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Variant: NM_130839.5(UBE3A):c.2444G>A (p.Gly815Glu)

Version: 1.0

CA391351072

654655 (ClinVar)

Gene: UBE3A (HGNC:7337)
Condition: Angelman syndrome (MONDO:0007113)
Inheritance Mode: Autosomal dominant inheritance
UUID: 52c90ff0-38d6-4a6f-8964-0ea960fb1997
Approved on: 2025/2/28
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HGVS expressions

NM_130839.5:c.2444G>A
NM_130839.5(UBE3A):c.2444G>A (p.Gly815Glu)
NC_000015.10:g.25340139C>T
CM000677.2:g.25340139C>T
NC_000015.9:g.25585286C>T
CM000677.1:g.25585286C>T
NC_000015.8:g.23136379C>T
NG_009268.1:g.103843G>A
ENST00000438097.6:c.2384G>A
ENST00000635914.1:c.2384G>A
ENST00000636667.1:c.-20G>A
ENST00000637886.1:c.2444G>A
ENST00000638011.1:c.2384G>A
ENST00000638155.1:c.2384G>A
ENST00000648336.2:c.2444G>A
ENST00000649550.1:c.2384G>A
ENST00000650110.1:c.2453G>A
ENST00000675177.1:c.2267G>A
ENST00000675593.1:n.5140G>A
ENST00000232165.7:c.2384G>A
ENST00000397954.6:c.2453G>A
ENST00000428984.6:c.2384G>A
ENST00000438097.5:c.2384G>A
ENST00000566215.5:c.2384G>A
ENST00000604860.3:c.395G>A
ENST00000614096.4:c.2444G>A
ENST00000626176.2:c.255G>A
ENST00000630424.2:c.2384G>A
NM_000462.3:c.2453G>A
NM_130838.1:c.2384G>A
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NM_000462.5:c.2453G>A
NM_001354505.1:c.2444G>A
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NM_001354507.1:c.2384G>A
NM_001354508.1:c.2384G>A
NM_001354509.1:c.2384G>A

NM_001354511.1:c.2384G>A
NM_001354512.1:c.2384G>A
NM_001354513.1:c.2384G>A
NM_001354523.1:c.2384G>A
NM_001354526.1:c.2384G>A
NM_001354538.1:c.2444G>A
NM_001354539.1:c.2384G>A
NM_001354540.1:c.2384G>A
NM_001354541.1:c.2384G>A
NM_001354542.1:c.2384G>A
NM_001354543.1:c.2384G>A
NM_001354544.1:c.2384G>A
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NM_001354546.1:c.2267G>A
NM_001354547.1:c.2228G>A
NM_001354548.1:c.2228G>A
NM_001354549.1:c.2219G>A
NM_001354550.1:c.1193G>A
NM_001354551.1:c.1133G>A
NM_130838.3:c.2384G>A
NM_130839.4:c.2444G>A
NR_146177.1:n.18393-51457C>T
NR_148916.1:n.2988G>A
NM_001354506.2:c.2384G>A
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NM_001354508.2:c.2384G>A
NM_001354509.2:c.2384G>A
NM_001354511.2:c.2384G>A
NM_001354512.2:c.2384G>A
NM_001354513.2:c.2384G>A
NM_001354523.2:c.2384G>A
NM_001354538.2:c.2444G>A
NM_001354539.2:c.2384G>A
NM_001354540.2:c.2384G>A
NM_001354541.2:c.2384G>A
NM_001354542.2:c.2384G>A
NM_001354543.2:c.2384G>A
NM_001354544.2:c.2384G>A
NM_001354545.2:c.2288G>A
NM_001354546.2:c.2267G>A
NM_001354547.2:c.2228G>A
NM_001354548.2:c.2228G>A
NM_001354549.2:c.2219G>A
NM_001354550.2:c.1193G>A
NM_001354551.2:c.1133G>A
NM_001374461.1:c.2384G>A
NM_130838.4:c.2384G>A
NR_148916.2:n.2956G>A

Met criteria codes 3

Expert Panel

Likely Pathogenic

PS2 PM2_Supporting PP3

Not Met criteria codes 2

BP4 PS4

Evidence Links 0

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for UBE3A Version 5.0.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The c.2444G>A (p.Gly815Glu) variant in UBE3A has been identified as a de novo occurrence with confirmed parental relationships in 1 individual with clinical features of Angelman syndrome (PS2; GeneDx: internal data). This variant is absent from gnomAD v4.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.705, which is above the threshold of 0.644, evidence that correlates with impact to UBE3A function (PP3). In summary, the p.Gly815Glu variant in UBE3A is classified as Likely Pathogenic for Angelman syndrome based on the ACMG/AMP criteria (PS2, PP3, PM2_supporting) (UBE3A specifications version 5.0; 02/28/2025).

Met criteria codes

PS2	✔	This variant has been identified as a de novo occurrence with confirmed parental relationships in 1 individual with clinical features of Angelman syndrome (PS2; GeneDx: internal data).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1 (PM2_Supporting).
PP3	✔	The computational predictor REVEL gives a score of 0.705, which is above the threshold of 0.644, evidence that correlates with impact to UBE3A function (PP3).

Not Met criteria codes

BP4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	✖	The p.Gly815Glu variant is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time.

Curation History

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