

Variant: *NM_130839.5(UBE3A):c.1338T>C (p.Phe446=)*

Version: 1.0

CA243572 [↗](#)

196572 (ClinVar) [↗](#)

Gene: UBE3A ([HGNC:7337](#))

Condition: Angelman syndrome ([MONDO:0007113](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 74809671-f253-470e-9e9f-c4ecdbf0029f

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HGVS expressions

NM_130839.5(UBE3A):c.1338T>C (p.Phe446=)

NM_130839.5:c.1338T>C

NC_000015.10:g.25370836A>G

CM000677.2:g.25370836A>G

NC_000015.9:g.25615983A>G

CM000677.1:g.25615983A>G

NC_000015.8:g.23167076A>G

NG_009268.1:g.73146T>C

ENST00000438097.6:c.1278T>C

ENST00000625778.3:c.1278T>C

ENST00000635914.1:c.1278T>C

ENST00000637886.1:c.1338T>C

ENST00000638011.1:c.1278T>C

ENST00000638155.1:c.1278T>C

ENST00000648336.2:c.1338T>C

ENST00000649550.1:c.1278T>C

ENST00000650110.1:c.1347T>C

ENST00000675000.1:n.2013T>C

ENST00000675177.1:c.1161T>C

ENST00000675593.1:n.4034T>C

ENST00000232165.7:c.1278T>C

ENST00000397954.6:c.1347T>C

ENST00000428984.6:c.1278T>C

ENST00000438097.5:c.1278T>C

ENST00000566215.5:c.1278T>C

ENST00000614096.4:c.1338T>C

ENST00000625778.2:c.1278T>C

ENST00000630424.2:c.1278T>C

NM_000462.3:c.1347T>C

NM_130838.1:c.1278T>C

NM_130839.2:c.1338T>C

NM_000462.5:c.1347T>C

NM_001354505.1:c.1338T>C

NM_001354506.1:c.1278T>C

NM_001354507.1:c.1278T>C

NM_001354508.1:c.1278T>C

NM_001354509.1:c.1278T>C

NM_001354511.1:c.1278T>C
NM_001354512.1:c.1278T>C
NM_001354513.1:c.1278T>C
NM_001354523.1:c.1278T>C
NM_001354526.1:c.1278T>C
NM_001354538.1:c.1338T>C
NM_001354539.1:c.1278T>C
NM_001354540.1:c.1278T>C
NM_001354541.1:c.1278T>C
NM_001354542.1:c.1278T>C
NM_001354543.1:c.1278T>C
NM_001354544.1:c.1278T>C
NM_001354545.1:c.1338T>C
NM_001354546.1:c.1161T>C
NM_001354547.1:c.1278T>C
NM_001354548.1:c.1278T>C
NM_001354549.1:c.1278T>C
NM_001354550.1:c.361+4629T>C
NM_001354551.1:c.301+4629T>C
NM_130838.3:c.1278T>C
NM_130839.4:c.1338T>C
NR_146177.1:n.18393-20760A>G
NR_148916.1:n.1886T>C
NM_001354506.2:c.1278T>C
NM_001354507.2:c.1278T>C
NM_001354508.2:c.1278T>C
NM_001354509.2:c.1278T>C
NM_001354511.2:c.1278T>C
NM_001354512.2:c.1278T>C
NM_001354513.2:c.1278T>C
NM_001354523.2:c.1278T>C
NM_001354538.2:c.1338T>C
NM_001354539.2:c.1278T>C
NM_001354540.2:c.1278T>C
NM_001354541.2:c.1278T>C
NM_001354542.2:c.1278T>C
NM_001354543.2:c.1278T>C
NM_001354544.2:c.1278T>C
NM_001354545.2:c.1338T>C
NM_001354546.2:c.1161T>C
NM_001354547.2:c.1278T>C
NM_001354548.2:c.1278T>C
NM_001354549.2:c.1278T>C
NM_001354550.2:c.361+4629T>C
NM_001354551.2:c.301+4629T>C
NM_001374461.1:c.1278T>C
NM_130838.4:c.1278T>C
NR_148916.2:n.1854T>C

Benign

BS1

BP7

Not Met criteria codes 1

BS2

Evidence Links 0

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.Phe426= variant in UBE3A (NM_130838.2) is 0.014% in European (non-Finnish) sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Phe426= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Phe426= variant in UBE3A (NM_130838.2) is classified as Likely Benign based on the ACMG/AMP criteria (BS1, BP7).

Met criteria codes

BS1

i

✓

The allele frequency of the p.Phe426= variant in UBE3A (NM_130838.2) is 0.014% in European (non-Finnish) sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).

BP7

i

✓

The p.Phe426= variant in UBE3A (NM_130838.2) is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide.

Not Met criteria codes

BS2

i

✗

The p.Phe426= variant in UBE3A (NM_130838.2) is observed in at least 4 unaffected individuals (gnomAD) (BS2).

Curation History

Showing 1 to 1 of 1 rows

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