

Variant: NM_004333.4(BRAF):c.1929A>G (p.Gly643=)

CA135110 [↗](#)

40391 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 043632b0-38d9-48f7-b0b4-993f0aa553b4

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HGVS expressions

NM_004333.4:c.1929A>G

NM_004333.4(BRAF):c.1929A>G (p.Gly643=)

NC_000007.14:g.140749350T>C

CM000669.2:g.140749350T>C

NC_000007.13:g.140449150T>C

CM000669.1:g.140449150T>C

NC_000007.12:g.140095619T>C

NG_007873.3:g.180415A>G

NM_001354609.1:c.1929A>G

NM_004333.5:c.1929A>G

NR_148928.1:n.3027A>G

ENST00000288602.10:c.1929A>G

ENST00000479537.5:n.213A>G

ENST00000496384.6:n.752A>G

ENST00000497784.1:n.1964A>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.1929A>G (p.Gly643=) variant in the BRAF gene is 66.117% (6984/10356) 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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