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Variant: NM_000330.4:c.16G>T

Version: 1.0

CA412379192

Gene: RS1 (HGNC:6247)
Condition: X-linked retinoschisis (MONDO:0010725)
Inheritance Mode: X-linked inheritance
UUID: f425418d-db13-40f9-83e7-90d376e9256e
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.16G>T
NC_000023.11:g.18672053C>A
CM000685.2:g.18672053C>A
NC_000023.10:g.18690173C>A
CM000685.1:g.18690173C>A
NC_000023.9:g.18600094C>A
NG_008659.3:g.10396G>T
ENST00000379984.4:c.16G>T
ENST00000379984.3:c.16G>T
NM_000330.3:c.16G>T

Likely Pathogenic

Met criteria codes 2

PVS1 PM2_Supporting

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 1.0.0*
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.16G>T (p.Glu6Ter) variant is a nonsense variant that introduces a premature stop codon in exon 1 of 6 and is predicted to trigger nonsense-mediated decay (PVS1). This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting). This variant has been reported in at least 1 proband meeting the PS4 requirement of a male diagnosed with X-linked retinoschisis (PMID: 23009889), however, PS4_Supporting requires at least 2 unrelated probands, so this criterion was not met. In summary, this variant is classified as likely 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 and PVS1.

Met criteria codes

PVS1	✔	This is a nonsense variant that introduces a premature stop codon between amino acids 1 and 223 that is predicted to either trigger nonsense-mediated decay or to disrupt a critical C-terminal region required for the proper function of RS1 (PVS1, PMID: 19849666).
PM2_Supporting	✔	This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting).

Not Met criteria codes

PS4	✖	This variant has been reported in at least 1 proband meeting the PS4 requirement of a male diagnosed with XLRS (PMID: 23009889). PS4_Supporting requires at least 2 unrelated probands, so this criterion was not met.

Curation History

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