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Variant: NM_001034853.2(RPGR):c.2856A>T (p.Glu952Asp)

Version: 1.0

CA327915374

2660291 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 9998ade5-386d-45a7-b0d2-58be5d5e7cb0
Approved on: 2025/9/5
Published on: 2025/9/5

HGVS expressions

NM_001034853.2:c.2856A>T
NM_001034853.2(RPGR):c.2856A>T (p.Glu952Asp)
NC_000023.11:g.38286143T>A
CM000685.2:g.38286143T>A
NC_000023.10:g.38145396T>A
CM000685.1:g.38145396T>A
NC_000023.9:g.38030340T>A
NG_009553.1:g.46393A>T
ENST00000494707.6:c.953+1722A>T
ENST00000642170.1:n.1826+4816A>T
ENST00000642395.2:c.1905+951A>T
ENST00000642739.1:c.1572+4816A>T
ENST00000644238.1:c.1386+4816A>T
ENST00000644337.1:c.1719+951A>T
ENST00000645032.1:c.2856A>T
ENST00000645124.1:c.*101+951A>T
ENST00000646020.1:c.*594+951A>T
ENST00000318842.11:c.1905+951A>T
ENST00000339363.7:c.2520+951A>T
ENST00000378505.6:c.2856A>T
ENST00000465127.1:c.172-379978T>A
ENST00000474584.5:c.*37+4816A>T
ENST00000482855.5:c.1905+951A>T
ENST00000494707.5:c.139+4816A>T
NM_000328.2:c.1905+951A>T
NM_001034853.1:c.2856A>T
NM_001367245.1:c.1902+951A>T
NM_001367246.1:c.1719+951A>T
NM_001367247.1:c.1572+4816A>T
NM_001367248.1:c.1602+4816A>T
NM_001367249.1:c.1569+4816A>T
NM_001367250.1:c.1569+4816A>T
NM_001367251.1:c.1386+4816A>T
NR_159803.1:n.2263+951A>T
NR_159804.1:n.1648+4816A>T
NR_159805.1:n.1714+4816A>T
NR_159806.1:n.1866+951A>T

NR_159807.1:n.1622+4816A>T
NR_159808.1:n.1826+4816A>T
NM_000328.3:c.1905+951A>T

Benign

Met criteria codes 2

BP4_Stand Alone | BA1

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.2856A>T (p.Glu952Asp) is a missense variant causing substitution of glutamic acid by aspartic acid at amino acid 952. This variant is present in gnomAD v4.1.0 at a frequency of 0.0001313 among hemizygous individuals, with 10 variant alleles / 76,136 total alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.00005 (BA1). The computational predictor REVEL gives a score of 0.003, which is below the ClinGen X-linked IRD VCEP threshold of less than or equal to 0.003 and predicts a non-damaging effect on RPGR function. Additionally, the splicing impact predictor SpliceAI gives a delta score of 0.00, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4_Very-Strong). In summary, this variant is classified as 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; BA1 and BP4_Very-Strong.

Met criteria codes

BP4_Stand Alone	✔	The computational predictor REVEL gives a score of 0.003, which is below the ClinGen X-linked IRD VCEP threshold of < 0.003 and predicts a non-damaging effect on RPGR function. Additionally, the splicing impact predictor SpliceAI gives a delta score of 0.00, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4_VeryStrong).
BA1	✔	This variant is present in gnomAD v.4.1.0 at a frequency of 0.0001313 among hemizygous individuals, with 10 variant alleles / 76136 total alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.00005 (BA1).

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001034853.2(RPGR):c.2856A>T (p...	Benign	RPGR-Related Retinopathy	2025-09-05	1.0	ClinGen X-linked Inherited Retinal Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RPGR Version 1.0.0	RPGR

Showing 1 to 1 of 1 rows

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