

Variant: *NM_001034853.2(RPGR):c.141T>G (p.Ser47=)*

Version: 1.0

CA226360 [↗](#)

98741 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: addc7984-3ab1-4e7c-9ca2-8980bd438504

Approved on: 2025/10/5

Published on: 2025/10/6

HGVS expressions

NM_001034853.2(RPGR):c.141T>G (p.Ser47=)

NM_001034853.2:c.141T>G

NC_000023.11:g.38323412A>C

CM000685.2:g.38323412A>C

NC_000023.10:g.38182665A>C

CM000685.1:g.38182665A>C

NC_000023.9:g.38067609A>C

NG_009553.1:g.9124T>G

ENST00000642170.1:n.551T>G

ENST00000642373.1:c.141T>G

ENST00000642395.2:c.141T>G

ENST00000642558.1:c.141T>G

ENST00000642739.1:c.141T>G

ENST00000644238.1:c.141T>G

ENST00000644337.1:c.141T>G

ENST00000645032.1:c.141T>G

ENST00000645124.1:c.141T>G

ENST00000646020.1:c.141T>G

ENST00000647261.1:c.141T>G

ENST00000318842.11:c.141T>G

ENST00000339363.7:c.141T>G

ENST00000378505.6:c.141T>G

ENST00000465127.1:c.172-342709A>C

ENST00000474584.5:c.141T>G

ENST00000482855.5:c.141T>G

NM_000328.2:c.141T>G

NM_001034853.1:c.141T>G

NM_001367245.1:c.141T>G

NM_001367246.1:c.141T>G

NM_001367247.1:c.141T>G

NM_001367248.1:c.171T>G

NM_001367249.1:c.141T>G

NM_001367250.1:c.141T>G

NM_001367251.1:c.141T>G

NR_159803.1:n.283T>G

NR_159804.1:n.283T>G

NR_159805.1:n.283T>G

NR_159806.1:n.283T>G
NR_159807.1:n.283T>G
NR_159808.1:n.551T>G
NM_000328.3:c.141T>G

Benign

Met criteria codes 3

BA1 BP4 BP7

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.141T>G (p.Ser47=) is a synonymous variant in exon 2 of RPGR (BP7). 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). This variant is present in gnomAD v.4.1.0 at a frequency of 0.0004126 among hemizygous individuals, with 162 variant alleles / 392,654 total hemizygous alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.00005 (BA1). 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, BP4, and BP7.

Met criteria codes

BA1			This variant is present in gnomAD v.4.1.0 at a frequency of 0.0004126 among hemizygous individuals, with 162 variant alleles/392654 total hemizygous alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.00005 (BA1).
BP4			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).
BP7			This silent variant NM_001034853.2(RPGR):c.141T>G causing a synonymous variant at codon 42 does not have an impact at splicing sites according to SpliceAI, which predicts a delta score of 0.00 for all splicing types, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP7).

Curation History

▾

▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.