Variant: NM_000330.4(RS1):c.150G>T (p.Trp50Cys)

Version: 1.0

CA10360804

1164535 (ClinVar)

Gene: RS1 (HGNC:6247)
Condition: X-linked retinoschisis (MONDO:0010725)
Inheritance Mode: X-linked inheritance
UUID: 544f0bd6-7c0f-4dee-aa2e-264746b632dd
Approved on: 2025-05-19
Published on: 2025-05-20

HGVS expressions

NM_000330.4:c.150G>T
NM_000330.4(RS1):c.150G>T (p.Trp50Cys)
NC_000023.11:g.18656687C>A
CM000685.2:g.18656687C>A
NC_000023.10:g.18674807C>A
CM000685.1:g.18674807C>A
NC_000023.9:g.18584728C>A
NG_008659.3:g.25762G>T
ENST00000379984.4:c.150G>T
ENST00000379984.3:c.150G>T
NM_000330.3:c.150G>T

Likely Benign

Met criteria codes 1

BS1

Not Met criteria codes 2

BP4 **PP3**

Evidence Links 0

Expert Panel

X-linked Inherited Retinal Disease VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen X-linked Inherited Retinal Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RS1 Version 1.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

X-linked Inherited Retinal Disease VCEP

The NM_000330.4(RS1):c.150G>T variant is a missense variant encoding the substitution of Tryptophan with Cysteine at amino acid 50. This variant is present in gnomAD v.4.1.0 at a frequency of 0.0001765 among hemizygous individuals, with 70 variant alleles / 396581 total alleles, which is higher than the ClinGen X-linked IRD VCEP BS1 threshold of >>0.00002 (BS1). The computational predictor REVEL gives a score of 0.625, which is between the ClinGen X-linked IRD VCEP thresholds of >0.664 and <0.290 and does not predict a damaging effect on RS1 function. Additionally, the splicing impact predictor SpliceAI gives a score of 0.05, which is below the ClinGen X-linked IRD VCEP recommended threshold of ≥0.2 and does not strongly predict an impact on splicing. Collectively, the BP4 and PP3 codes do not apply. In

summary, this variant is classified as likely benign for X-linked retinoschisis based on the ClinGen X-linked Inherited Retinal Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RS1 Version 1.0.0: BS1. (date of approval 01/24/2025).

Met criteria codes

BS1	✓	This variant is present in gnomAD v.4.1.0 at a frequency of 0.0001765 among hemizygous individuals, with 70 variant alleles / 396581 total alleles, which is higher than the ClinGen X-linked IRD VCEP BS1 threshold of >>0.00002 (BS1).

Not Met criteria codes

BP4	✗	The computational predictor REVEL gives a score of 0.625, which is between the ClinGen X-linked IRD VCEP threshold of 0.664 to 0.290 and does not predict a damaging effect on RS1 function. Additionally, the splicing impact predictor SpliceAI gives a score of 0.05, which is below the ClinGen X-linked IRD VCEP recommended threshold of ≥0.2 and does not strongly predict an impact on splicing. Collectively, both BP4 and PP3 codes do not apply.
PP3	✗	The computational predictor REVEL gives a score of 0.625, which is between the ClinGen X-linked IRD VCEP threshold of 0.664 to 0.290 and does not predict a damaging effect on RS1 function. Additionally, the splicing impact predictor SpliceAI gives a score of 0.05, which is below the ClinGen X-linked IRD VCEP recommended threshold of ≥0.2 and does not strongly predict an impact on splicing. Collectively, both BP4 and PP3 codes do not apply.

Curation History

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