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Variant: NM_000330.4:c.52+2T>A

Version: 1.0

CA412378903

Gene: RS1 (HGNC:6247)
Condition: X-linked retinoschisis (MONDO:0010725)
Inheritance Mode: X-linked inheritance
UUID: d91e17a5-2824-4d5c-b3de-ac7d49bf1de3
Approved on: Sat, 26 Sep 2026 00:00:00 +0000
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HGVS expressions

NM_000330.4:c.52+2T>A
NC_000023.11:g.18672015A>T
CM000685.2:g.18672015A>T
NC_000023.10:g.18690135A>T
CM000685.1:g.18690135A>T
NC_000023.9:g.18600056A>T
NG_008659.3:g.10434T>A
ENST00000379984.4:c.52+2T>A
ENST00000379984.3:c.52+2T>A
NM_000330.3:c.52+2T>A

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PP4

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.52+2T>A variant is a canonical splice site variant in intron 1, located 2 nucleotides after exon 1. This variant occurs at a canonical splice site in intron 1 and is predicted to disrupt splicing and induce skipping of exon 2, which is expected to introduce a premature stop codon and disrupt a critical functional domain in RS1 (PVS1). This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting). At least one proband harboring this variant exhibits the following phenotypes, including the appearance of schisis and reduced visual acuity before age 13 years, which together are specific for X-linked retinoschisis (PMID: 33124204, PP4). 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: PVS1, PM2_Supporting, and PP4.

Met criteria codes

PVS1	✔	This variant occurs at a canonical splice site in intron 1 and is predicted to disrupt splicing and induce skipping of exon 2, which is expected to introduce a premature stop codon and disrupt a critical functional domain in RS1 (PVS1).
PM2_Supporting	✔	This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting).
PP4	✔	At least one proband harboring this variant exhibits the following phenotypes, including the appearance of schisis and reduced visual acuity before age 13 years, which together are specific for X-linked retinoschisis (PMID: 33124204, PP4).

Curation History

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