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Variant: NM_001034853.2(RPGR):c.1693C>T (p.Gln565Ter)

Version: 1.0

CA412736970

2907569 (ClinVar)

Gene: RPGR (HGNC:6103)
Condition: RPGR-related retinopathy (MONDO:0100437)
Inheritance Mode: X-linked inheritance
UUID: 8b6340ac-c705-4eb4-b43c-4fee589a452c
Approved on: 2026/1/25
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HGVS expressions

NM_001034853.2(RPGR):c.1693C>T (p.Gln565Ter)
NM_001034853.2:c.1693C>T
NC_000023.11:g.38287921G>A
CM000685.2:g.38287921G>A
NC_000023.10:g.38147174G>A
CM000685.1:g.38147174G>A
NC_000023.9:g.38032118G>A
NG_009553.1:g.44615C>T
ENST00000494707.6:c.897C>T
ENST00000642170.1:n.1826+3038C>T
ENST00000642395.2:c.1693C>T
ENST00000642739.1:c.1572+3038C>T
ENST00000644238.1:c.1386+3038C>T
ENST00000644337.1:c.1507C>T
ENST00000645032.1:c.1693C>T
ENST00000645124.1:c.1573-676C>T
ENST00000646020.1:c.*386C>T
ENST00000318842.11:c.1693C>T
ENST00000339363.7:c.1693C>T
ENST00000378505.6:c.1693C>T
ENST00000465127.1:c.172-378200G>A
ENST00000474584.5:c.*37+3038C>T
ENST00000482855.5:c.1693C>T
ENST00000494707.5:c.139+3038C>T
NM_000328.2:c.1693C>T
NM_001034853.1:c.1693C>T
NM_001367245.1:c.1690C>T
NM_001367246.1:c.1507C>T
NM_001367247.1:c.1572+3038C>T
NM_001367248.1:c.1602+3038C>T
NM_001367249.1:c.1569+3038C>T
NM_001367250.1:c.1569+3038C>T
NM_001367251.1:c.1386+3038C>T
NR_159803.1:n.2055C>T
NR_159804.1:n.1648+3038C>T
NR_159805.1:n.1714+3038C>T
NR_159806.1:n.1715-676C>T

NR_159807.1:n.1622+3038C>T
NR_159808.1:n.1826+3038C>T
NM_000328.3:c.1693C>T

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PP1_Moderate

Not Met criteria codes 2

PS4 PP4

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.1693C>T (p.Gln565Ter) is a nonsense variant in codon 565 that introduces a premature stop codon into exon 14 of 15, and is predicted to lead to nonsense-mediated decay in the RPGR gene in which loss-of-function is an established mechanism of disease (PVS1). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). At least one female proband (>40) harboring this variant has been confirmed to have an affected male relative and exhibits a delayed phenotype including night blindness, and moderately decreased cone and rod electroretinogram responses (PMID: 31645972). The variant has been reported to segregate with retinal dystrophy through at least 3 affected meioses from 1 family (PP1_Moderate; PMID: 31645972). In summary, this variant is classified as pathogenic 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; PVS1, PM2_Supporting, and PP1_Moderate.

Met criteria codes

PVS1	✔	This is a nonsense variant that introduces a premature stop codon into exon 14 of 15, and is predicted to lead to nonsense-mediated decay in the RPGR gene in which loss-of-function is an established mechanism of disease (PVS1).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP1_Moderate	✔	The variant has been reported to segregate with retinal dystrophy through at least 3 affected meioses from 1 family (PMID: 31645972, PP1_Moderate).

Not Met criteria codes

PS4	✖	This variant has been reported in at least 1 family (PMID: 31645972). However, the PS4 requirement of some functional vision impairment in affected males by age 30, with decreased or absent cone and/or rod ERG responses was not given for the affected male, so PS4 was not met.

PP4		✖	At least one female proband (>40) harboring this variant exhibits a phenotype including night blindness, moderately decreased cone and rod responses (ERG), and a delayed phenotype. Since no male proband (<30) was described, the required criteria to use PP4 are not met.

Curation History

Showing 1 to 1 of 1 rows

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