

Variant: NM_002834.4(PTPN11):c.1678C>T (p.Leu560Phe)

CA134647 [↗](#)

44599 (ClinVar) [↗](#)

Gene: PTPN11 ([HGNC:5781](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1de1c18c-c026-4034-afda-2b4516c6cdae

Approved on: 2017-04-03

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HGVS expressions

NM_002834.4:c.1678C>T

NM_002834.4(PTPN11):c.1678C>T (p.Leu560Phe)

NC_000012.12:g.112502222C>T

CM000674.2:g.112502222C>T

NC_000012.11:g.112940026C>T

CM000674.1:g.112940026C>T

NC_000012.10:g.111424409C>T

NG_007459.1:g.88491C>T

ENST00000639857.2:c.1678C>T

ENST00000685487.1:c.*880C>T

ENST00000687120.1:n.1061C>T

ENST00000687906.1:c.1564C>T

ENST00000688597.1:c.1303C>T

ENST00000688701.1:n.922C>T

ENST00000690210.1:c.1678C>T

ENST00000690472.1:n.887C>T

ENST00000692624.1:c.*224C>T

ENST00000351677.7:c.1678C>T

ENST00000351677.6:c.1678C>T

ENST00000635625.1:c.1690C>T

NM_002834.3:c.1678C>T

NM_001330437.1:c.1690C>T

NM_001330437.2:c.1690C>T

NM_001374625.1:c.1675C>T

NM_002834.5:c.1678C>T

Likely Benign

Met criteria codes **2**

BS3 **BP5**

Evidence Links **1**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen RASopathy Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for PTPN11 Version 2.1.0

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

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

RASopathy VCEP

The c.1678C>T (p.Leu560Phe) variant in PTPN11 (NM_002834.5(PTPN11):c.1678C>T (p.Leu560Phe)) has been identified in individuals with some features of a RASopathy but none were diagnosed with a RASopathy (PS4 not met; GeneDx, Partners LMM, APHP-Robert Debré Hospital internal data; GTR ID's: 28338, 26957, 21766; SCV000061285.5; SCV000208999.2; SCV000207690.1). This variant has been identified in a patient with an alternate molecular basis for disease (BP5; GeneDx internal data: GTR ID: 26957; SCV000208999.2). In vitro functional studies provide some evidence that the p.Leu560Phe variant does not impact protein function (BS3; PMID: 15987685). In summary, this variant meets criteria to be classified as likely benign. RASopathy-specific ACMG/AMP criteria applied: BS3, BP5 (Version 2.1; 09/17/2024).

Met criteria codes

BS3		In vitro functional studies provide some evidence that the p.Leu560Phe variant does not impact protein function (BS3; PMID: 15987685).
		In vitro functional studies provide some evidence that the p.Leu560Phe variant does not impact protein function (BS3; PMID: 15987685). PubMed:15987685
BP5		This variant has been identified in a patient with an alternate molecular basis for disease (BP5; GeneDx internal data: GTR ID: 26957; SCV000208999.2).

Curation History

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
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No matching records found