

Variant: *NM_001034853.2:c.2101G>T*

Version: 1.0

CA412731451 [↗](#)

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

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

Inheritance Mode: X-linked inheritance (primarily recessive with milder female expression)

UUID: 56f6daef-8cbf-497b-bded-49d18392a82b

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HGVS expressions

NM_001034853.2:c.2101G>T

NC_000023.11:g.38286898C>A
CM000685.2:g.38286898C>A
NC_000023.10:g.38146151C>A
CM000685.1:g.38146151C>A
NC_000023.9:g.38031095C>A
NG_009553.1:g.45638G>T
ENST00000494707.6:c.953+967G>T
ENST00000642170.1:n.1826+4061G>T
ENST00000642395.2:c.1905+196G>T
ENST00000642739.1:c.1572+4061G>T
ENST00000644238.1:c.1386+4061G>T
ENST00000644337.1:c.1719+196G>T
ENST00000645032.1:c.2101G>T
ENST00000645124.1:c.*101+196G>T
ENST00000646020.1:c.*594+196G>T
ENST00000318842.11:c.1905+196G>T
ENST00000339363.7:c.2520+196G>T
ENST00000378505.6:c.2101G>T
ENST00000465127.1:c.172-379223C>A
ENST00000474584.5:c.*37+4061G>T
ENST00000482855.5:c.1905+196G>T
ENST00000494707.5:c.139+4061G>T
NM_000328.2:c.1905+196G>T
NM_001034853.1:c.2101G>T
NM_001367245.1:c.1902+196G>T
NM_001367246.1:c.1719+196G>T
NM_001367247.1:c.1572+4061G>T
NM_001367248.1:c.1602+4061G>T
NM_001367249.1:c.1569+4061G>T
NM_001367250.1:c.1569+4061G>T
NM_001367251.1:c.1386+4061G>T
NR_159803.1:n.2263+196G>T
NR_159804.1:n.1648+4061G>T
NR_159805.1:n.1714+4061G>T
NR_159806.1:n.1866+196G>T
NR_159807.1:n.1622+4061G>T

NR_159808.1:n.1826+4061G>T
NM_000328.3:c.1905+196G>T

Likely Pathogenic

Met criteria codes 2

PVS1

PM2_Supporting

Not Met criteria codes 1

PS4

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.2101G>T (p.Glu701Ter) is a nonsense variant introducing a premature stop in codon 701 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). This variant has been reported in a submission to the Leiden Open Variation Database (LOVD 3.0) as a disease-causing variant (DB-ID RPGR_000065), however, no affected individual was described meeting one of the PS4 requirements of some functional vision impairment in an affected male by age 30 years, or decreased or absent electroretinogram responses, so PS4_Supporting was not met. 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; PVS1 and PM2_Supporting.

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 v4.0.0 (PM2_Supporting).

Not Met criteria codes

PS4



This variant has been reported in a submission to the Leiden Open Variation Database (LOVD 3.0) as a disease-causing variant (DB-ID RPGR_000065), however, no affected individual was described meeting one of the PS4 requirements of some functional vision impairment in an affected male by age 30 years, or decreased or absent cone and/or rod electroretinogram responses, so PS4_Supporting was not met.

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