

Variant: *NM_001034853.2(RPGR):c.2719G>T (p.Glu907Ter)*

Version: 1.0

CA412729944 [↗](#)

3702951 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

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HGVS expressions

NM_001034853.2:c.2719G>T

NM_001034853.2(RPGR):c.2719G>T (p.Glu907Ter)

NC_000023.11:g.38286280C>A

CM000685.2:g.38286280C>A

NC_000023.10:g.38145533C>A

CM000685.1:g.38145533C>A

NC_000023.9:g.38030477C>A

NG_009553.1:g.46256G>T

ENST00000494707.6:c.953+1585G>T

ENST00000642170.1:n.1826+4679G>T

ENST00000642395.2:c.1905+814G>T

ENST00000642739.1:c.1572+4679G>T

ENST00000644238.1:c.1386+4679G>T

ENST00000644337.1:c.1719+814G>T

ENST00000645032.1:c.2719G>T

ENST00000645124.1:c.*101+814G>T

ENST00000646020.1:c.*594+814G>T

ENST00000318842.11:c.1905+814G>T

ENST00000339363.7:c.2520+814G>T

ENST00000378505.6:c.2719G>T

ENST00000465127.1:c.172-379841C>A

ENST00000474584.5:c.*37+4679G>T

ENST00000482855.5:c.1905+814G>T

ENST00000494707.5:c.139+4679G>T

NM_000328.2:c.1905+814G>T

NM_001034853.1:c.2719G>T

NM_001367245.1:c.1902+814G>T

NM_001367246.1:c.1719+814G>T

NM_001367247.1:c.1572+4679G>T

NM_001367248.1:c.1602+4679G>T

NM_001367249.1:c.1569+4679G>T

NM_001367250.1:c.1569+4679G>T

NM_001367251.1:c.1386+4679G>T

NR_159803.1:n.2263+814G>T

NR_159804.1:n.1648+4679G>T

NR_159805.1:n.1714+4679G>T

NR_159806.1:n.1866+814G>T

NR_159807.1:n.1622+4679G>T
NR_159808.1:n.1826+4679G>T
NM_000328.3:c.1905+814G>T

Pathogenic

Met criteria codes 4

PP1_Moderate

PM2_Supporting

PVS1

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

NM_001034853.2(RPGR):c.2719G>T (p.Glu907Ter) is a nonsense variant that introduces a premature stop codon between amino acids 1-1132 that is predicted to disrupt a critical C-terminal region required for proper glutamylation of RPGR (PVS1, PMID: 36445968). This variant is absent from hemizygous individuals in gnomAD v4.1.1 (PM2_Supporting). At least one proband harboring this variant exhibits a phenotype including Cone dystrophy (1), 2nd decade onset (1), abnormal color vision (0.5), decreased central vision acuity (0.5), ERG absent (0), pigmentary retinopathy (0.5), and macular atrophy (1), which together are specific for RPGR-related retinopathy (4.5 points, PMID: 35806195, PP4). The variant has been reported to segregate with retinal dystrophy through at least 3 affected meioses from 1 family. (PP1_moderate; PMID: 41686256). 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, PP1_strong, PP4, and PM2_supporting.

Met criteria codes

PP1_Moderate



The variant has been reported to segregate with retinal dystrophy through at least 3 affected meioses from 1 family. (PP1_moderate; PMID: 41686256).

PM2_Supporting



This variant is absent from hemizygous individuals in gnomAD v4.1.1 (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)

PP4



At least one proband harboring this variant exhibits a phenotype including Cone dystrophy (1), 2nd decade onset (1), abnormal color vision (0.5), decreased central vision acuity (0.5), ERG absent (0), pigmentary retinopathy (0.5), and macular atrophy (1), which together are specific for RPGR-related retinopathy (4.5 points, PMID: 35806195, PP4).

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