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Variant: NM_130838.1(UBE3A):c.1745_1747delCTT (p.Ser582del)

Version: 1.0

CA333369

155987 (ClinVar)

Gene: UBE3A (HGNC:7337)
Condition: Angelman syndrome (MONDO:0007113)
Inheritance Mode: Autosomal dominant inheritance (with paternal imprinting (HP:0012274))
UUID: 63c4c18c-b6ad-4d46-90d9-5ab500c96c58
Approved on: 2021/3/26
Published on: 2021/5/17

HGVS expressions

NM_130838.1:c.1745_1747del
NM_130838.1(UBE3A):c.1745_1747delCTT (p.Ser582del)
NM_130838.1:c.1745_1747delCTT
NC_000015.10:g.25356847_25356849del
CM000677.2:g.25356847_25356849del
NC_000015.9:g.25601994_25601996del
CM000677.1:g.25601994_25601996del
NC_000015.8:g.23153087_23153089del
NG_009268.1:g.87137_87139del
ENST00000438097.6:c.1745_1747del
ENST00000625778.3:c.1745_1747del
ENST00000635914.1:c.1745_1747del
ENST00000637886.1:c.1805_1807del
ENST00000638011.1:c.1745_1747del
ENST00000638155.1:c.1745_1747del
ENST00000648336.2:c.1805_1807del
ENST00000649550.1:c.1745_1747del
ENST00000650110.1:c.1814_1816del
ENST00000675177.1:c.1628_1630del
ENST00000675593.1:n.4501_4503del
ENST00000232165.7:c.1745_1747del
ENST00000397954.6:c.1814_1816del
ENST00000428984.6:c.1745_1747del
ENST00000438097.5:c.1745_1747del
ENST00000566215.5:c.1745_1747del
ENST00000614096.4:c.1805_1807del
ENST00000625778.2:c.1745_1747del
ENST00000630424.2:c.1745_1747del
ENST00000631247.1:n.294_296del
NM_000462.3:c.1814_1816del
NM_130839.2:c.1805_1807del
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NM_001354505.1:c.1805_1807del
NM_001354506.1:c.1745_1747del
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NM_001354512.1:c.1745_1747del
NM_001354513.1:c.1745_1747del
NM_001354523.1:c.1745_1747del
NM_001354526.1:c.1745_1747del
NM_001354538.1:c.1805_1807del
NM_001354539.1:c.1745_1747del
NM_001354540.1:c.1745_1747del
NM_001354541.1:c.1745_1747del
NM_001354542.1:c.1745_1747del
NM_001354543.1:c.1745_1747del
NM_001354544.1:c.1745_1747del
NM_001354545.1:c.1805_1807del
NM_001354546.1:c.1628_1630del
NM_001354547.1:c.1745_1747del
NM_001354548.1:c.1745_1747del
NM_001354549.1:c.1745_1747del
NM_001354550.1:c.554_556del
NM_001354551.1:c.494_496del
NM_130838.3:c.1745_1747del
NM_130839.4:c.1805_1807del
NR_146177.1:n.18393-34749_18393-34747del
NR_148916.1:n.2349_2351del
NM_001354506.2:c.1745_1747del
NM_001354507.2:c.1745_1747del
NM_001354508.2:c.1745_1747del
NM_001354509.2:c.1745_1747del
NM_001354511.2:c.1745_1747del
NM_001354512.2:c.1745_1747del
NM_001354513.2:c.1745_1747del
NM_001354523.2:c.1745_1747del
NM_001354538.2:c.1805_1807del
NM_001354539.2:c.1745_1747del
NM_001354540.2:c.1745_1747del
NM_001354541.2:c.1745_1747del
NM_001354542.2:c.1745_1747del
NM_001354543.2:c.1745_1747del
NM_001354544.2:c.1745_1747del
NM_001354545.2:c.1805_1807del
NM_001354546.2:c.1628_1630del
NM_001354547.2:c.1745_1747del
NM_001354548.2:c.1745_1747del
NM_001354549.2:c.1745_1747del
NM_001354550.2:c.554_556del
NM_001354551.2:c.494_496del
NM_001374461.1:c.1745_1747del
NM_130838.4:c.1745_1747del
NM_130839.5:c.1805_1807del
NR_148916.2:n.2317_2319del

Likely Pathogenic

Met criteria codes 4

PM6_Strong | PM2_Supporting | PS4_Supporting | PP4

Evidence Links 1

Expert Panel

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.Ser582del variant in UBE3A is absent from gnomAD (PM2_Supporting). The p.Ser582del variant in UBE3A has been reported in at least 2 de novo occurrences (biological parentage unconfirmed) in individuals with Angelman syndrome (PMID 25212744, internal database) (PM6_Strong). The p.Ser582del variant has been observed in at least 1 other individual with Angelman syndrome (PMID 25212744) (PS4_Supporting). The p.Ser582del variant in UBE3A has been reported in an individual with a clinical phenotype suggestive of Angelman syndrome (PMID 25212744) (PP4). In summary, the p.Ser582del variant in UBE3A is classified as likely pathogenic for Angelman syndrome based on the ACMG/AMP criteria (PM2_supporting, PM6_strong, PS4_supporting, PP4).

Met criteria codes

PM6_Strong ✔	The p.Ser582del variant in UBE3A has been reported in at least 2 de novo occurrences (biological parentage unconfirmed) in individuals with Angelman syndrome (PMID 25212744, internal database) Review of UBE3A variants identified in a cohort of 2515 AS patients. Variant identified in 1 patient, found to be de novo PubMed:25212744
PM2_Supporting ✔	The p.Ser582del variant in UBE3A is absent from gnomAD
PS4_Supporting ✔	The p.Ser582del variant has been observed in at least 1 other individual with Angelman syndrome (PMID 25212744) Review of UBE3A variants identified in a cohort of 2515 AS patients. Variant identified in 1 patient, found to be de novo PubMed:25212744
PP4 ✔	The p.Ser582del variant in UBE3A has been reported in an individual with a clinical phenotype suggestive of Angelman syndrome (PMID 25212744)

Curation History

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