The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001034853.2(RPGR):c.980T>G (p.Leu327Ter)

Version: 1.0

CA226468

98816 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 9ffae707-f809-420d-9bf1-4297cd9f8028
Approved on: 2025/9/7
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HGVS expressions

NM_001034853.2(RPGR):c.980T>G (p.Leu327Ter)
NM_001034853.2:c.980T>G
NC_000023.11:g.38301326A>C
CM000685.2:g.38301326A>C
NC_000023.10:g.38160579A>C
CM000685.1:g.38160579A>C
NC_000023.9:g.38045523A>C
NG_009553.1:g.31210T>G
ENST00000494707.6:c.184T>G
ENST00000642170.1:n.1234T>G
ENST00000642395.2:c.980T>G
ENST00000642558.1:c.887T>G
ENST00000642739.1:c.980T>G
ENST00000644238.1:c.980T>G
ENST00000644337.1:c.980T>G
ENST00000645032.1:c.980T>G
ENST00000645124.1:c.980T>G
ENST00000646020.1:c.1040T>G
ENST00000318842.11:c.980T>G
ENST00000339363.7:c.980T>G
ENST00000378505.6:c.980T>G
ENST00000464437.1:c.46T>G
ENST00000465127.1:c.172-364795A>C
ENST00000474584.5:c.980T>G
ENST00000482855.5:c.980T>G
ENST00000494841.1:n.243T>G
NM_000328.2:c.980T>G
NM_001034853.1:c.980T>G
NM_001367245.1:c.977T>G
NM_001367246.1:c.980T>G
NM_001367247.1:c.980T>G
NM_001367248.1:c.1010T>G
NM_001367249.1:c.977T>G
NM_001367250.1:c.977T>G
NM_001367251.1:c.980T>G
NR_159803.1:n.1182T>G
NR_159804.1:n.1031T>G

NR_159805.1:n.1122T>G
NR_159806.1:n.1122T>G
NR_159807.1:n.1122T>G
NR_159808.1:n.1234T>G
NM_000328.3:c.980T>G

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PS4_Supporting

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.980T>G (p.Leu327Ter) is a nonsense variant introducing a premature stop in codon 327 within exon 9 of 15, which is predicted to trigger nonsense-mediated decay (PVS1). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). This variant has been reported in at least 2 apparently unrelated probands meeting one of the PS4 requirements of a male with some functional vision impairment by age 30 years and/or decreased or absent electroretinogram responses, or a female with functional visual abnormality and documentation of a male relative affected with retinitis pigmentosa (PMIDs: 17325176, 32531858, PS4_Supporting). 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 PS4_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 v4.1.0 (PM2_Supporting).
PS4_Supporting	✔	This variant has been reported in at least 2 apparently unrelated probands meeting one of the PS4 requirements of a male with some functional vision impairment by age 30 and/or decreased or absent ERG responses, or a female with functional visual abnormality and documentation of a male relative affected with retinitis pigmentosa (PMIDs: 17325176, 32531858, PS4_Supporting).

Curation History

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