

Variant: NM_004333.4(BRAF):c.375T>G (p.Ser125=)

CA135125 [↗](#)

44825 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0fdcd6c2-d132-4727-b944-17e200706a31

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Published on: 2018-12-10

HGVS expressions

NM_004333.4(BRAF):c.375T>G (p.Ser125=)

NM_004333.4:c.375T>G

NC_000007.14:g.140834738A>C

CM000669.2:g.140834738A>C

NC_000007.13:g.140534538A>C

CM000669.1:g.140534538A>C

NC_000007.12:g.140181007A>C

NG_007873.3:g.95027T>G

NM_001354609.1:c.375T>G

NM_004333.5:c.375T>G

NR_148928.1:n.600T>G

ENST00000288602.10:c.375T>G

ENST00000469930.1:n.381T>G

ENST00000497784.1:n.330T>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.375T>G (p.Ser125=) variant in the BRAF gene is 0.054% (9/8644) of East Asian 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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