

Variant: NM_001083962.2(TCF4):c.706C>G (p.Pro236Ala)

Version: 1.0

CA16043067 [↗](#)

373746 (ClinVar) [↗](#)

Gene: TCF4 ([HGNC:6925](#))

Condition: Pitt-Hopkins syndrome ([MONDO:0012589](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: a94a40b0-3f10-4035-8a9f-54bedbf96d01

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HGVS expressions

NM_001083962.2:c.706C>G

NM_001083962.2(TCF4):c.706C>G (p.Pro236Ala)

NC_000018.10:g.55275702G>C

CM000680.2:g.55275702G>C

NC_000018.9:g.52942933G>C

CM000680.1:g.52942933G>C

NC_000018.8:g.51093931G>C

NG_011716.1:g.317928C>G

NG_011716.2:g.365292C>G

ENST00000354452.8:c.706C>G

ENST00000630720.3:c.226C>G

ENST00000635822.2:c.706C>G

ENST00000635990.2:n.386C>G

ENST00000636400.2:c.634C>G

ENST00000636751.2:c.*414C>G

ENST00000636822.2:c.316C>G

ENST00000637115.2:c.*596C>G

ENST00000637169.2:c.58C>G

ENST00000637239.2:n.773C>G

ENST00000637250.2:n.400C>G

ENST00000637923.2:c.304C>G

ENST00000638154.3:c.736C>G

ENST00000643689.1:c.316C>G

ENST00000674764.1:c.*317C>G

ENST00000675707.1:c.316C>G

ENST00000354452.7:c.706C>G

ENST00000356073.8:c.706C>G

ENST00000398339.5:c.1012C>G

ENST00000457482.7:c.226C>G

ENST00000537578.5:c.634C>G

ENST00000537856.7:c.316C>G

ENST00000540999.5:c.634C>G

ENST00000543082.5:c.580C>G

ENST00000544241.6:c.493C>G

ENST00000561831.7:c.226C>G

ENST00000561992.5:c.316C>G

ENST00000562030.3:c.316C>G

ENST00000562607.5:c.316C>G
ENST00000562638.5:c.316C>G
ENST00000562680.5:n.797C>G
ENST00000563686.5:n.561C>G
ENST00000563760.5:n.298C>G
ENST00000564228.5:c.493C>G
ENST00000564403.6:c.724C>G
ENST00000564999.5:c.706C>G
ENST00000565018.6:c.454C>G
ENST00000565580.3:n.455C>G
ENST00000566279.5:c.526C>G
ENST00000566286.5:c.700C>G
ENST00000566514.5:c.667C>G
ENST00000566777.5:c.316C>G
ENST00000567880.5:c.526C>G
ENST00000568169.5:c.718C>G
ENST00000568186.5:c.316C>G
ENST00000568673.5:c.634C>G
ENST00000568740.5:c.631C>G
ENST00000569012.5:c.316C>G
ENST00000570177.6:c.316C>G
ENST00000570287.6:c.226C>G
ENST00000616053.4:c.454C>G
ENST00000625849.2:n.566C>G
ENST00000625925.2:c.316C>G
ENST00000626584.2:c.58C>G
ENST00000627136.2:n.524-14169C>G
ENST00000628078.2:c.316C>G
ENST00000628636.2:c.316C>G
ENST00000628689.2:c.175+3849C>G
ENST00000629343.2:c.316C>G
ENST00000629387.2:c.706C>G
ENST00000630268.2:c.316C>G
ENST00000630319.2:c.475C>G
ENST00000630712.2:c.316C>G
ENST00000630720.2:c.226C>G
ENST00000630828.2:c.496C>G
ENST00000631043.2:n.505C>G
NM_001083962.1:c.706C>G
NM_001243226.2:c.1012C>G
NM_001243227.1:c.634C>G
NM_001243228.1:c.724C>G
NM_001243230.1:c.700C>G
NM_001243231.1:c.580C>G
NM_001243232.1:c.493C>G
NM_001243233.1:c.316C>G
NM_001243234.1:c.226C>G
NM_001243235.1:c.226C>G
NM_001243236.1:c.226C>G
NM_001306207.1:c.634C>G
NM_001306208.1:c.493C>G
NM_003199.2:c.706C>G

NM_001330604.2:c.706C>G
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NM_001348212.1:c.316C>G
NM_001348213.1:c.316C>G
NM_001348214.1:c.226C>G
NM_001348215.1:c.58C>G
NM_001348216.1:c.226C>G
NM_001348217.1:c.634C>G
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NM_001348219.1:c.634C>G
NM_001348220.1:c.631C>G
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NM_001243236.2:c.226C>G
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NM_001348214.2:c.226C>G
NM_001348215.2:c.58C>G
NM_001348216.2:c.226C>G
NM_001348218.2:c.634C>G
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NM_001369567.1:c.706C>G
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NM_001369569.1:c.703C>G
NM_001369570.1:c.703C>G
NM_001369571.1:c.706C>G
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NM_001369580.1:c.634C>G
NM_001369581.1:c.631C>G
NM_001369582.1:c.634C>G
NM_001369583.1:c.634C>G
NM_001369584.1:c.631C>G
NM_001369585.1:c.631C>G
NM_001369586.1:c.634C>G
NM_003199.3:c.706C>G
NM_001243230.2:c.700C>G

Likely Benign

Met criteria codes 2

BS2BP4

Not Met criteria codes 3

BS1PS4PM2

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.Pro236Ala variant in TCF4 in gnomAD v4.1 is 0.00000062 in the European (non-Finnish) population (not sufficient to meet BS1 criteria). The p.Pro236Ala variant is observed in at least 2 unaffected individuals (Internal database - GeneDx, Internal database - Ambry) (BS2). Computational analysis prediction tools suggest that the p.Pro236Ala variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4). In summary, the p.Pro236Ala variant in TCF4 is classified as a likely benign variant based on the ACMG/AMP criteria (BS2, BP4).

Met criteria codes

BS2

i

✓

The p.Pro236Ala variant is observed in at least 2 unaffected individuals (Internal database - GeneDx, Internal database - Ambry) (BS2).

BP4

i

✓

Computational analysis prediction tools suggest that the p.Pro236Ala variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4).

Not Met criteria codes

BS1

i

✗

The highest population minor allele frequency of the p.Pro236Ala variant in TCF4 in gnomAD v4.1 is 0.00000062 in the European (non-Finnish) population (not sufficient to meet BS1 criteria).

PS4

i

✗

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM2

✗

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

▾

▾

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