

Variant: NM_130839.5(UBE3A):c.2563C>T (p.Leu855Phe)

Version: 2.0

CA294630 [↗](#)

156620 (ClinVar) [↗](#)

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

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

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

UUID: 0b0cd308-4e86-4727-9fb9-c9120a58828c

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HGVS expressions

NM_130839.5:c.2563C>T

NM_130839.5(UBE3A):c.2563C>T (p.Leu855Phe)

NC_000015.10:g.25339193G>A

CM000677.2:g.25339193G>A

NC_000015.9:g.25584340G>A

CM000677.1:g.25584340G>A

NC_000015.8:g.23135433G>A

NG_009268.1:g.104789C>T

ENST00000438097.6:c.2503C>T

ENST00000635914.1:c.*888C>T

ENST00000636667.1:c.100C>T

ENST00000637886.1:c.2563C>T

ENST00000638011.1:c.2503C>T

ENST00000638155.1:c.2503C>T

ENST00000648336.2:c.2563C>T

ENST00000649550.1:c.2503C>T

ENST00000650110.1:c.2572C>T

ENST00000675177.1:c.2386C>T

ENST00000232165.7:c.2503C>T

ENST00000397954.6:c.2572C>T

ENST00000428984.6:c.2503C>T

ENST00000438097.5:c.2503C>T

ENST00000566215.5:c.2503C>T

ENST00000604860.3:c.514C>T

ENST00000614096.4:c.2563C>T

ENST00000626176.2:c.374C>T

ENST00000630424.2:c.2503C>T

NM_000462.3:c.2572C>T

NM_130838.1:c.2503C>T

NM_130839.2:c.2563C>T

NM_000462.5:c.2572C>T

NM_001354505.1:c.2563C>T

NM_001354506.1:c.2503C>T

NM_001354507.1:c.2503C>T

NM_001354508.1:c.2503C>T

NM_001354509.1:c.2503C>T

NM_001354511.1:c.2503C>T

NM_001354512.1:c.2503C>T
NM_001354513.1:c.2503C>T
NM_001354523.1:c.2503C>T
NM_001354526.1:c.2503C>T
NM_001354538.1:c.2563C>T
NM_001354539.1:c.2503C>T
NM_001354540.1:c.2503C>T
NM_001354541.1:c.2503C>T
NM_001354542.1:c.2503C>T
NM_001354543.1:c.2503C>T
NM_001354544.1:c.2503C>T
NM_001354545.1:c.2407C>T
NM_001354546.1:c.2386C>T
NM_001354547.1:c.2347C>T
NM_001354548.1:c.2347C>T
NM_001354549.1:c.2338C>T
NM_001354550.1:c.1312C>T
NM_001354551.1:c.1252C>T
NM_130838.3:c.2503C>T
NM_130839.4:c.2563C>T
NR_146177.1:n.18393-52403G>A
NR_148916.1:n.3107C>T
NM_001354506.2:c.2503C>T
NM_001354507.2:c.2503C>T
NM_001354508.2:c.2503C>T
NM_001354509.2:c.2503C>T
NM_001354511.2:c.2503C>T
NM_001354512.2:c.2503C>T
NM_001354513.2:c.2503C>T
NM_001354523.2:c.2503C>T
NM_001354538.2:c.2563C>T
NM_001354539.2:c.2503C>T
NM_001354540.2:c.2503C>T
NM_001354541.2:c.2503C>T
NM_001354542.2:c.2503C>T
NM_001354543.2:c.2503C>T
NM_001354544.2:c.2503C>T
NM_001354545.2:c.2407C>T
NM_001354546.2:c.2386C>T
NM_001354547.2:c.2347C>T
NM_001354548.2:c.2347C>T
NM_001354549.2:c.2338C>T
NM_001354550.2:c.1312C>T
NM_001354551.2:c.1252C>T
NM_001374461.1:c.2503C>T
NM_130838.4:c.2503C>T
NR_148916.2:n.3075C>T

Likely Pathogenic

Met criteria codes 5

Expert Panel

Rett and Angelman-like Disorders VCEP

- [Criteria Specification:](#) ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for UBE3A Version 7.0
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Leu835Phe variant in UBE3A (NM_130838.4) has been reported in an individual with a clinical phenotype suggestive of Angelman syndrome (internal database - University of Chicago Genetic Services Laboratories) (PP4). The variant has been reported to segregate in three informative meioses (internal database - University of Chicago Genetic Services Laboratories) (PP1_Moderate). The p.Leu835Phe variant has been observed in at least 1 other affected individual (internal database - GeneDx) (PS4_Supporting). The p.Leu835Phe variant in UBE3A is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools suggest a deleterious impact; however, this information does not predict clinical significance on its own (PP3). In summary, the p.Leu835Phe variant in UBE3A is classified as Likely Pathogenic for Angelman syndrome based on the ACMG/AMP criteria (PP1_Moderate, PS4_Supporting, PM2_Supporting, PP3, PP4). (UBE3A Specifications v7.0; curation approved on 8/24/2026)

Met criteria codes

PP1_Moderate			The variant has been reported to segregate in three informative meioses (internal database - University of Chicago Genetic Services Laboratories) (PP1_Moderate).
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PP4			The p.Leu835Phe variant in UBE3A has been reported in an individual with a clinical phenotype suggestive of Angelman syndrome (internal database - University of Chicago Genetic Services Laboratories) (PP4).
PM2_Supporting			The p.Leu835Phe variant in UBE3A is absent from gnomAD v4.1 (PM2_Supporting).
PS4_Supporting			The p.Leu835Phe variant has been observed in at least 1 other affected individual (internal database - GeneDx) (PS4_Supporting).

Curation History

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