

*Variant: NM\_004333.4(BRAF):c.1383A>G (p.Gln461=)*

CA135073 [↗](#)

40363 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** ab868908-3f55-4346-8bad-adcda350acb4

**Approved on:** 2017-04-18

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### HGVS expressions

**NM\_004333.4:c.1383A>G**

**NM\_004333.4(BRAF):c.1383A>G (p.Gln461=)**

NM\_001354609.1:c.1383A>G

NM\_004333.5:c.1383A>G

NR\_148928.1:n.1688A>G

ENST00000288602.10:c.1383A>G

ENST00000496384.6:n.206A>G

ENST00000497784.1:n.1418A>G

NC\_000007.14:g.140781625T>C

CM000669.2:g.140781625T>C

NC\_000007.13:g.140481425T>C

CM000669.1:g.140481425T>C

NC\_000007.12:g.140127894T>C

NG\_007873.3:g.148140A>G

**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.1383A>G (p.Gln461=) variant in the BRAF gene is 0.986% (120/10404) of African 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**



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Curation History 



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