

Variant: NM_001348220.1:c.1411G>T

Version: 1.0

CA402531414 [↗](#)

690393 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0a189478-9ece-44c0-87b9-f803923296b4

Approved on: 2021/5/11

Published on: 2021/5/17

HGVS expressions

NM_001348220.1:c.1411G>T

NC_000018.10:g.55234548C>A

CM000680.2:g.55234548C>A

NC_000018.9:g.52901779C>A

CM000680.1:g.52901779C>A

NC_000018.8:g.51052777C>A

NG_011716.1:g.359082G>T

NG_011716.2:g.406446G>T

ENST00000354452.8:c.1486G>T

ENST00000635822.2:c.1486G>T

ENST00000635990.2:n.1166G>T

ENST00000636400.2:c.1414G>T

ENST00000636751.2:c.*1194G>T

ENST00000636822.2:c.1096G>T

ENST00000637115.2:c.*1376G>T

ENST00000637169.2:c.838G>T

ENST00000637239.2:n.1553G>T

ENST00000637250.2:n.1180G>T

ENST00000637923.2:c.1084G>T

ENST00000638154.3:c.1513G>T

ENST00000643689.1:c.1096G>T

ENST00000674764.1:c.*1097G>T

ENST00000675707.1:c.1096G>T

ENST00000354452.7:c.1486G>T

ENST00000356073.8:c.1486G>T

ENST00000398339.5:c.1792G>T

ENST00000457482.7:c.1006G>T

ENST00000537578.5:c.1414G>T

ENST00000537856.7:c.1096G>T

ENST00000540999.5:c.1414G>T

ENST00000543082.5:c.1360G>T

ENST00000544241.6:c.1273G>T

ENST00000561831.7:c.1006G>T

ENST00000561992.5:c.1096G>T

ENST00000562680.5:n.1577G>T

ENST00000564228.5:c.1273G>T

ENST00000564403.6:c.1504G>T

ENST00000564999.5:c.1486G>T
ENST00000565018.6:c.1234G>T
ENST00000566279.5:c.1306G>T
ENST00000566286.5:c.1477G>T
ENST00000567880.5:c.1306G>T
ENST00000568673.5:c.1414G>T
ENST00000568740.5:c.1411G>T
ENST00000570177.6:c.1096G>T
ENST00000570287.6:c.1006G>T
ENST00000616053.4:c.1234G>T
ENST00000626466.1:n.521G>T
ENST00000626584.2:c.838G>T
ENST00000629387.2:c.1486G>T
NM_001083962.1:c.1486G>T
NM_001243226.2:c.1792G>T
NM_001243227.1:c.1414G>T
NM_001243228.1:c.1504G>T
NM_001243230.1:c.1477G>T
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NM_001243236.1:c.1006G>T
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NM_001330604.2:c.1483G>T
NM_001330605.2:c.1096G>T
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NM_001348213.1:c.1096G>T
NM_001348214.1:c.1003G>T
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NM_001369584.1:c.1411G>T
NM_001369585.1:c.1411G>T
NM_001369586.1:c.1417G>T
NM_003199.3:c.1486G>T
NM_001243230.2:c.1477G>T

Likely Pathogenic

Met criteria codes 3

PS2

PP3

PM2_Supporting

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 p.Gly496Cys variant in TCF4 occurs in the confirmed de novo state in an individual with a neurodevelopmental disorder consistent with TCF4 abnormality (PS2). The p.Gly496Cys variant in TCF4 is absent from gnomAD (PM2_supporting). This variant occurs at the last amino acid residue in exon 16 and is predicted to affect the canonical splice donor site in intron 16 by several splice prediction programs. In summary, the p.Gly496Cys variant in TCF4 is classified as Likely Pathogenic based on the ACMG/AMP criteria (PS2, PM2_supporting, PP3).

Met criteria codes

PS2



The p.Gly496Cys variant in TCF4 occurs in the confirmed de novo state in an individual with a neurodevelopmental disorder consistent with TCF4 abnormality

PP3			This variant occurs at the last amino acid residue in exon 16 and is predicted to affect the canonical splice donor site in intron 16 by several splice prediction programs.
PM2_Supporting			The p.Gly496Cys variant in TCF4 is absent from gnomAD.

Curation History 

Showing 1 to 1 of 1 rows

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