

Variant: NM_002834.4(PTPN11):c.48A>G (p.Ala16=)

CA134668 [↗](#)

44608 (ClinVar) [↗](#)

Gene: PTPN11 ([HGNC:5781](#))

Condition: RASopathy ([MONDO:0021060](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 44c75d04-cfb3-4a52-840e-30d2eb641915

Approved on: 2017-04-18

Published on: 2018-12-10

HGVS expressions

NM_002834.4:c.48A>G

NM_002834.4(PTPN11):c.48A>G (p.Ala16=)

NM_002834.3:c.48A>G

NM_080601.1:c.48A>G

NM_001330437.1:c.48A>G

NM_080601.2:c.48A>G

ENST00000351677.6:c.48A>G

ENST00000392597.5:c.48A>G

ENST00000635625.1:n.48A>G

NC_000012.12:g.112446309A>G

CM000674.2:g.112446309A>G

NC_000012.11:g.112884113A>G

CM000674.1:g.112884113A>G

NC_000012.10:g.111368496A>G

NG_007459.1:g.32578A>G

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Insufficient Evidence"

Met criteria codes **1**

BS1

Evidence Links **0**

Expert Panel

[RASopathy VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

RASopathy VCEP

The filtering allele frequency of the c.48A>G (p.Ala16=) variant in the PTPN11 gene is 0.0284% (9/16512) of South Asian chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Met criteria codes

BS1





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Curation History

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