

Variant: *NM\_001083962.1(TCF4):c.687C>A (p.Ser229=)*

Version: 1.0

CA8970418 [↗](#)

327305 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** 78cdc311-810b-46b0-84bf-d4d745a07f25

**Approved on:** 2021/3/26

**Published on:** 2021/5/17

### *HGVS expressions*

**NM\_001083962.1:c.687C>A**

**NM\_001083962.1(TCF4):c.687C>A (p.Ser229=)**

NC\_000018.10:g.55275721G>T

CM000680.2:g.55275721G>T

NC\_000018.9:g.52942952G>T

CM000680.1:g.52942952G>T

NC\_000018.8:g.51093950G>T

NG\_011716.1:g.317909C>A

NG\_011716.2:g.365273C>A

ENST00000354452.8:c.687C>A

ENST00000630720.3:c.207C>A

ENST00000635822.2:c.687C>A

ENST00000635990.2:n.367C>A

ENST00000636400.2:c.615C>A

ENST00000636751.2:c.\*395C>A

ENST00000636822.2:c.297C>A

ENST00000637115.2:c.\*577C>A

ENST00000637169.2:c.39C>A

ENST00000637239.2:n.754C>A

ENST00000637250.2:n.381C>A

ENST00000637923.2:c.285C>A

ENST00000638154.3:c.717C>A

ENST00000643689.1:c.297C>A

ENST00000674764.1:c.\*298C>A

ENST00000675707.1:c.297C>A

ENST00000354452.7:c.687C>A

ENST00000356073.8:c.687C>A

ENST00000398339.5:c.993C>A

ENST00000457482.7:c.207C>A

ENST00000537578.5:c.615C>A

ENST00000537856.7:c.297C>A

ENST00000540999.5:c.615C>A

ENST00000543082.5:c.561C>A

ENST00000544241.6:c.474C>A

ENST00000561831.7:c.207C>A

ENST00000561992.5:c.297C>A

ENST00000562030.3:c.297C>A

ENST00000562607.5:c.297C>A  
ENST00000562638.5:c.297C>A  
ENST00000562680.5:n.778C>A  
ENST00000563686.5:n.542C>A  
ENST00000563760.5:n.279C>A  
ENST00000564228.5:c.474C>A  
ENST00000564403.6:c.705C>A  
ENST00000564999.5:c.687C>A  
ENST00000565018.6:c.435C>A  
ENST00000565580.3:n.436C>A  
ENST00000565908.6:c.615C>A  
ENST00000566279.5:c.507C>A  
ENST00000566286.5:c.681C>A  
ENST00000566514.5:c.648C>A  
ENST00000566777.5:c.297C>A  
ENST00000567880.5:c.507C>A  
ENST00000568169.5:c.699C>A  
ENST00000568186.5:c.297C>A  
ENST00000568673.5:c.615C>A  
ENST00000568740.5:c.612C>A  
ENST00000569012.5:c.297C>A  
ENST00000570177.6:c.297C>A  
ENST00000570287.6:c.207C>A  
ENST00000616053.4:c.435C>A  
ENST00000625849.2:n.547C>A  
ENST00000625925.2:c.297C>A  
ENST00000626584.2:c.39C>A  
ENST00000627136.2:n.524-14188C>A  
ENST00000628078.2:c.297C>A  
ENST00000628636.2:c.297C>A  
ENST00000628689.2:c.175+3830C>A  
ENST00000629343.2:c.297C>A  
ENST00000629387.2:c.687C>A  
ENST00000630268.2:c.297C>A  
ENST00000630319.2:c.456C>A  
ENST00000630712.2:c.297C>A  
ENST00000630720.2:c.207C>A  
ENST00000630828.2:c.477C>A  
ENST00000631043.2:n.486C>A  
NM\_001243226.2:c.993C>A  
NM\_001243227.1:c.615C>A  
NM\_001243228.1:c.705C>A  
NM\_001243230.1:c.681C>A  
NM\_001243231.1:c.561C>A  
NM\_001243232.1:c.474C>A  
NM\_001243233.1:c.297C>A  
NM\_001243234.1:c.207C>A  
NM\_001243235.1:c.207C>A  
NM\_001243236.1:c.207C>A  
NM\_001306207.1:c.615C>A  
NM\_001306208.1:c.474C>A  
NM\_003199.2:c.687C>A

NM\_001330604.2:c.687C>A  
NM\_001330605.2:c.297C>A  
NM\_001348211.1:c.561C>A  
NM\_001348212.1:c.297C>A  
NM\_001348213.1:c.297C>A  
NM\_001348214.1:c.207C>A  
NM\_001348215.1:c.39C>A  
NM\_001348216.1:c.207C>A  
NM\_001348217.1:c.615C>A  
NM\_001348218.1:c.615C>A  
NM\_001348219.1:c.615C>A  
NM\_001348220.1:c.612C>A  
NM\_001083962.2:c.687C>A  
NM\_001243226.3:c.993C>A  
NM\_001243227.2:c.615C>A  
NM\_001243228.2:c.705C>A  
NM\_001243231.2:c.561C>A  
NM\_001243233.2:c.297C>A  
NM\_001243234.2:c.207C>A  
NM\_001243235.2:c.207C>A  
NM\_001243236.2:c.207C>A  
NM\_001330604.3:c.687C>A  
NM\_001330605.3:c.297C>A  
NM\_001348211.2:c.561C>A  
NM\_001348212.2:c.297C>A  
NM\_001348213.2:c.297C>A  
NM\_001348214.2:c.207C>A  
NM\_001348215.2:c.39C>A  
NM\_001348216.2:c.207C>A  
NM\_001348218.2:c.615C>A  
NM\_001348219.2:c.615C>A  
NM\_001369567.1:c.687C>A  
NM\_001369568.1:c.687C>A  
NM\_001369569.1:c.684C>A  
NM\_001369570.1:c.684C>A  
NM\_001369571.1:c.687C>A  
NM\_001369572.1:c.687C>A  
NM\_001369573.1:c.684C>A  
NM\_001369574.1:c.687C>A  
NM\_001369575.1:c.615C>A  
NM\_001369576.1:c.612C>A  
NM\_001369577.1:c.615C>A  
NM\_001369578.1:c.612C>A  
NM\_001369579.1:c.615C>A  
NM\_001369580.1:c.615C>A  
NM\_001369581.1:c.612C>A  
NM\_001369582.1:c.615C>A  
NM\_001369583.1:c.615C>A  
NM\_001369584.1:c.612C>A  
NM\_001369585.1:c.612C>A  
NM\_001369586.1:c.615C>A  
NM\_003199.3:c.687C>A

Likely Benign

Met criteria codes 2

BS1 BP7

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Ser229= variant in TCF4 is 0.027% in East Asian sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The silent p.Ser229= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Ser229= variant in TCF4 is classified as likely benign based on the ACMG/AMP criteria (BS1, BP7).

Met criteria codes

BS1



The allele frequency of the p.Ser229= variant in TCF4 is 0.027% in East Asian sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions

BP7



The silent p.Ser229= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

Curation History



Showing 1 to 1 of 1 rows

|  |
|--|
|  |
|--|

