

Variant: NM_001034853.2(RPGR):c.2491G>T (p.Glu831Ter)

Version: 1.0

CA412730514 [↗](#)

1297121 (ClinVar) [↗](#)

Gene: RPGR ([HGNC:6103](#))

Condition: RPGR-related retinopathy ([MONDO:0100437](#))

Inheritance Mode: X-linked inheritance

UUID: 423a2e20-8f42-4e6a-88fa-5dcaa75eb3c0

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HGVS expressions

NM_001034853.2:c.2491G>T

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NC_000023.11:g.38286508C>A

CM000685.2:g.38286508C>A

NC_000023.10:g.38145761C>A

CM000685.1:g.38145761C>A

NC_000023.9:g.38030705C>A

NG_009553.1:g.46028G>T

ENST00000494707.6:c.953+1357G>T

ENST00000642170.1:n.1826+4451G>T

ENST00000642395.2:c.1905+586G>T

ENST00000642739.1:c.1572+4451G>T

ENST00000644238.1:c.1386+4451G>T

ENST00000644337.1:c.1719+586G>T

ENST00000645032.1:c.2491G>T

ENST00000645124.1:c.*101+586G>T

ENST00000646020.1:c.*594+586G>T

ENST00000318842.11:c.1905+586G>T

ENST00000339363.7:c.2520+586G>T

ENST00000378505.6:c.2491G>T

ENST00000465127.1:c.172-379613C>A

ENST00000474584.5:c.*37+4451G>T

ENST00000482855.5:c.1905+586G>T

ENST00000494707.5:c.139+4451G>T

NM_000328.2:c.1905+586G>T

NM_001034853.1:c.2491G>T

NM_001367245.1:c.1902+586G>T

NM_001367246.1:c.1719+586G>T

NM_001367247.1:c.1572+4451G>T

NM_001367248.1:c.1602+4451G>T

NM_001367249.1:c.1569+4451G>T

NM_001367250.1:c.1569+4451G>T

NM_001367251.1:c.1386+4451G>T

NR_159803.1:n.2263+586G>T

NR_159804.1:n.1648+4451G>T

NR_159805.1:n.1714+4451G>T

NR_159806.1:n.1866+586G>T

NR_159807.1:n.1622+4451G>T
NR_159808.1:n.1826+4451G>T
NM_000328.3:c.1905+586G>T

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 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.2491G>T (p.Glu831Ter) is a nonsense variant introducing a premature stop in codon 831 within exon 15 of 15 that is predicted to disrupt a critical C-terminal region required for proper glutamylation of RPGR (PVS1, PMID: 36445968). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). At least one proband harboring this variant exhibits a phenotype including presentation with night blindness (0.5 pts) with onset at age 5 years (1 pt), reduced visual acuity (0.5 pts), diminished electroretinogram responses from rods, fundus appearance showing widespread retinal pigment epithelium degeneration with pigment deposits (0.5 pts), optic disc pallor (0.5 pts), severe macular degeneration, artery attenuation, and genotyping by next-generation sequencing with a 483-gene panel showing no alternative basis for inherited 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

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).
PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP4			At least one proband harboring this variant exhibits a phenotype including presentation with night blindness (0.5 pts) with onset at age 5 years (1 pt), reduced visual acuity (0.5 pts), diminished electroretinogram responses from rods, fundus appearance showing widespread retinal pigment epithelium degeneration with pigment deposits (0.5 pts), optic disc pallor (0.5 pts), severe macular degeneration, artery attenuation, and genotyping by next-generation sequencing with a 483-gene panel showing no alternative basis for inherited retinal disease (2 pts), which together are specific for RPGR-related retinopathy (5 points, PMID: 36276946, PP4).



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