

Variant: NM_001083962.2(TCF4):c.504A>G (p.Val168=)

Version: 1.0

CA293878 [↗](#)

139404 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a7e99de5-02f7-4772-8727-96988307a984

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HGVS expressions

NM_001083962.2:c.504A>G

NM_001083962.2(TCF4):c.504A>G (p.Val168=)

NC_000018.10:g.55350404T>C

CM000680.2:g.55350404T>C

NC_000018.9:g.53017635T>C

CM000680.1:g.53017635T>C

NC_000018.8:g.51168633T>C

NG_011716.1:g.243226A>G

NG_011716.2:g.290590A>G

ENST00000354452.8:c.504A>G

ENST00000635822.2:c.504A>G

ENST00000635990.2:n.184A>G

ENST00000636400.2:c.432A>G

ENST00000636751.2:c.*212A>G

ENST00000636822.2:c.114A>G

ENST00000637115.2:c.*394A>G

ENST00000637239.2:n.571A>G

ENST00000637250.2:n.198A>G

ENST00000638154.3:c.534A>G

ENST00000643689.1:c.114A>G

ENST00000674764.1:c.*115A>G

ENST00000675707.1:c.114A>G

ENST00000354452.7:c.504A>G

ENST00000356073.8:c.504A>G

ENST00000398339.5:c.810A>G

ENST00000537578.5:c.432A>G

ENST00000537856.7:c.114A>G

ENST00000540999.5:c.432A>G

ENST00000543082.5:c.378A>G

ENST00000544241.6:c.291A>G

ENST00000561992.5:c.114A>G

ENST00000562030.3:c.114A>G

ENST00000562607.5:c.114A>G

ENST00000562638.5:c.114A>G

ENST00000562680.5:n.595A>G

ENST00000563686.5:n.359A>G

ENST00000563824.5:c.432A>G

ENST00000563888.6:c.429A>G
ENST00000564228.5:c.291A>G
ENST00000564403.6:c.504A>G
ENST00000564999.5:c.504A>G
ENST00000565018.6:c.297+53050A>G
ENST00000565580.3:n.298+53050A>G
ENST00000565908.6:c.432A>G
ENST00000566279.5:c.369+53050A>G
ENST00000566286.5:c.498A>G
ENST00000566376.3:n.184A>G
ENST00000566514.5:c.465A>G
ENST00000566777.5:c.114A>G
ENST00000567880.5:c.369+53050A>G
ENST00000568147.5:c.468A>G
ENST00000568169.5:c.516A>G
ENST00000568186.5:c.114A>G
ENST00000568673.5:c.432A>G
ENST00000568740.5:c.429A>G
ENST00000569012.5:c.114A>G
ENST00000570177.6:c.114A>G
ENST00000590810.5:c.150A>G
ENST00000616053.4:c.297+53050A>G
ENST00000625849.2:n.364A>G
ENST00000625925.2:c.114A>G
ENST00000626425.2:c.429A>G
ENST00000626584.2:c.-100+53050A>G
ENST00000626595.2:c.504A>G
ENST00000627136.2:n.478A>G
ENST00000627685.2:c.432A>G
ENST00000628078.2:c.114A>G
ENST00000628636.2:c.114A>G
ENST00000629343.2:c.114A>G
ENST00000629387.2:c.504A>G
ENST00000630268.2:c.114A>G
ENST00000630319.2:c.273A>G
ENST00000630712.2:c.114A>G
ENST00000630828.2:c.294A>G
ENST00000631043.2:n.303A>G
NM_001083962.1:c.504A>G
NM_001243226.2:c.810A>G
NM_001243227.1:c.432A>G
NM_001243228.1:c.504A>G
NM_001243230.1:c.498A>G
NM_001243231.1:c.378A>G
NM_001243232.1:c.291A>G
NM_001243233.1:c.114A>G
NM_001306207.1:c.432A>G
NM_001306208.1:c.291A>G
NM_003199.2:c.504A>G
NM_001330604.2:c.504A>G
NM_001330605.2:c.114A>G
NM_001348211.1:c.378A>G

	NM_001348212.1:c.114A>G NM_001348213.1:c.114A>G NM_001348217.1:c.432A>G NM_001348218.1:c.432A>G NM_001348219.1:c.432A>G NM_001348220.1:c.429A>G NM_001243226.3:c.810A>G NM_001243227.2:c.432A>G NM_001243228.2:c.504A>G NM_001243231.2:c.378A>G NM_001243233.2:c.114A>G NM_001330604.3:c.504A>G NM_001330605.3:c.114A>G NM_001348211.2:c.378A>G NM_001348212.2:c.114A>G NM_001348213.2:c.114A>G NM_001348218.2:c.432A>G NM_001348219.2:c.432A>G NM_001369567.1:c.504A>G NM_001369568.1:c.504A>G NM_001369569.1:c.501A>G NM_001369570.1:c.501A>G NM_001369571.1:c.504A>G NM_001369572.1:c.504A>G NM_001369573.1:c.501A>G NM_001369574.1:c.504A>G NM_001369575.1:c.432A>G NM_001369576.1:c.429A>G NM_001369577.1:c.432A>G NM_001369578.1:c.429A>G NM_001369579.1:c.432A>G NM_001369580.1:c.432A>G NM_001369581.1:c.429A>G NM_001369582.1:c.432A>G NM_001369583.1:c.432A>G NM_001369584.1:c.429A>G NM_001369585.1:c.429A>G NM_001369586.1:c.432A>G NM_003199.3:c.504A>G NM_001243230.2:c.498A>G
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Benign

Met criteria codes 2

BS2_Supporting

BA1

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Val168= variant in TCF4 is 0.057% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). The p.Val168= variant is observed in 1 unaffected individual (Baylor Genetics internal database) (BS2_supporting). In summary, p.Val168= variant in TCF4 is classified as benign based on the ACMG/AMP criteria (BA1, BS2_supporting).

Met criteria codes

BS2_Supporting			The p.Val168= variant is observed in 1 unaffected individual (Baylor Genetics internal database).
BA1			The allele frequency of the p.Val168= variant in TCF4 is 0.057% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1).

Curation History



 

 

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