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Variant: NM_001083962.2(TCF4):c.1486+4G>C

Version: 1.0

CA8970200

516726 (ClinVar)

Gene: TCF4 (HGNC:6925)
Condition: Pitt-Hopkins syndrome (MONDO:0012589)
Inheritance Mode: Autosomal dominant inheritance
UUID: 956ee048-4ac3-4468-9b1b-36d9440cdc1c
Approved on: 2024-02-23
Published on: 2024-03-31

HGVS expressions

NM_001083962.2:c.1486+4G>C
NM_001083962.2(TCF4):c.1486+4G>C
NC_000018.10:g.55234544C>G
CM000680.2:g.55234544C>G
NC_000018.9:g.52901775C>G
CM000680.1:g.52901775C>G
NC_000018.8:g.51052773C>G
NG_011716.1:g.359086G>C
NG_011716.2:g.406450G>C
ENST00000354452.8:c.1486+4G>C
ENST00000635822.2:c.1486+4G>C
ENST00000635990.2:n.1166+4G>C
ENST00000636400.2:c.1414+4G>C
ENST00000636751.2:c.*1194+4G>C
ENST00000636822.2:c.1096+4G>C
ENST00000637115.2:c.*1376+4G>C
ENST00000637169.2:c.838+4G>C
ENST00000637239.2:n.1553+4G>C
ENST00000637250.2:n.1180+4G>C
ENST00000637923.2:c.1084+4G>C
ENST00000638154.3:c.1513+4G>C
ENST00000643689.1:c.1096+4G>C
ENST00000674764.1:c.*1097+4G>C
ENST00000675707.1:c.1096+4G>C
ENST00000354452.7:c.1486+4G>C
ENST00000356073.8:c.1486+4G>C
ENST00000398339.5:c.1792+4G>C
ENST00000457482.7:c.1006+4G>C
ENST00000537578.5:c.1414+4G>C
ENST00000537856.7:c.1096+4G>C
ENST00000540999.5:c.1414+4G>C
ENST00000543082.5:c.1360+4G>C
ENST00000544241.6:c.1273+4G>C
ENST00000561831.7:c.1006+4G>C
ENST00000561992.5:c.1096+4G>C
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ENST00000564999.5:c.1486+4G>C
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NM_001369586.1:c.1417+4G>C
NM_003199.3:c.1486+4G>C
NM_001243230.2:c.1477+4G>C

Likely Benign

Met criteria codes 3

BS2 BP5 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 c.1486+4G>C variant in TCF4 is present in 2 XX and 1 XY individual(s) in gnomAD v2 (0.005%) (not sufficient to meet BS1 criteria). Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4). The c.1486+4G>C variant is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2). The c.1486+4G>C variant is

found in a patient with an alternate molecular basis of disease (internal database - Invitae) (BP5). In summary, the c.1486+4G>C variant in TCF4 is classified as likely benign based on the ACMG/AMP criteria (BP4, BS2, BP5).

Met criteria codes

BS2	✔	The c.1486+4G>C variant is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2).
BP5	✔	The c.1486+4G>C variant is found in a patient with an alternate molecular basis of disease (internal database - Invitae) (BP5).
BP4	✔	Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4).

Not Met criteria codes

BS1	✖	The c.1486+4G>C variant in TCF4 is present in 2 XX and 1 XY individual(s) in gnomAD v2 (0.005%) (not sufficient to meet BS1 criteria).

Curation History

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