

Variant: NM_001034853.2(RPGR):c.2695G>T (p.Glu899Ter)

Version: 1.0

CA412730005 [↗](#)

866316 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 34d28fdd-694f-4c62-9cbd-9ad808ec2be4

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HGVS expressions

NM_001034853.2:c.2695G>T

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NC_000023.11:g.38286304C>A

CM000685.2:g.38286304C>A

NC_000023.10:g.38145557C>A

CM000685.1:g.38145557C>A

NC_000023.9:g.38030501C>A

NG_009553.1:g.46232G>T

ENST00000494707.6:c.953+1561G>T

ENST00000642170.1:n.1826+4655G>T

ENST00000642395.2:c.1905+790G>T

ENST00000642739.1:c.1572+4655G>T

ENST00000644238.1:c.1386+4655G>T

ENST00000644337.1:c.1719+790G>T

ENST00000645032.1:c.2695G>T

ENST00000645124.1:c.*101+790G>T

ENST00000646020.1:c.*594+790G>T

ENST00000318842.11:c.1905+790G>T

ENST00000339363.7:c.2520+790G>T

ENST00000378505.6:c.2695G>T

ENST00000465127.1:c.172-379817C>A

ENST00000474584.5:c.*37+4655G>T

ENST00000482855.5:c.1905+790G>T

ENST00000494707.5:c.139+4655G>T

NM_000328.2:c.1905+790G>T

NM_001034853.1:c.2695G>T

NM_001367245.1:c.1902+790G>T

NM_001367246.1:c.1719+790G>T

NM_001367247.1:c.1572+4655G>T

NM_001367248.1:c.1602+4655G>T

NM_001367249.1:c.1569+4655G>T

NM_001367250.1:c.1569+4655G>T

NM_001367251.1:c.1386+4655G>T

NR_159803.1:n.2263+790G>T

NR_159804.1:n.1648+4655G>T

NR_159805.1:n.1714+4655G>T

NR_159806.1:n.1866+790G>T

NR_159807.1:n.1622+4655G>T
NR_159808.1:n.1826+4655G>T
NM_000328.3:c.1905+790G>T

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.2695G>T (p.Glu899Ter) is a nonsense variant that introduces a premature stop in codon 899 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 4 years (1 pt), reduced visual acuity (0.5 pts), decreased electroretinogram responses from rods, mild macular degeneration, pigment deposits (0.5 pts), 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 (4.5 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 presentation with night blindness (0.5 pts) with onset at age 4 years (1 pt), reduced visual acuity (0.5 pts), decreased electroretinogram responses from rods, mild macular degeneration, pigment deposits (0.5 pts), 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 (4.5 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).

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