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Variant: NM_001083962.2(TCF4):c.466C>A (p.Pro156Thr)

Version: 1.0

CA8970510

664780 (ClinVar)

Gene: TCF4 (HGNC:6925)
Condition: Pitt-Hopkins syndrome (MONDO:0012589)
Inheritance Mode: Autosomal dominant inheritance
UUID: 009efeaf-274d-43f3-83d3-095679e3c249
Approved on: 2024/6/25
Published on: 2024/8/23

HGVS expressions

NM_001083962.2(TCF4):c.466C>A (p.Pro156Thr)
NM_001083962.2:c.466C>A
NC_000018.10:g.55350907G>T
CM000680.2:g.55350907G>T
NC_000018.9:g.53018138G>T
CM000680.1:g.53018138G>T
NC_000018.8:g.51169136G>T
NG_011716.1:g.242723C>A
NG_011716.2:g.290087C>A
ENST00000354452.8:c.466C>A
ENST00000635822.2:c.466C>A
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ENST00000636400.2:c.394C>A
ENST00000636751.2:c.*174C>A
ENST00000636822.2:c.76C>A
ENST00000637115.2:c.*356C>A
ENST00000637239.2:n.533C>A
ENST00000637250.2:n.160C>A
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ENST00000674764.1:c.*77C>A
ENST00000675707.1:c.76C>A
ENST00000354452.7:c.466C>A
ENST00000356073.8:c.466C>A
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ENST00000537578.5:c.394C>A
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ENST00000543082.5:c.340C>A
ENST00000544241.6:c.253C>A
ENST00000561992.5:c.76C>A
ENST00000562030.3:c.76C>A
ENST00000562607.5:c.76C>A
ENST00000562638.5:c.76C>A
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ENST00000563686.5:n.321C>A
ENST00000563824.5:c.394C>A

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ENST00000564228.5:c.253C>A
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ENST00000564999.5:c.466C>A
ENST00000565018.6:c.297+52547C>A
ENST00000565580.3:n.298+52547C>A
ENST00000565908.6:c.394C>A
ENST00000566279.5:c.369+52547C>A
ENST00000566286.5:c.460C>A
ENST00000566376.3:n.146C>A
ENST00000566514.5:c.427C>A
ENST00000566777.5:c.76C>A
ENST00000567880.5:c.369+52547C>A
ENST00000568147.5:c.430C>A
ENST00000568169.5:c.478C>A
ENST00000568186.5:c.76C>A
ENST00000568673.5:c.394C>A
ENST00000568740.5:c.391C>A
ENST00000569012.5:c.76C>A
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ENST00000627685.2:c.394C>A
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NM_001369586.1:c.394C>A
NM_003199.3:c.466C>A
NM_001243230.2:c.460C>A

Likely Benign

Met criteria codes 2

BS2 BP4

Not Met criteria codes 1

BS1

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for TCF4 Version 3.0.0*

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The highest population minor allele frequency of the p.Pro156Thr variant in TCF4 in gnomAD v4.1 is 0.000067 in the Admixed American population (not sufficient to meet BS1 criteria). The p.Pro156Thr variant is observed in at least 2 unaffected individuals (Internal database - Ambry) (BS2). Computational analysis prediction tools suggest that the p.Pro156Thr variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4). In summary, the p.Pro156Thr variant in TCF4 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP4).

Met criteria codes

BS2	✔	The p.Pro156Thr variant is observed in at least 2 unaffected individuals (Internal database - Ambry) (BS2).
BP4	✔	Computational analysis prediction tools suggest that the p.Pro156Thr variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4).

Not Met criteria codes

BS1	✖	The highest population minor allele frequency of the p.Pro156Thr variant in TCF4 in gnomAD v4.1 is 0.000067 in the Admixed American population (not sufficient to meet BS1 criteria).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001083962.2(TCF4):c.466C>A (p.P...	Likely Benign	Pitt-Hopkins Syndrome	2024-08-23	1.0	ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for TCF4 Version 3.0.0	TCF4

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