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Variant: NM_002880.3(RAF1):c.1830A>G (p.Gln610=)

CA134712

40627 (ClinVar)

Gene: RAF1 (HGNC:5894)
Condition: RASopathy (MONDO:0021060)
Inheritance Mode: Autosomal dominant inheritance
UUID: 04430f0e-93d8-4407-9ac9-c6268cb1dbba
Approved on: 2017-04-18
Published on: 2018-12-10

HGVS expressions

NM_002880.3(RAF1):c.1830A>G (p.Gln610=)
NM_002880.3:c.1830A>G
NC_000003.12:g.12584631T>C
CM000665.2:g.12584631T>C
NC_000003.11:g.12626130T>C
CM000665.1:g.12626130T>C
NC_000003.10:g.12601130T>C
NG_007467.1:g.84549A>G
NM_001354689.1:c.1890A>G
NM_001354690.1:c.1830A>G
NM_001354691.1:c.1587A>G
NM_001354692.1:c.1587A>G
NM_001354693.1:c.1731A>G
NM_001354694.1:c.1647A>G
NM_001354695.1:c.1488A>G
NR_148940.1:n.2358A>G
NR_148941.1:n.2304A>G
NR_148942.1:n.2243A>G
ENST00000251849.8:c.1830A>G
ENST00000423275.5:c.*1507A>G
ENST00000432427.2:n.1467A>G
ENST00000442415.6:c.1890A>G
ENST00000471449.1:n.519A>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.1830A>G (p.Gln610=) variant in the RAF1 gene is 0.11% (88/66740) of European 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.1830A>G (p.Gln610=) variant in the RAF1 gene is 0.11% (88/66740) of European 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)

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_002880.3(RAF1):c.1830A>G (p.Gln...	Benign	RASopathy	2018-12-10	1.0	-	RAF1

Showing 1 to 1 of 1 rows

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