Variant: NM_002709.3(PPP1CB):c.53-9G>A

Version: 1.0

CA1589161

747303 (ClinVar)

Gene: PPP1CB (HGNC:5500)
Condition: RASopathy (MONDO:0021060)
Inheritance Mode: Autosomal dominant inheritance
UUID: 0c73b719-a9d3-4ddf-ad9f-d72869456e33
Approved on: 2024-12-03
Published on: 2025-03-25

HGVS expressions

NM_002709.3:c.53-9G>A
NM_002709.3(PPP1CB):c.53-9G>A
NC_000002.12:g.28776842G>A
CM000664.2:g.28776842G>A
NC_000002.11:g.28999708G>A
CM000664.1:g.28999708G>A
NC_000002.10:g.28853212G>A
NG_052878.1:g.30095G>A
ENST00000418910.2:c.53-9G>A
ENST00000420282.6:c.53-9G>A
ENST00000427786.2:c.-32-9G>A
ENST00000441461.6:c.53-9G>A
ENST00000455580.6:c.-32-9G>A
ENST00000703171.1:c.53-9G>A
ENST00000703172.1:c.-32-9G>A
ENST00000703173.1:c.53-9G>A
ENST00000703174.1:c.53-9G>A
ENST00000703176.1:c.20-9G>A
ENST00000703177.1:c.-32-9G>A
ENST00000395366.3:c.53-9G>A
ENST00000296122.10:c.53-9G>A
ENST00000358506.6:c.53-9G>A
ENST00000395366.2:c.53-9G>A
ENST00000420282.5:c.53-9G>A
ENST00000427786.1:c.-32-9G>A
ENST00000441461.5:c.53-9G>A
ENST00000455580.5:c.-32-9G>A
ENST00000464273.1:n.167-9G>A
NM_002709.2:c.53-9G>A
NM_206876.1:c.53-9G>A
NM_206876.2:c.53-9G>A

Benign

Met criteria codes 1

BA1

Expert Panel

RASopathy VCEP

Evidence Links 0

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.53-9G>A variant in PPP1CB is an intronic splicing variant located in intron 1 of PPP1CB. The filtering allele frequency in gnomAD v2 is 0.5284% in the African American 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. (RASopathy VCEP specifications version 1.3; 12/3/2024)

Met criteria codes

BA1	ℹ	✔	This variant has a minor allele frequency of 0.5599% (417/74482) in the African American population in gnomAD v4, which is higher than the ClinGen RASopathy VCEP threshold (>0.0005) for BA1

Curation History

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