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Variant: NM_001034853.2:c.851C>G

Version: 1.0

CA412741475

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 9528d860-e0cf-47ad-9ed9-405d03ad84b5
Approved on: 2025/9/7
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HGVS expressions

NM_001034853.2:c.851C>G
NC_000023.11:g.38304718G>C
CM000685.2:g.38304718G>C
NC_000023.10:g.38163971G>C
CM000685.1:g.38163971G>C
NC_000023.9:g.38048915G>C
NG_009553.1:g.27818C>G
ENST00000494707.6:c.55C>G
ENST00000642170.1:n.1189-3347C>G
ENST00000642373.1:c.*430C>G
ENST00000642395.2:c.851C>G
ENST00000642558.1:c.758C>G
ENST00000642739.1:c.851C>G
ENST00000644238.1:c.851C>G
ENST00000644337.1:c.851C>G
ENST00000645032.1:c.851C>G
ENST00000645124.1:c.851C>G
ENST00000646020.1:c.851C>G
ENST00000647261.1:c.851C>G
ENST00000318842.11:c.851C>G
ENST00000339363.7:c.851C>G
ENST00000378505.6:c.851C>G
ENST00000465127.1:c.172-361403G>C
ENST00000474584.5:c.851C>G
ENST00000482855.5:c.851C>G
ENST00000494841.1:n.114C>G
NM_000328.2:c.851C>G
NM_001034853.1:c.851C>G
NM_001367245.1:c.848C>G
NM_001367246.1:c.851C>G
NM_001367247.1:c.851C>G
NM_001367248.1:c.881C>G
NM_001367249.1:c.848C>G
NM_001367250.1:c.848C>G
NM_001367251.1:c.851C>G
NR_159803.1:n.993C>G
NR_159804.1:n.902C>G

NR_159805.1:n.993C>G
NR_159806.1:n.993C>G
NR_159807.1:n.993C>G
NR_159808.1:n.1189-3347C>G
NM_000328.3:c.851C>G

Pathogenic

Met criteria codes 3

PP4 PM2_Supporting PVS1

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 1.0.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

X-linked Inherited Retinal Disease VCEP

NM_001034853.2(RPGR):c.851C>G (p.Ser284Ter) is a nonsense variant that introduces a premature stop in codon 284 within exon 8 of 15 and is predicted to trigger nonsense-mediated decay (PVS1). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). At least one proband harboring this variant exhibits a phenotype including early onset (1pts), night blindness (0.5 pts), reduced visual acuity (0.5 pts), pigment deposits (0.5 pts), optic disc pallor (0.5 pts), and genotyping by next-generation sequencing with a panel of 483 genes that did not identify an alternative basis for retinal disease (2 pts), which together are specific for RPGR-related retinopathy (5 points, PMID: 36276946, PP4). In summary, this variant is classified as 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; PVS1, PM2_supporting, and PP4.

Met criteria codes

PP4	✔	At least one proband harboring this variant exhibits a phenotype including early onset (1pts), night blindness (0.5 pts), reduced visual acuity (0.5 pts), pigment deposits (0.5 pts), optic disc pallor (0.5 pts), and genotyping by next-generation sequencing with a panel of 483 genes that did not identify an alternative basis for retinal disease (2 pts), which together are specific for RPGR-related retinopathy (5 points, PMID: 36276946, PP4).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PVS1	✔	This is a nonsense variant that introduces a premature stop codon between amino acids 1-1132 that is predicted to either trigger nonsense-mediated decay or to disrupt a critical C-terminal region required for proper glutamylation of RPGR (PVS1, PMID: 36445968).

Curation History

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