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.838_842del (p.Phe279_Leu280insTer)

Version: 1.0

CA2580616965

2138542 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 5a3264a5-fa8e-4cdf-a7a3-9b61469a95ff
Approved on: 2025/9/7
Published on: 2025/9/7

HGVS expressions

NM_001034853.2:c.838_842del
NM_001034853.2(RPGR):c.838_842del (p.Phe279_Leu280insTer)
NC_000023.11:g.38304732_38304736del
CM000685.2:g.38304732_38304736del
NC_000023.10:g.38163985_38163989del
CM000685.1:g.38163985_38163989del
NC_000023.9:g.38048929_38048933del
NG_009553.1:g.27805_27809del
ENST00000494707.6:c.42_46del
ENST00000642170.1:n.1189-3360_1189-3356del
ENST00000642373.1:c.*417_*421del
ENST00000642395.2:c.838_842del
ENST00000642558.1:c.745_749del
ENST00000642739.1:c.838_842del
ENST00000644238.1:c.838_842del
ENST00000644337.1:c.838_842del
ENST00000645032.1:c.838_842del
ENST00000645124.1:c.838_842del
ENST00000646020.1:c.838_842del
ENST00000647261.1:c.838_842del
ENST00000318842.11:c.838_842del
ENST00000339363.7:c.838_842del
ENST00000378505.6:c.838_842del
ENST00000465127.1:c.172-361389_172-361385del
ENST00000474584.5:c.838_842del
ENST00000482855.5:c.838_842del
ENST00000494841.1:n.101_105del
NM_000328.2:c.838_842del
NM_001034853.1:c.838_842del
NM_001367245.1:c.835_839del
NM_001367246.1:c.838_842del
NM_001367247.1:c.838_842del
NM_001367248.1:c.868_872del
NM_001367249.1:c.835_839del
NM_001367250.1:c.835_839del

NM_001367251.1:c.838_842del
NR_159803.1:n.980_984del
NR_159804.1:n.889_893del
NR_159805.1:n.980_984del
NR_159806.1:n.980_984del
NR_159807.1:n.980_984del
NR_159808.1:n.1189-3360_1189-3356del
NM_000328.3:c.838_842del

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.838_842del (p.Phe279_Leu280insTer) is a frameshift variant due to 7-nucleotide deletion which results in a premature stop codon within exon 8 of 15 and 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 (PMID: 17325176, PMID: 21857984; 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

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, 21857984; 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 v4.1.0 (PM2_Supporting).

Curation History

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