

Variant: NM_002880.3(RAF1):c.1141G>A (p.Asp381Asn)

CA297118 [↗](#)

181509 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: f46177ab-5c80-48cf-bd17-6acc5668f182

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HGVS expressions

NM_002880.3(RAF1):c.1141G>A (p.Asp381Asn)

NM_002880.3:c.1141G>A

NM_001354689.1:c.1201G>A

NM_001354690.1:c.1141G>A

NM_001354691.1:c.898G>A

NM_001354692.1:c.898G>A

NM_001354693.1:c.1042G>A

NM_001354694.1:c.958G>A

NM_001354695.1:c.799G>A

NR_148940.1:n.1669G>A

NR_148941.1:n.1615G>A

NR_148942.1:n.1554G>A

ENST00000251849.8:c.1141G>A

ENST00000423275.5:c.*818G>A

ENST00000432427.2:n.778G>A

ENST00000442415.6:c.1201G>A

ENST00000460610.1:n.98G>A

ENST00000465826.5:n.498G>A

ENST00000475353.1:n.309G>A

ENST00000494557.1:n.157G>A

NC_000003.12:g.12591760C>T

CM000665.2:g.12591760C>T

NC_000003.11:g.12633259C>T

CM000665.1:g.12633259C>T

NC_000003.10:g.12608259C>T

NG_007467.1:g.77420G>A

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information **!**

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

RASopathy VCEP

The filtering allele frequency of the c.1141G>A (p.Asp381Asn) variant in the RAF1 gene is 0.162% (36/16508) of South 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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