

Variant: NM_001034853.2(RPGR):c.1100del (p.Pro367fs)

Version: 1.0

CA226343 [↗](#)

98728 (ClinVar) [↗](#)

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

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

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

UUID: ae32c582-19d1-42ce-958d-43011fbc888e

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HGVS expressions

- NM_001034853.2:c.1100del
- NM_001034853.2(RPGR):c.1100del (p.Pro367fs)
- NC_000023.11:g.38299102del
- CM000685.2:g.38299102del
- NC_000023.10:g.38158355del
- CM000685.1:g.38158355del
- NC_000023.9:g.38043299del
- NG_009553.1:g.33435del
- ENST00000494707.6:c.304del
- ENST00000642170.1:n.1354del
- ENST00000642395.2:c.1100del
- ENST00000642558.1:c.1007del
- ENST00000642739.1:c.1100del
- ENST00000644238.1:c.1060-1649del
- ENST00000644337.1:c.1060-1649del
- ENST00000645032.1:c.1100del
- ENST00000645124.1:c.1100del
- ENST00000646020.1:c.1160del
- ENST00000318842.11:c.1100del
- ENST00000339363.7:c.1100del
- ENST00000378505.6:c.1100del
- ENST00000464437.1:c.166del
- ENST00000465127.1:c.172-367019del
- ENST00000474584.5:c.1100del
- ENST00000482855.5:c.1100del
- ENST00000494841.1:n.363del
- NM_000328.2:c.1100del
- NM_001034853.1:c.1100del
- NM_001367245.1:c.1097del
- NM_001367246.1:c.1060-1649del
- NM_001367247.1:c.1100del
- NM_001367248.1:c.1130del
- NM_001367249.1:c.1097del
- NM_001367250.1:c.1097del
- NM_001367251.1:c.1060-1649del
- NR_159803.1:n.1302del
- NR_159804.1:n.1151del

NR_159805.1:n.1242del
NR_159806.1:n.1242del
NR_159807.1:n.1242del
NR_159808.1:n.1354del
NM_000328.3:c.1100del

Pathogenic

Met criteria codes 3

PS4_Supporting

PVS1

PM2_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.1100del (p.Pro367LeufsTer14) is a frameshift variant due to deletion of 1 nucleotide and introduces a premature stop codon after 14 amino acids in exon 10 of 15 that is predicted to result in 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 (PMID: 36017691, PMID: 10937588, 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, PS4_Supporting and PM2_Supporting. (date of approval 05/16/2025).

Met criteria codes

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: 36017691 and 10937588, PS4_Supporting).

PVS1



This is a frameshift 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).



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