

Variant: NM_002880.3(RAF1):c.1755A>G (p.Val585=)

CA134709 [↗](#)

40626 (ClinVar) [↗](#)

Gene: RAF1 ([HGNC:5894](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d1aca5e8-fdc6-4f63-acc4-05595b97c993

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HGVS expressions

NM_002880.3:c.1755A>G

NM_002880.3(RAF1):c.1755A>G (p.Val585=)

NM_001354689.1:c.1815A>G

NM_001354690.1:c.1755A>G

NM_001354691.1:c.1512A>G

NM_001354692.1:c.1512A>G

NM_001354693.1:c.1656A>G

NM_001354694.1:c.1572A>G

NM_001354695.1:c.1413A>G

NR_148940.1:n.2283A>G

NR_148941.1:n.2229A>G

NR_148942.1:n.2168A>G

ENST00000251849.8:c.1755A>G

ENST00000423275.5:c.*1432A>G

ENST00000432427.2:n.1392A>G

ENST00000442415.6:c.1815A>G

ENST00000471449.1:n.444A>G

NC_000003.12:g.12584895T>C

CM000665.2:g.12584895T>C

NC_000003.11:g.12626394T>C

CM000665.1:g.12626394T>C

NC_000003.10:g.12601394T>C

NG_007467.1:g.84285A>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.1755A>G (p.Val585=) variant in the RAF1 gene is 6.222% (690/10404) 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			The filtering allele frequency of the c.1755A>G (p.Val585=) variant in the RAF1 gene is 6.222% (690/10404) 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)
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Curation History



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