

Variant: NM_002524.5(NRAS):c.250A>G (p.Ile84Val)

Version: 1.1

CA1020748 [↗](#)

424299 (ClinVar) [↗](#)

Gene: NRAS ([HGNC:4893](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 21f25196-ecca-494f-bb6c-53ab02e41537

Approved on: 2024/12/3

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HGVS expressions

NM_002524.5:c.250A>G

NM_002524.5(NRAS):c.250A>G (p.Ile84Val)

NC_000001.11:g.114713840T>C

CM000663.2:g.114713840T>C

NC_000001.10:g.115256461T>C

CM000663.1:g.115256461T>C

NC_000001.9:g.115057984T>C

NG_007572.1:g.8055A>G

ENST00000369535.5:c.250A>G

ENST00000369535.4:c.250A>G

NM_002524.4:c.250A>G

Uncertain Significance

Met criteria codes **1**

BP4

Not Met criteria codes **3**

PM2

BS1

BA1

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen RASopathy Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for NRAS Version 2.3.0

[↗](#) **Criteria Specification Approval History**

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

Evidence submitted by expert panel

RASopathy VCEP

The NM_002524.5:c.250A>G variant in NRAS is a missense variant predicted to cause substitution of isoleucine by valine at amino acid 84 (p.Ile84Val). The highest population minor allele frequency in gnomAD v2.1.1 is 0.00003870 (5/129184 alleles) in the European (non-Finnish) population (PM2_Supporting, BS1, and BA1 are not met). The computational predictor REVEL gives a score of 0.247, which is below the threshold of 0.3, evidence that does not predict a damaging effect on NRAS function (BP4). In summary, this variant meets the criteria to be classified as uncertain significance for autosomal dominant RASopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen RASopathy VCEP: BP4. (ClinGen RASopathy VCEP specifications version 2.3; 12/3/2024)

Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.247, which is below the threshold of 0.3, evidence that does not predict a damaging effect on NRAS function (BP4).
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Not Met criteria codes

PM2			The highest population minor allele frequency in gnomAD v2.1.1 is 0.00003870 (5/129184 alleles) in the European (non-Finnish) population (PM2_Supporting, BS1, and BA1 are not met).
BS1			0.000015% European alleles (5/129184) v2 0.0014% European alleles (1/68028) v3
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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