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Variant: NM_000330.4(RS1):c.103C>T (p.Gln35Ter)

Version: 1.0

CA412376480

1805017 (ClinVar)

Gene: RS1 (HGNC:6247)
Condition: X-linked retinoschisis (MONDO:0010725)
Inheritance Mode: X-linked inheritance
UUID: 982b9872-f29f-4e7c-9e7f-99db8e0fae83
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.103C>T
NM_000330.4(RS1):c.103C>T (p.Gln35Ter)
NC_000023.11:g.18656734G>A
CM000685.2:g.18656734G>A
NC_000023.10:g.18674854G>A
CM000685.1:g.18674854G>A
NC_000023.9:g.18584775G>A
NG_008659.3:g.25715C>T
ENST00000379984.4:c.103C>T
ENST00000379984.3:c.103C>T
NM_000330.3:c.103C>T

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PS4_Supporting

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

NM_000330.4(RS1):c.103C>T (p.Gln35Ter) is a nonsense variant that introduces a premature stop codon in exon 3 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 two apparently unrelated probands meeting the PS4 requirements of a male diagnosed with X-linked retinoschisis (PMIDs: 23847049, 15932525, PS4_Supporting). 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 PS4_Supporting.

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).
PS4_Supporting	✔	This variant has been reported in at least two apparently unrelated probands meeting the PS4 requirements of a male diagnosed with X-linked retinoschisis (PMIDs: 23847049, 15932525, PS4_Supporting).

Curation History

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