

Variant: *NM_000330.4(RS1):c.330T>A (p.Cys110Ter)*

Version: 1.0

CA226688 [↗](#)

98939 (ClinVar) [↗](#)

Gene: RS1 ([HGNC:6247](#))

Condition: X-linked retinoschisis ([MONDO:0010725](#))

Inheritance Mode: X-linked inheritance (recessive (HP:0001419))

UUID: 8a105601-4a7a-49d8-b79f-28645a57a26a

Approved on: Sat, 26 Sep 2026 00:00:00 +0000

Published on: Sat, 26 Sep 2026 00:00:00 +0000

HGVS expressions

NM_000330.4:c.330T>A

NM_000330.4(RS1):c.330T>A (p.Cys110Ter)

NC_000023.11:g.18644622A>T

CM000685.2:g.18644622A>T

NC_000023.10:g.18662742A>T

CM000685.1:g.18662742A>T

NC_000023.9:g.18572663A>T

NG_008475.1:g.224018A>T

NG_008659.3:g.37827T>A

ENST00000379984.4:c.330T>A

ENST00000379984.3:c.330T>A

ENST00000379989.6:c.2714-1385A>T

ENST00000379996.7:c.2714-1385A>T

ENST00000476595.1:n.821T>A

NM_000330.3:c.330T>A

NM_001037343.1:c.2714-1385A>T

NM_003159.2:c.2714-1385A>T

NM_001037343.2:c.2714-1385A>T

NM_003159.3:c.2714-1385A>T

Likely Pathogenic

Met criteria codes **3**

PP1

PM2_Supporting

PVS1_Strong

Not Met criteria codes **1**

PS4

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*

[↗](#) **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.330T>A variant is a nonsense variant in amino acid 110, which results in a premature stop codon and initiates the nonsense-mediated decay pathway. This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting). This is a nonsense variant that introduces a premature stop codon between amino acids 1-223 that is predicted to either trigger nonsense-mediated decay or to disrupt a critical C-terminal region required for proper multimerization of RS1 (PVS1, PMID: 19849666). The variant has been reported to segregate with retinal dystrophy through 1 meiosis in a family [two affected brothers]. (PP1; PMID: 30652005). This variant has been reported in at least 1 proband meeting the PS4 requirement of retinoschisis (PMID: 10636421, 30652005). PS4_Supporting requires at least 2 unrelated probands, so this criterion was not met. In summary, this variant is classified as pathogenic 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: PM2_supporting, PVS1, and PP1.

Met criteria codes

PP1			The variant has been reported to segregate with retinal dystrophy through 1 meiosis in a family [two affected brothers]. (PP1; PMID: 30652005).
PM2_Supporting			This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting).
PVS1_Strong			This is a nonsense variant that introduces a premature stop codon between amino acids 1-223 that is predicted to either trigger nonsense-mediated decay or to disrupt a critical C-terminal region required for proper multimerization of RS1 (PVS1, PMID: 19849666).

Not Met criteria codes

PS4			This variant has been reported in at least 1 proband meeting the PS4 requirement of retinoschisis (PMID: 10636421, 30652005). PS4_Supporting requires at least 2 unrelated probands, so this criterion was not met.
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Curation History

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