Variant: NM_001034853.2(RPGR):c.1059+6G>A

Version: 1.0

CA10385560

255828 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 419e2509-9f07-4f06-8a67-8c919cbe6dd4
Approved on: 2025-08-01
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HGVS expressions

NM_001034853.2:c.1059+6G>A
NM_001034853.2(RPGR):c.1059+6G>A
NC_000023.11:g.38301241C>T
CM000685.2:g.38301241C>T
NC_000023.10:g.38160494C>T
CM000685.1:g.38160494C>T
NC_000023.9:g.38045438C>T
NG_009553.1:g.31295G>A
ENST00000494707.6:c.263+6G>A
ENST00000642170.1:n.1313+6G>A
ENST00000642395.2:c.1059+6G>A
ENST00000642558.1:c.966+6G>A
ENST00000642739.1:c.1059+6G>A
ENST00000644238.1:c.1059+6G>A
ENST00000644337.1:c.1059+6G>A
ENST00000645032.1:c.1059+6G>A
ENST00000645124.1:c.1059+6G>A
ENST00000646020.1:c.1119+6G>A
ENST00000318842.11:c.1059+6G>A
ENST00000339363.7:c.1059+6G>A
ENST00000378505.6:c.1059+6G>A
ENST00000464437.1:c.125+6G>A
ENST00000465127.1:c.172-364880C>T
ENST00000474584.5:c.1059+6G>A
ENST00000482855.5:c.1059+6G>A
ENST00000494841.1:n.322+6G>A
NM_000328.2:c.1059+6G>A
NM_001034853.1:c.1059+6G>A
NM_001367245.1:c.1056+6G>A
NM_001367246.1:c.1059+6G>A
NM_001367247.1:c.1059+6G>A
NM_001367248.1:c.1089+6G>A
NM_001367249.1:c.1056+6G>A
NM_001367250.1:c.1056+6G>A
NM_001367251.1:c.1059+6G>A
NR_159803.1:n.1261+6G>A
NR_159804.1:n.1110+6G>A

NR_159805.1:n.1201+6G>A
NR_159806.1:n.1201+6G>A
NR_159807.1:n.1201+6G>A
NR_159808.1:n.1313+6G>A
NM_000328.3:c.1059+6G>A

Benign

Met criteria codes 2

BA1 BP4

Not Met criteria codes 3

PM2 PM1 BP3

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.1059+6G>A an intron 9 variant located six nucleotides after exon 9. This variant is present in gnomAD v4.1.0 at a frequency of 0.0002996 among hemizygous individuals, with 114 variant alleles / 380,467 total alleles, which is higher than the ClinGen X-linked IRD VCEP BA1 threshold of >0.00005 (BA1). The splicing impact predictor SpliceAI gives a delta score of 0.01, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4). 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. (date of approval 05/16/2025).

Met criteria codes

BA1	✔	This variant is present in gnomAD v.4.1.0 at a frequency of 0.0002996 among hemizygous individuals, with 114 variant alleles / 380,467 total 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 0.01, which is below the ClinGen X-linked IRD VCEP recommended threshold of <0.1 and does not strongly predict an impact on splicing (BP4).

Not Met criteria codes

PM2	✖	This variant is present in gnomAD v.2.1.1 at a frequency of [0.0002996] among hemizygous individuals, with [114] variant alleles / [380467] total alleles, which is higher than the ClinGen X-linked IRD VCEP PM2_Supporting threshold of <0.0000005 and fails to meet this criterion.
PM1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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