The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001034853.2:c.934G>C

Version: 1.0

CA412741115

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 5658615a-e1f2-472a-9487-026ccd987913
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.934G>C
NC_000023.11:g.38304635C>G
CM000685.2:g.38304635C>G
NC_000023.10:g.38163888C>G
CM000685.1:g.38163888C>G
NC_000023.9:g.38048832C>G
NG_009553.1:g.27901G>C
ENST00000494707.6:c.138G>C
ENST00000642170.1:n.1189-3264G>C
ENST00000642395.2:c.934G>C
ENST00000642558.1:c.841G>C
ENST00000642739.1:c.934G>C
ENST00000644238.1:c.934G>C
ENST00000644337.1:c.934G>C
ENST00000645032.1:c.934G>C
ENST00000645124.1:c.934G>C
ENST00000646020.1:c.934G>C
ENST00000647261.1:c.934G>C
ENST00000318842.11:c.934G>C
ENST00000339363.7:c.934G>C
ENST00000378505.6:c.934G>C
ENST00000465127.1:c.172-361486C>G
ENST00000474584.5:c.934G>C
ENST00000482855.5:c.934G>C
ENST00000494841.1:n.197G>C
NM_000328.2:c.934G>C
NM_001034853.1:c.934G>C
NM_001367245.1:c.931G>C
NM_001367246.1:c.934G>C
NM_001367247.1:c.934G>C
NM_001367248.1:c.964G>C
NM_001367249.1:c.931G>C
NM_001367250.1:c.931G>C
NM_001367251.1:c.934G>C
NR_159803.1:n.1076G>C
NR_159804.1:n.985G>C
NR_159805.1:n.1076G>C

NR_159806.1:n.1076G>C
NR_159807.1:n.1076G>C
NR_159808.1:n.1189-3264G>C
NM_000328.3:c.934G>C

Likely Pathogenic

Met criteria codes 5

PS1_Moderate PP1 PP3 PP4 PM2_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 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.934G>C variant is a predicted missense variant encoding the substitution of aspartic acid with histidine at amino acid 312. However, the splicing impact predictor SpliceAI gives a score of 0.97 (donor loss), which is above the ClinGen X-linked IRD VCEP recommended threshold of ≥0.2 and predicts a damaging impact on splicing (PP3). The variant is located outside the +/- 1,2 dinucleotide and another variant with the same predicted splicing impact, c.934+1G>T, was previously classified as a pathogenic variant in the same donor/acceptor motif for the RPGR-related X-linked IRD VCEP (PS1_moderate). This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting). At least one proband harboring this variant exhibits a phenotype including childhood-onset (1), decreased central vision acuity (0.5), night blindness (0.5), optic nerve pallor (0.5), pigmentary retinopathy (0.5), rod cone dystrophy (1), X-linked inheritance (2), which together are specific for RPGR-related recessive retinopathy (6 points, PMID: 36882936, PP4). The variant has been reported to segregate with retinal dystrophy through at least 2 affected meiosis from 1 family. (PP1; PMID: 36882936).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: PS1_moderate, PP3, PP4, PP1, and PM2_supporting.

Met criteria codes

PS1_Moderate	✔	This variant is outside the +/- 1,2 dinucleotide and meets the PP3 code due to predicted splicing disruption and another predicted splicing variant, c.934+1G>T, was previously classified as a pathogenic variant in the same donor/acceptor motif for the RPGR-related X-linked IRD VCEP (PS1_moderate).
PP1	✔	The variant has been reported to segregate with retinal dystrophy through at least 2 affected meiosis from 1 family. (PP1; PMID: 36882936).
PP3	✔	The splicing impact predictor SpliceAI gives a score of 0.97 (donor loss), which is above the ClinGen X-linked IRD VCEP recommended threshold of ≥0.2 and predicts a damaging impact on splicing (PP3).
PP4	✔	At least one proband harboring this variant exhibits a phenotype including childhood-onset (1), decreased central vision acuity (0.5), night blindness (0.5), optic nerve pallor (0.5), pigmentary retinopathy (0.5), rod cone dystrophy

(1), X-linked inheritance (2), which together are specific for RPGR-related recessive retinopathy (6 points, PMID: 36882936, PP4).

PM2_Supporting ✔ This variant is absent from hemizygous individuals in gnomAD v4.1.0 (PM2_Supporting).

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001034853.2:c.934G>C	Likely Pathogenic	RPGR-Related Retinopathy	2026-09-26	1.0	ClinGen X-linked Inherited Retinal Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RPGR Version 1.0	RPGR

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.