

Variant: *NM_130839.5(UBE3A):c.1272C>T (p.Pro424=)*

Version: 1.0

CA16606643 [↗](#)

378843 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 368dce9e-5faf-4081-a47b-86f65f9c96b4

Approved on: 2023/6/16

Published on: 2023/6/21

HGVS expressions

NM_130839.5(UBE3A):c.1272C>T (p.Pro424=)

NM_130839.5:c.1272C>T

NC_000015.10:g.25370902G>A

CM000677.2:g.25370902G>A

NC_000015.9:g.25616049G>A

CM000677.1:g.25616049G>A

NC_000015.8:g.23167142G>A

NG_009268.1:g.73080C>T

ENST00000438097.6:c.1212C>T

ENST00000625778.3:c.1212C>T

ENST00000635914.1:c.1212C>T

ENST00000637886.1:c.1272C>T

ENST00000638011.1:c.1212C>T

ENST00000638155.1:c.1212C>T

ENST00000648336.2:c.1272C>T

ENST00000649550.1:c.1212C>T

ENST00000650110.1:c.1281C>T

ENST00000675000.1:n.1947C>T

ENST00000675177.1:c.1095C>T

ENST00000675593.1:n.3968C>T

ENST00000232165.7:c.1212C>T

ENST00000397954.6:c.1281C>T

ENST00000428984.6:c.1212C>T

ENST00000438097.5:c.1212C>T

ENST00000566215.5:c.1212C>T

ENST00000614096.4:c.1272C>T

ENST00000625778.2:c.1212C>T

ENST00000630424.2:c.1212C>T

NM_000462.3:c.1281C>T

NM_130838.1:c.1212C>T

NM_130839.2:c.1272C>T

NM_000462.5:c.1281C>T

NM_001354505.1:c.1272C>T

NM_001354506.1:c.1212C>T

NM_001354507.1:c.1212C>T

NM_001354508.1:c.1212C>T

NM_001354509.1:c.1212C>T

NM_001354511.1:c.1212C>T
NM_001354512.1:c.1212C>T
NM_001354513.1:c.1212C>T
NM_001354523.1:c.1212C>T
NM_001354526.1:c.1212C>T
NM_001354538.1:c.1272C>T
NM_001354539.1:c.1212C>T
NM_001354540.1:c.1212C>T
NM_001354541.1:c.1212C>T
NM_001354542.1:c.1212C>T
NM_001354543.1:c.1212C>T
NM_001354544.1:c.1212C>T
NM_001354545.1:c.1272C>T
NM_001354546.1:c.1095C>T
NM_001354547.1:c.1212C>T
NM_001354548.1:c.1212C>T
NM_001354549.1:c.1212C>T
NM_001354550.1:c.361+4563C>T
NM_001354551.1:c.301+4563C>T
NM_130838.3:c.1212C>T
NM_130839.4:c.1272C>T
NR_146177.1:n.18393-20694G>A
NR_148916.1:n.1820C>T
NM_001354506.2:c.1212C>T
NM_001354507.2:c.1212C>T
NM_001354508.2:c.1212C>T
NM_001354509.2:c.1212C>T
NM_001354511.2:c.1212C>T
NM_001354512.2:c.1212C>T
NM_001354513.2:c.1212C>T
NM_001354523.2:c.1212C>T
NM_001354538.2:c.1272C>T
NM_001354539.2:c.1212C>T
NM_001354540.2:c.1212C>T
NM_001354541.2:c.1212C>T
NM_001354542.2:c.1212C>T
NM_001354543.2:c.1212C>T
NM_001354544.2:c.1212C>T
NM_001354545.2:c.1272C>T
NM_001354546.2:c.1095C>T
NM_001354547.2:c.1212C>T
NM_001354548.2:c.1212C>T
NM_001354549.2:c.1212C>T
NM_001354550.2:c.361+4563C>T
NM_001354551.2:c.301+4563C>T
NM_001374461.1:c.1212C>T
NM_130838.4:c.1212C>T
NR_148916.2:n.1788C>T

Likely Benign

BP7BP4

Not Met criteria codes 1

BS1

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 c.1212C>T p.Pro404= variant in UBE3A (NM_130838.2) is present in gnomAD v2.1.1 at a frequency of 0.0062% in the European (non-Finnish) sub population (no criteria met). The silent p.Pro404= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP4, BP7). In summary, the c.1212C>T p.Pro404= variant in UBE3A is classified as Likely Benign based on the ACMG/AMP criteria (BP4, BP7).

Met criteria codes

BP7

The silent p.(Pro404=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide.

BP4

The silent p.(Pro404=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide.

Not Met criteria codes

BS1

The c.1212C>T p.(Pro404=) variant in UBE3A (NM_130838.2) is present in gnomAD v2.1.1 at a frequency of 0.0062% in the European (non-Finnish) sub population (no criteria met).

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

[ClinGen Terms of Use.](#)

[✕ Powered by BCM's Genboree.](#)