

Variant: NM_001083962.2(TCF4):c.1113G>A (p.Ser371=)

Version: 1.0

CA8970269 [↗](#)

436960 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 63afad84-522d-4576-9d78-80ec0aac5f08

Approved on: 2026/1/28

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HGVS expressions

NM_001083962.2(TCF4):c.1113G>A (p.Ser371=)

NM_001083962.2:c.1113G>A

NC_000018.10:g.55257348C>T

CM000680.2:g.55257348C>T

NC_000018.9:g.52924579C>T

CM000680.1:g.52924579C>T

NC_000018.8:g.51075577C>T

NG_011716.1:g.336282G>A

NG_011716.2:g.383646G>A

ENST00000354452.8:c.1113G>A

ENST00000630720.3:c.630G>A

ENST00000635822.2:c.1113G>A

ENST00000635990.2:n.793G>A

ENST00000636400.2:c.1041G>A

ENST00000636751.2:c.*821G>A

ENST00000636822.2:c.723G>A

ENST00000637115.2:c.*1003G>A

ENST00000637169.2:c.465G>A

ENST00000637239.2:n.1180G>A

ENST00000637250.2:n.807G>A

ENST00000637923.2:c.711G>A

ENST00000638154.3:c.1140G>A

ENST00000643689.1:c.723G>A

ENST00000674764.1:c.*724G>A

ENST00000675707.1:c.723G>A

ENST00000354452.7:c.1113G>A

ENST00000356073.8:c.1113G>A

ENST00000398339.5:c.1419G>A

ENST00000457482.7:c.633G>A

ENST00000537578.5:c.1041G>A

ENST00000537856.7:c.723G>A

ENST00000540999.5:c.1041G>A

ENST00000543082.5:c.987G>A

ENST00000544241.6:c.900G>A

ENST00000561831.7:c.633G>A

ENST00000561992.5:c.723G>A

ENST00000562680.5:n.1204G>A

ENST00000563760.5:n.626G>A
ENST00000564228.5:c.900G>A
ENST00000564403.6:c.1131G>A
ENST00000564999.5:c.1113G>A
ENST00000565018.6:c.861G>A
ENST00000566279.5:c.933G>A
ENST00000566286.5:c.1104G>A
ENST00000567880.5:c.933G>A
ENST00000568673.5:c.1041G>A
ENST00000568740.5:c.1038G>A
ENST00000570177.6:c.723G>A
ENST00000570287.6:c.633G>A
ENST00000616053.4:c.861G>A
ENST00000626466.1:n.148G>A
ENST00000626584.2:c.465G>A
ENST00000628689.2:c.*256G>A
ENST00000629387.2:c.1113G>A
ENST00000630720.2:c.630G>A
NM_001083962.1:c.1113G>A
NM_001243226.2:c.1419G>A
NM_001243227.1:c.1041G>A
NM_001243228.1:c.1131G>A
NM_001243230.1:c.1104G>A
NM_001243231.1:c.987G>A
NM_001243232.1:c.900G>A
NM_001243233.1:c.723G>A
NM_001243234.1:c.633G>A
NM_001243235.1:c.633G>A
NM_001243236.1:c.633G>A
NM_001306207.1:c.1041G>A
NM_001306208.1:c.900G>A
NM_003199.2:c.1113G>A
NM_001330604.2:c.1110G>A
NM_001330605.2:c.723G>A
NM_001348211.1:c.987G>A
NM_001348212.1:c.723G>A
NM_001348213.1:c.723G>A
NM_001348214.1:c.630G>A
NM_001348215.1:c.465G>A
NM_001348216.1:c.633G>A
NM_001348217.1:c.1041G>A
NM_001348218.1:c.1041G>A
NM_001348219.1:c.1041G>A
NM_001348220.1:c.1038G>A
NM_001243226.3:c.1419G>A
NM_001243227.2:c.1041G>A
NM_001243228.2:c.1131G>A
NM_001243231.2:c.987G>A
NM_001243233.2:c.723G>A
NM_001243234.2:c.633G>A
NM_001243235.2:c.633G>A
NM_001243236.2:c.633G>A

	NM_001330604.3:c.1110G>A NM_001330605.3:c.723G>A NM_001348211.2:c.987G>A NM_001348212.2:c.723G>A NM_001348213.2:c.723G>A NM_001348214.2:c.630G>A NM_001348215.2:c.465G>A NM_001348216.2:c.633G>A NM_001348218.2:c.1041G>A NM_001348219.2:c.1041G>A NM_001369567.1:c.1113G>A NM_001369568.1:c.1113G>A NM_001369569.1:c.1110G>A NM_001369570.1:c.1110G>A NM_001369571.1:c.1113G>A NM_001369572.1:c.1113G>A NM_001369573.1:c.1110G>A NM_001369574.1:c.1110G>A NM_001369575.1:c.1041G>A NM_001369576.1:c.1038G>A NM_001369577.1:c.1038G>A NM_001369578.1:c.1038G>A NM_001369579.1:c.1038G>A NM_001369580.1:c.1038G>A NM_001369581.1:c.1038G>A NM_001369582.1:c.1041G>A NM_001369583.1:c.1041G>A NM_001369584.1:c.1038G>A NM_001369585.1:c.1038G>A NM_001369586.1:c.1044G>A NM_003199.3:c.1113G>A NM_001243230.2:c.1104G>A
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Benign

Met criteria codes 2

BA1BP7

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 5.0.0](#)

[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.Ser371= variant in TCF4 in gnomAD v4.1 is 0.0006424 in the Ashkenazi Jewish population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.000083) for BA1, and therefore meets

this criterion (BA1). The silent p.Ser371= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). The p.Ser371= variant is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time. In summary, the p.Ser371= variant in TCF4 is classified as Benign based on the ACMG/AMP criteria (BA1, BP7). (TCF4 Specifications v.5.0; curation approved on 01/28/2026)

Met criteria codes

BA1			The highest population minor allele frequency of the p.Ser371= variant in TCF4 in gnomAD v4.1 is 0.0006424 in the Ashkenazi Jewish population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.000083) for BA1, and therefore meets this criterion (BA1).
BP7			The silent p.Ser371= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). Splice AI = 0.0500 (Donor loss was 0.05 and all other categories are 0).

Curation History



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