Variant: NM_002709.3(PPP1CB):c.201A>G (p.Gln67=)

CA1589199

667025 (ClinVar)

Gene: PPP1CB (HGNC:5500)
Condition: RASopathy (MONDO:0021060)
Inheritance Mode: Autosomal dominant inheritance
UUID: 92c34e06-f193-405c-a099-e2ab6cede4d0
Approved on: 2024-12-03
Published on: 2025-03-25

HGVS expressions

NM_002709.3:c.201A>G
NM_002709.3(PPP1CB):c.201A>G (p.Gln67=)
NC_000002.12:g.28778825A>G
CM000664.2:g.28778825A>G
NC_000002.11:g.29001691A>G
CM000664.1:g.29001691A>G
NC_000002.10:g.28855195A>G
NG_052878.1:g.32078A>G
ENST00000418910.2:c.201A>G
ENST00000420282.6:c.201A>G
ENST00000427786.2:c.*161A>G
ENST00000441461.6:c.201A>G
ENST00000455580.6:c.117A>G
ENST00000703171.1:c.*248A>G
ENST00000703172.1:c.117A>G
ENST00000703173.1:c.201A>G
ENST00000703174.1:c.201A>G
ENST00000703176.1:c.168A>G
ENST00000703177.1:c.*161A>G
ENST00000395366.3:c.201A>G
ENST00000296122.10:c.201A>G
ENST00000358506.6:c.201A>G
ENST00000395366.2:c.201A>G
ENST00000420282.5:c.201A>G
ENST00000427786.1:c.*161A>G
ENST00000441461.5:c.201A>G
ENST00000455580.5:c.117A>G
NM_002709.2:c.201A>G
NM_206876.1:c.201A>G
NM_206876.2:c.201A>G

Benign

Met criteria codes 3

BA1 BP7 BP4

Evidence Links 0

Expert Panel

RASopathy VCEP

Criteria Specification Information

Criteria Specification: *ClinGen RASopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PPP1CB Version 1.3.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

RASopathy VCEP

The c.201A>G (p.Gln67=) variant in PPP1CB is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, the computational predictor REVEL does not predict a damaging effect on PPP1CB function (BP4, BP7). The filtering allele frequency in gnomAD v2 is 60.64% in the European (non-Finnish) population, which is higher than the ClinGen RASopathy VCEP threshold (>0.0005) for BA1, and therefore meets this criterion (BA1). In summary, this variant meets the criteria to be classified as benign for autosomal dominant RASopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen RASopathy VCEP: BA1, BP4, BP7. (RASopathy VCEP specifications version 1.3; 12/3/2024)

Met criteria codes

BA1	✔	The highest population minor allele frequency in gnomAD v4 is 62.67% (3738/5964) in the Middle Eastern population, which is higher than the ClinGen RASopathy VCEP threshold (>0.0005) for BA1
BP7	✔	The variant is not predicted by SpliceAI to impact splicing
BP4	✔	The computational predictor REVEL does not predict a damaging effect on PPP1CB function

Curation History

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene

No matching records found