

Variant: NM_004333.4(BRAF):c.36G>A (p.Ala12=)

CA281921 [↗](#)

40333 (ClinVar) [↗](#)

Gene: BRAF ([HGNC:673](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 37d415c0-90e7-425c-84bc-6361413d50aa

Approved on: 2017-04-18

Published on: 2018-12-10

HGVS expressions

NM_004333.4:c.36G>A

NM_004333.4(BRAF):c.36G>A (p.Ala12=)

NM_001354609.1:c.36G>A

NM_004333.5:c.36G>A

NR_148928.1:n.261G>A

ENST00000288602.10:c.36G>A

ENST00000469930.1:n.42G>A

NC_000007.14:g.140924668C>T

CM000669.2:g.140924668C>T

NC_000007.13:g.140624468C>T

CM000669.1:g.140624468C>T

NC_000007.12:g.140270937C>T

NG_007873.3:g.5097G>A

Benign

Met criteria codes **1**

BA1

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.36G>A (p.Ala12=) variant in the BRAF gene is 0.116% (14/7290) of Latino chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Met criteria codes

BA1



The filtering allele frequency of the c.36G>A (p.Ala12=) variant in the BRAF gene is 0.116% (1/90) of Latino chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Curation History [↗](#)

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