

Variant: NM_130838.1(UBE3A):c.1147G>T (p.Glu383Ter)

Version: 1.0

CA391354119 [↗](#)

546267 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance (with paternal imprinting (HP:0012274))

UUID: 5a71dd7c-8402-4233-9b65-cc71afc0f62a

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HGVS expressions

NM_130838.1(UBE3A):c.1147G>T (p.Glu383Ter)

NM_130838.1:c.1147G>T

NC_000015.10:g.25370967C>A

CM000677.2:g.25370967C>A

NC_000015.9:g.25616114C>A

CM000677.1:g.25616114C>A

NC_000015.8:g.23167207C>A

NG_009268.1:g.73015G>T

ENST00000438097.6:c.1147G>T

ENST00000625778.3:c.1147G>T

ENST00000635914.1:c.1147G>T

ENST00000637886.1:c.1207G>T

ENST00000638011.1:c.1147G>T

ENST00000638155.1:c.1147G>T

ENST00000648336.2:c.1207G>T

ENST00000649550.1:c.1147G>T

ENST00000650110.1:c.1216G>T

ENST00000675000.1:n.1882G>T

ENST00000675177.1:c.1030G>T

ENST00000675593.1:n.3903G>T

ENST00000232165.7:c.1147G>T

ENST00000397954.6:c.1216G>T

ENST00000428984.6:c.1147G>T

ENST00000438097.5:c.1147G>T

ENST00000566215.5:c.1147G>T

ENST00000614096.4:c.1207G>T

ENST00000625778.2:c.1147G>T

ENST00000630424.2:c.1147G>T

NM_000462.3:c.1216G>T

NM_130839.2:c.1207G>T

NM_000462.5:c.1216G>T

NM_001354505.1:c.1207G>T

NM_001354506.1:c.1147G>T

NM_001354507.1:c.1147G>T

NM_001354508.1:c.1147G>T

NM_001354509.1:c.1147G>T

NM_001354511.1:c.1147G>T

NM_001354512.1:c.1147G>T
NM_001354513.1:c.1147G>T
NM_001354523.1:c.1147G>T
NM_001354526.1:c.1147G>T
NM_001354538.1:c.1207G>T
NM_001354539.1:c.1147G>T
NM_001354540.1:c.1147G>T
NM_001354541.1:c.1147G>T
NM_001354542.1:c.1147G>T
NM_001354543.1:c.1147G>T
NM_001354544.1:c.1147G>T
NM_001354545.1:c.1207G>T
NM_001354546.1:c.1030G>T
NM_001354547.1:c.1147G>T
NM_001354548.1:c.1147G>T
NM_001354549.1:c.1147G>T
NM_001354550.1:c.361+4498G>T
NM_001354551.1:c.301+4498G>T
NM_130838.3:c.1147G>T
NM_130839.4:c.1207G>T
NR_146177.1:n.18393-20629C>A
NR_148916.1:n.1755G>T
NM_001354506.2:c.1147G>T
NM_001354507.2:c.1147G>T
NM_001354508.2:c.1147G>T
NM_001354509.2:c.1147G>T
NM_001354511.2:c.1147G>T
NM_001354512.2:c.1147G>T
NM_001354513.2:c.1147G>T
NM_001354523.2:c.1147G>T
NM_001354538.2:c.1207G>T
NM_001354539.2:c.1147G>T
NM_001354540.2:c.1147G>T
NM_001354541.2:c.1147G>T
NM_001354542.2:c.1147G>T
NM_001354543.2:c.1147G>T
NM_001354544.2:c.1147G>T
NM_001354545.2:c.1207G>T
NM_001354546.2:c.1030G>T
NM_001354547.2:c.1147G>T
NM_001354548.2:c.1147G>T
NM_001354549.2:c.1147G>T
NM_001354550.2:c.361+4498G>T
NM_001354551.2:c.301+4498G>T
NM_001374461.1:c.1147G>T
NM_130838.4:c.1147G>T
NM_130839.5:c.1207G>T
NR_148916.2:n.1723G>T

Pathogenic

PP4

PVS1

PM2_Supporting

Not Met criteria codes

1

PS4

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 p.Glu383Ter variant in UBE3A is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1). The p.Glu383Ter variant in UBE3A has been reported in a patient with a clinical phenotype suggestive of Angelman syndrome (PP4). This variant is absent from gnomAD (PM2_supporting). In summary, the p.Glu383Ter variant in UBE3A is classified as Pathogenic for Angelman syndrome based on the ACMG/AMP criteria (PVS1, PM2_supporting, PP4).

Met criteria codes

PP4

✓

The patient is reported to have a clinical phenotype suggestive of Angelman syndrome

PVS1

✓

The p.Glu383Ter variant in UBE3A is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism.

PM2_Supporting

✓

The p.Glu383Ter variant in UBE3A is absent from gnomAD.

Not Met criteria codes

PS4

✗

This variant has not been reported in the published literature.

Curation History

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