Variant: NM_001034853.2(RPGR):c.2901_2903del (p.Glu969del)

Version: 1.0

CA10385235

1199928 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 213abb16-7e96-432f-a07c-30fc63455df7
Approved on: 2025-05-20
Published on: 2025-05-21

HGVS expressions

NM_001034853.2:c.2901_2903del
NM_001034853.2(RPGR):c.2901_2903del (p.Glu969del)
NC_000023.11:g.38286098_38286100del
CM000685.2:g.38286098_38286100del
NC_000023.10:g.38145351_38145353del
CM000685.1:g.38145351_38145353del
NC_000023.9:g.38030295_38030297del
NG_009553.1:g.46438_46440del
ENST00000494707.6:c.953+1767_953+1769del
ENST00000642170.1:n.1826+4861_1826+4863del
ENST00000642395.2:c.1905+996_1905+998del
ENST00000642739.1:c.1572+4861_1572+4863del
ENST00000644238.1:c.1386+4861_1386+4863del
ENST00000644337.1:c.1719+996_1719+998del
ENST00000645032.1:c.2901_2903del
ENST00000645124.1:c.*101+996_*101+998del
ENST00000646020.1:c.*594+996_*594+998del
ENST00000318842.11:c.1905+996_1905+998del
ENST00000339363.7:c.2520+996_2520+998del
ENST00000378505.6:c.2901_2903del
ENST00000465127.1:c.172-380023_172-380021del
ENST00000474584.5:c.*37+4861_*37+4863del
ENST00000482855.5:c.1905+996_1905+998del
ENST00000494707.5:c.139+4861_139+4863del
NM_000328.2:c.1905+996_1905+998del
NM_001034853.1:c.2901_2903del
NM_001367245.1:c.1902+996_1902+998del
NM_001367246.1:c.1719+996_1719+998del
NM_001367247.1:c.1572+4861_1572+4863del
NM_001367248.1:c.1602+4861_1602+4863del
NM_001367249.1:c.1569+4861_1569+4863del
NM_001367250.1:c.1569+4861_1569+4863del
NM_001367251.1:c.1386+4861_1386+4863del
NR_159803.1:n.2263+996_2263+998del
NR_159804.1:n.1648+4861_1648+4863del

NR_159805.1:n.1714+4861_1714+4863del
NR_159806.1:n.1866+996_1866+998del
NR_159807.1:n.1622+4861_1622+4863del
NR_159808.1:n.1826+4861_1826+4863del
NM_000328.3:c.1905+996_1905+998del

Likely Benign

Met criteria codes 1

BP3

Not Met criteria codes 2

PM4 BS1

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.2901_2903del (p.Glu968del) is an in-frame deletion variant. This variant is a short in-frame deletion of 3 base pairs that encodes amino acid 968, and is located within a low-complexity region (PMID: 27162334) that extends approximately from amino acids 787–1043 in RPGR (BP3). This variant is present in gnomAD v4.1.0 at a frequency of 0.000009574 among hemizygous individuals, with 1 variant allele / 104,450 total alleles, which is higher than the ClinGen X-linked IRD VCEP BS1 threshold of >0.000005. However, the VCEP recommended not to apply the BS1 code based on the gnomAD quality control data indicating low coverage and sequencing quality at this site. In summary, this variant is classified as likely benign 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; BP3. (date of approval 05/16/2025).

Met criteria codes

BP3	✔	This variant is a short in-frame deletion of 3 base pairs that encodes amino acid 968 within a low-complexity region (PMID: 27162334) that extends approximately from amino acids 787–1043 in RPGR (BP3).

Not Met criteria codes

PM4	✖	N/A
BS1	✖	This variant is present in gnomAD v.4.1.0 at a frequency of 0.000009574 among hemizygous individuals, with 1 variant alleles / 104450 total alleles, which is higher than the ClinGen X-linked IRD VCEP BS1 threshold of >0.000005 (BS1). Only One individual was found to carry this variant and be healthy. The specific age (>30) and clinical evaluation are unknown. Therefore, the panel decided not to use this code.

Curation History

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