Variant: NM_001034853.2(RPGR):c.552G>T (p.Gln184His)

Version: 1.0

CA10385632

255836 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: baf6cb27-7358-4016-a937-da7795cf76f4
Approved on: 2025-09-07
Published on: 2025-09-07

HGVS expressions

NM_001034853.2:c.552G>T
NM_001034853.2(RPGR):c.552G>T (p.Gln184His)
NC_000023.11:g.38317383C>A
CM000685.2:g.38317383C>A
NC_000023.10:g.38176636C>A
CM000685.1:g.38176636C>A
NC_000023.9:g.38061580C>A
NG_009553.1:g.15153G>T
ENST00000642170.1:n.962G>T
ENST00000642373.1:c.*131G>T
ENST00000642395.2:c.552G>T
ENST00000642558.1:c.459G>T
ENST00000642739.1:c.552G>T
ENST00000644238.1:c.552G>T
ENST00000644337.1:c.552G>T
ENST00000645032.1:c.552G>T
ENST00000645124.1:c.552G>T
ENST00000646020.1:c.552G>T
ENST00000647261.1:c.552G>T
ENST00000318842.11:c.552G>T
ENST00000339363.7:c.552G>T
ENST00000378505.6:c.552G>T
ENST00000465127.1:c.172-348738C>A
ENST00000470183.1:n.245G>T
ENST00000474584.5:c.552G>T
ENST00000482855.5:c.552G>T
NM_000328.2:c.552G>T
NM_001034853.1:c.552G>T
NM_001367245.1:c.552G>T
NM_001367246.1:c.552G>T
NM_001367247.1:c.552G>T
NM_001367248.1:c.582G>T
NM_001367249.1:c.549G>T
NM_001367250.1:c.552G>T
NM_001367251.1:c.552G>T
NR_159803.1:n.694G>T
NR_159804.1:n.670+24G>T

NR_159805.1:n.694G>T
NR_159806.1:n.694G>T
NR_159807.1:n.694G>T
NR_159808.1:n.962G>T
NM_000328.3:c.552G>T

Benign

Met criteria codes 2

BA1 BP4

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.552G>T (p.Gln184His) is a missense variant causing substitution of glutamine by histidine at amino acid 184. This variant is present in gnomAD v4.1.0 at a frequency of 0.003034 among hemizygous individuals, with 1,194 variant alleles / 393,499 total hemizygous alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.000005 (BA1). The computational predictor REVEL gives a score of 0.17, which is below the ClinGen X-linked IRD VCEP threshold of <0.183 and predicts a non-damaging effect on RPGR function. Additionally, the splicing impact predictor SpliceAI gives a delta score of 0.03, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4_Moderate). 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_Moderate..

Met criteria codes

BA1	✔	This variant is present in gnomAD v.4.1.0 at a frequency of 0.003034 among hemizygous individuals, with 1,194 variant alleles/393,499 total hemizygous alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.000005 (BA1).
BP4	✔	The computational predictor REVEL gives a score of 0.17, which is below the ClinGen X-linked IRD VCEP threshold of <0.183 and predicts a non-damaging effect on RPGR function. Additionally, the splicing impact predictor SpliceAI gives a delta score of 0.03, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4_Moderate).

Curation History

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