

Variant: NM_004333.4(BRAF):c.111G>A (p.Ser37=)

CA175346 [↗](#)

162804 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 7e872ce6-f7c9-4c3d-a39e-8a174058fd77

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HGVS expressions

NM_004333.4(BRAF):c.111G>A (p.Ser37=)

NM_004333.4:c.111G>A

NC_000007.14:g.140924593C>T

CM000669.2:g.140924593C>T

NC_000007.13:g.140624393C>T

CM000669.1:g.140624393C>T

NC_000007.12:g.140270862C>T

NG_007873.3:g.5172G>A

NM_001354609.1:c.111G>A

NM_004333.5:c.111G>A

NR_148928.1:n.336G>A

ENST00000288602.10:c.111G>A

ENST00000469930.1:n.117G>A

ENST00000497784.1:n.66G>A

Likely Benign

Met criteria codes **2**

BP5 **BP7**

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

RASopathy VCEP

The c.111G>A (p.Ser37=) variant in BRAF is a synonymous (silent) variant at a nucleotide that is not highly conserved and is not predicted to impact splicing (BP7). This variant has been identified in a patient with an alternate molecular basis for disease (BP5; Partners LMM, GeneDx internal data, GTR ID's: SCV000197179.4, SCV000512270.5). In summary, this variant meets criteria to be classified as likely benign. RASopathy-specific ACMG/AMP criteria applied (PMID:29493581): BP7, BP5.

Met criteria codes

BP5



	This variant has been identified in a patient with an alternate molecular basis for disease (BP5; Partners LMM, GeneDx internal data, GTR ID's: SCV000197179.4, SCV000512270.5).	
BP7	 	The c.111G>A (p.Ser37=) variant is a synonymous (silent) variant at a nucleotide that is not highly conserved and is not predicted to impact splicing (BP7).

Curation History 	
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