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Variant: NM_001034853.2:c.644G>A

Version: 1.0

CA412743026

4692558 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 513f58f9-3ad0-4fcd-97f7-451fc9021e76
Approved on: Sat, 26 Sep 2026 00:00:00 +0000
Published on: Sat, 26 Sep 2026 00:00:00 +0000

HGVS expressions

NM_001034853.2:c.644G>A
NC_000023.11:g.38310749C>T
CM000685.2:g.38310749C>T
NC_000023.10:g.38170002C>T
CM000685.1:g.38170002C>T
NC_000023.9:g.38054946C>T
NG_009553.1:g.21787G>A
ENST00000642170.1:n.1054G>A
ENST00000642373.1:c.*223G>A
ENST00000642395.2:c.644G>A
ENST00000642558.1:c.551G>A
ENST00000642739.1:c.644G>A
ENST00000644238.1:c.644G>A
ENST00000644337.1:c.644G>A
ENST00000645032.1:c.644G>A
ENST00000645124.1:c.644G>A
ENST00000646020.1:c.644G>A
ENST00000647261.1:c.644G>A
ENST00000318842.11:c.644G>A
ENST00000339363.7:c.644G>A
ENST00000378505.6:c.644G>A
ENST00000465127.1:c.172-355372C>T
ENST00000474584.5:c.644G>A
ENST00000482855.5:c.644G>A
NM_000328.2:c.644G>A
NM_001034853.1:c.644G>A
NM_001367245.1:c.641G>A
NM_001367246.1:c.644G>A
NM_001367247.1:c.644G>A
NM_001367248.1:c.674G>A
NM_001367249.1:c.641G>A
NM_001367250.1:c.641G>A
NM_001367251.1:c.644G>A
NR_159803.1:n.786G>A
NR_159804.1:n.695G>A
NR_159805.1:n.786G>A
NR_159806.1:n.786G>A

NR_159807.1:n.786G>A
NR_159808.1:n.1054G>A
NM_000328.3:c.644G>A

Likely Pathogenic

Met criteria codes 4

PM2_Supporting PP3_Strong PP1 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 RPGR 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_001034853.2(RPGR):c.644G>A variant is a missense variant encoding the substitution of glycine with glutamate at amino acid 215. This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting). The variant has been reported to segregate with retinal dystrophy through an affected mother and son. (PP1; PMID: 31652454). The computational predictor REVEL gives a score of 0.958, which is above the ClinGen X-linked IRD VCEP threshold of >0.932 and predicts a damaging effect on RPGR function (PP3_strong). The computational splicing predictor SpliceAI gives a delta score of 0.02 for acceptor gain, which is below the ClinGen X-linked IRD VCEP threshold of >0.2 and does not predict that the variant disrupts RPGR splicing. In summary, this variant is classified as likely pathogenic for RPGR-related retinopathy based on the ClinGen X-linked Inherited Retinal Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RPGR Version 1.0.0: PP3_strong, PP1, and PM2_supporting.

Met criteria codes

PM2_Supporting	✔	This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting)
PP3_Strong	✔	The computational predictor REVEL gives a score of 0.958, which is above the ClinGen X-linked IRD VCEP threshold of >0.932 and predicts a damaging effect on RPGR function (PP3_strong). The computational splicing predictor SpliceAI gives a delta score of 0.02 for acceptor gain, which is below the ClinGen X-linked IRD VCEP threshold of >0.2 and does not predict that the variant disrupts RPGR splicing.
PP1	✔	The variant has been reported to segregate with retinal dystrophy through an affected mother and son. (PP1; PMID: 31652454).
PP4	✔	At least one proband harboring this variant exhibits a phenotype including X-linked inheritance (2) NGS sequencing (2), and pigmentary retinopathy (0.5), which together are specific for RPGR-related recessive retinopathy (4.5 points, PMID: 31652454).

Curation History

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