Variant: NM_001083962.2(TCF4):c.269A>G (p.Asn90Ser)

Version: 1.0

CA234991

167731 (ClinVar)

Gene: TCF4 (HGNC:6925)
Condition: Pitt-Hopkins syndrome (MONDO:0012589)
Inheritance Mode: Autosomal dominant inheritance
UUID: 8ab7cf89-4df9-4ee4-ac6a-8b22d634ebb3
Approved on: 2021-03-26
Published on: 2021-05-17

HGVS expressions

NM_001083962.2:c.269A>G
NM_001083962.2(TCF4):c.269A>G (p.Asn90Ser)
NC_000018.10:g.55461054T>C
CM000680.2:g.55461054T>C
NC_000018.9:g.53128285T>C
CM000680.1:g.53128285T>C
NC_000018.8:g.51279283T>C
NG_011716.1:g.132576A>G
NG_011716.2:g.179940A>G
ENST00000354452.8:c.269A>G
ENST00000635822.2:c.269A>G
ENST00000636400.2:c.197A>G
ENST00000636751.2:c.197A>G
ENST00000637115.2:c.*159A>G
ENST00000637239.2:n.336A>G
ENST00000638154.3:c.299A>G
ENST00000674598.1:n.739A>G
ENST00000674764.1:c.143A>G
ENST00000354452.7:c.269A>G
ENST00000356073.8:c.269A>G
ENST00000398339.5:c.575A>G
ENST00000537578.5:c.197A>G
ENST00000540999.5:c.197A>G
ENST00000543082.5:c.143A>G
ENST00000562543.5:c.269A>G
ENST00000562847.5:c.32A>G
ENST00000563686.5:n.124A>G
ENST00000563824.5:c.197A>G
ENST00000563888.6:c.197A>G
ENST00000564343.5:c.197A>G
ENST00000564403.6:c.269A>G
ENST00000564999.5:c.269A>G
ENST00000565018.6:c.197A>G
ENST00000565580.3:n.198A>G
ENST00000565908.6:c.197A>G
ENST00000566279.5:c.269A>G
ENST00000566286.5:c.263A>G

ENST00000566514.5:c.230A>G
ENST00000567880.5:c.269A>G
ENST00000568147.5:c.233A>G
ENST00000568169.5:c.281A>G
ENST00000568673.5:c.197A>G
ENST00000568740.5:c.197A>G
ENST00000569357.4:c.478A>G
ENST00000616053.4:c.197A>G
ENST00000625716.2:n.199A>G
ENST00000626425.2:c.197A>G
ENST00000626595.2:c.269A>G
ENST00000627136.2:n.243A>G
ENST00000627320.2:c.*199A>G
ENST00000627685.2:c.197A>G
ENST00000627784.2:c.269A>G
ENST00000629387.2:c.269A>G
ENST00000630319.2:c.74-57536A>G
NM_001083962.1:c.269A>G
NM_001243226.2:c.575A>G
NM_001243227.1:c.197A>G
NM_001243228.1:c.269A>G
NM_001243230.1:c.263A>G
NM_001243231.1:c.143A>G
NM_001306207.1:c.197A>G
NM_003199.2:c.269A>G
NR_132985.1:n.178+8344T>C
NM_001330604.2:c.269A>G
NM_001348211.1:c.143A>G
NM_001348217.1:c.197A>G
NM_001348218.1:c.197A>G
NM_001348219.1:c.197A>G
NM_001348220.1:c.197A>G
NM_001243226.3:c.575A>G
NM_001243227.2:c.197A>G
NM_001243228.2:c.269A>G
NM_001243231.2:c.143A>G
NM_001330604.3:c.269A>G
NM_001348211.2:c.143A>G
NM_001348218.2:c.197A>G
NM_001348219.2:c.197A>G
NM_001369567.1:c.269A>G
NM_001369568.1:c.269A>G
NM_001369569.1:c.269A>G
NM_001369570.1:c.269A>G
NM_001369571.1:c.269A>G
NM_001369572.1:c.269A>G
NM_001369573.1:c.269A>G
NM_001369574.1:c.269A>G
NM_001369575.1:c.197A>G
NM_001369576.1:c.197A>G
NM_001369577.1:c.197A>G
NM_001369578.1:c.197A>G

NM_001369579.1:c.197A>G
NM_001369580.1:c.197A>G
NM_001369581.1:c.197A>G
NM_001369582.1:c.197A>G
NM_001369583.1:c.197A>G
NM_001369584.1:c.197A>G
NM_001369585.1:c.197A>G
NM_001369586.1:c.197A>G
NM_003199.3:c.269A>G
NM_001243230.2:c.263A>G

Benign

Met criteria codes 3

BA1 BS2 BP4

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.Asn90Ser variant in TCF4 is 1.5% in Ashkenazi Jewish 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.Asn90Ser variant is observed in at least 2 unaffected individuals (internal database) (BS2). Computational analysis prediction tools suggest that the p.Asn90Ser variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4). In summary, the p.Asn90Ser variant in TCF4 is classified as benign based on the ACMG/AMP criteria (BA1, BS2, BP4).

Met criteria codes

BA1	✔	The allele frequency of the p.Asn90Ser variant in TCF4 is 1.5% in Ashkenazi Jewish 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.
BS2	✔	The p.Asn90Ser variant is observed in at least 2 unaffected individuals (internal database)
BP4	✔	Computational analysis prediction tools suggest that the p.Asn90Ser variant does not have a deleterious impact; however this information does not predict clinical significance on its own

Curation History

Showing 1 to 1 of 1 rows