

Variant: NM_001083962.2(TCF4):c.850C>T (p.His284Tyr)

Version: 1.0

CA357196 [↗](#)

224133 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UID: 0b00488e-5915-4a4f-b331-26c1c3a95595

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HGVS expressions

NM_001083962.2:c.850C>T

NM_001083962.2(TCF4):c.850C>T (p.His284Tyr)

NC_000018.10:g.55269903G>A

CM000680.2:g.55269903G>A

NC_000018.9:g.52937134G>A

CM000680.1:g.52937134G>A

NC_000018.8:g.51088132G>A

NG_011716.1:g.323727C>T

NG_011716.2:g.371091C>T

ENST00000354452.8:c.850C>T

ENST00000630720.3:c.370C>T

ENST00000635822.2:c.850C>T

ENST00000635990.2:n.530C>T

ENST00000636400.2:c.778C>T

ENST00000636751.2:c.*558C>T

ENST00000636822.2:c.460C>T

ENST00000637115.2:c.*740C>T

ENST00000637169.2:c.202C>T

ENST00000637239.2:n.917C>T

ENST00000637250.2:n.544C>T

ENST00000637923.2:c.448C>T

ENST00000638154.3:c.880C>T

ENST00000643689.1:c.460C>T

ENST00000674764.1:c.*461C>T

ENST00000675707.1:c.460C>T

ENST00000354452.7:c.850C>T

ENST00000356073.8:c.850C>T

ENST00000398339.5:c.1156C>T

ENST00000457482.7:c.370C>T

ENST00000537578.5:c.778C>T

ENST00000537856.7:c.460C>T

ENST00000540999.5:c.778C>T

ENST00000543082.5:c.724C>T

ENST00000544241.6:c.637C>T

ENST00000561831.7:c.370C>T

ENST00000561992.5:c.460C>T

ENST00000562030.3:c.460C>T

ENST00000562607.5:c.460C>T
ENST00000562680.5:n.941C>T
ENST00000563686.5:n.705C>T
ENST00000563760.5:n.442C>T
ENST00000564228.5:c.637C>T
ENST00000564403.6:c.868C>T
ENST00000564999.5:c.850C>T
ENST00000565018.6:c.598C>T
ENST00000566279.5:c.670C>T
ENST00000566286.5:c.844C>T
ENST00000566514.5:c.811C>T
ENST00000567880.5:c.670C>T
ENST00000568673.5:c.778C>T
ENST00000568740.5:c.775C>T
ENST00000569012.5:c.460C>T
ENST00000570146.3:c.114C>T
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ENST00000616053.4:c.598C>T
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ENST00000627136.2:n.524-8370C>T
ENST00000628078.2:c.460C>T
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ENST00000629343.2:c.460C>T
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ENST00000630720.2:c.370C>T
ENST00000630828.2:c.640C>T
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NM_001243227.1:c.778C>T
NM_001243228.1:c.868C>T
NM_001243230.1:c.844C>T
NM_001243231.1:c.724C>T
NM_001243232.1:c.637C>T
NM_001243233.1:c.460C>T
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NM_001243235.1:c.370C>T
NM_001243236.1:c.370C>T
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NM_003199.2:c.850C>T
NM_001330604.2:c.850C>T
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NM_001369586.1:c.778C>T
NM_003199.3:c.850C>T
NM_001243230.2:c.844C>T

Uncertain Significance

Not Met criteria codes 20

BA1 PP1 PP3 PP4 PM6
PM2 PM1 PM3 PM5 BS2
BS1 BS4 BS3 PS1 PS2 PS3
PS4 BP4 BP2 BP5

Expert Panel

[Rett and Angelman-like Disorders VCEP](#)



















Criteria Specification Information





















Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.His284Tyr variant in TCF4 has been reported in an individual with a neurodevelopmental phenotype (PMID 28554332). The p.His284Tyr variant is present in two XY individuals in gnomAD v4 (0.0003%) (not sufficient to meet BS1 criteria). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.His284Tyr variant in TCF4 is classified as a Variant of Unknown Significance based on the ACMG/AMP criteria (no criteria met).

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			Computational prediction analysis tools are inconclusive for this variant
PP4			The p.His284Tyr variant in TCF4 has been reported in an individual with a neurodevelopmental phenotype (PMID 28554332).
PM6			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
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS1			The p.His284Tyr variant in TCF4 is present in 2 XY individuals in gnomAD v4 (0.0003%) (not sufficient to meet BS1 criteria)
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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