives a delta score of 0.08, which is below the ClinGen LCA/eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4, BP7). 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, BP4, and BP7. (VCEP specifications version 1.0.0; date of approval 09/24/2025).

Met criteria codes

BA1			This variant is present in gnomAD v.4.1.0 at a Grpmax allele frequency of 0.03109, with 37005 alleles /1179952 total alleles in the European (non-Finnish) population, which is higher than the ClinGen LCA/eoRD VCEP BA1 threshold of >0.0057 (BA1).
BS2			This variant has been found in the homozygous state in 748 adult individuals in gnomAD which exceeds the LCA/eoRD VCEP threshold of ≥6 (gnomAD version 4.1.0; BS2)
BP4			The splicing impact predictor SpliceAI gives a delta score of 0.08, which is below the ClinGen LCA/eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing (BP4).
BP7			The splicing impact predictor SpliceAI gives a delta score of 0.08, which is below the ClinGen LCA/eoRD VCEP recommended threshold of <0.1 and does not predict an impact on splicing and the nucleotide has a phyloP score of 2.35057 and is not predicted to be highly conserved (BP4, BP7).

Curation History

▾

▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.