Variant: NM_130839.5(UBE3A):c.592G>T (p.Ala198Ser)

Version: 1.0

CA267785992

850340 (ClinVar)

Gene: UBE3A (HGNC:7337)
Condition: Angelman syndrome (MONDO:0007113)
Inheritance Mode: Autosomal dominant inheritance
UUID: e83dd0cf-c86a-44e9-a79b-24e43f81a16d
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HGVS expressions

NM_130839.5:c.592G>T
NM_130839.5(UBE3A):c.592G>T (p.Ala198Ser)
NC_000015.10:g.25371582C>A
CM000677.2:g.25371582C>A
NC_000015.9:g.25616729C>A
CM000677.1:g.25616729C>A
NC_000015.8:g.23167822C>A
NG_009268.1:g.72400G>T
ENST00000438097.6:c.532G>T
ENST00000625778.3:c.532G>T
ENST00000635914.1:c.532G>T
ENST00000637886.1:c.592G>T
ENST00000638011.1:c.532G>T
ENST00000638155.1:c.532G>T
ENST00000648336.2:c.592G>T
ENST00000649550.1:c.532G>T
ENST00000650110.1:c.601G>T
ENST00000675000.1:n.1267G>T
ENST00000675177.1:c.415G>T
ENST00000675593.1:n.3288G>T
ENST00000232165.7:c.532G>T
ENST00000397954.6:c.601G>T
ENST00000428984.6:c.532G>T
ENST00000438097.5:c.532G>T
ENST00000566215.5:c.532G>T
ENST00000614096.4:c.592G>T
ENST00000625778.2:c.532G>T
ENST00000626068.2:c.613G>T
ENST00000626793.2:n.643G>T
ENST00000630424.2:c.532G>T
NM_000462.3:c.601G>T
NM_130838.1:c.532G>T
NM_130839.2:c.592G>T
NM_000462.5:c.601G>T
NM_001354505.1:c.592G>T
NM_001354506.1:c.532G>T
NM_001354507.1:c.532G>T

NM_001354508.1:c.532G>T
NM_001354509.1:c.532G>T
NM_001354511.1:c.532G>T
NM_001354512.1:c.532G>T
NM_001354513.1:c.532G>T
NM_001354523.1:c.532G>T
NM_001354526.1:c.532G>T
NM_001354538.1:c.592G>T
NM_001354539.1:c.532G>T
NM_001354540.1:c.532G>T
NM_001354541.1:c.532G>T
NM_001354542.1:c.532G>T
NM_001354543.1:c.532G>T
NM_001354544.1:c.532G>T
NM_001354545.1:c.592G>T
NM_001354546.1:c.415G>T
NM_001354547.1:c.532G>T
NM_001354548.1:c.532G>T
NM_001354549.1:c.532G>T
NM_001354550.1:c.361+3883G>T
NM_001354551.1:c.301+3883G>T
NM_130838.3:c.532G>T
NM_130839.4:c.592G>T
NR_146177.1:n.18393-20014C>A
NR_148916.1:n.1140G>T
NM_001354506.2:c.532G>T
NM_001354507.2:c.532G>T
NM_001354508.2:c.532G>T
NM_001354509.2:c.532G>T
NM_001354511.2:c.532G>T
NM_001354512.2:c.532G>T
NM_001354513.2:c.532G>T
NM_001354523.2:c.532G>T
NM_001354538.2:c.592G>T
NM_001354539.2:c.532G>T
NM_001354540.2:c.532G>T
NM_001354541.2:c.532G>T
NM_001354542.2:c.532G>T
NM_001354543.2:c.532G>T
NM_001354544.2:c.532G>T
NM_001354545.2:c.592G>T
NM_001354546.2:c.415G>T
NM_001354547.2:c.532G>T
NM_001354548.2:c.532G>T
NM_001354549.2:c.532G>T
NM_001354550.2:c.361+3883G>T
NM_001354551.2:c.301+3883G>T
NM_001374461.1:c.532G>T
NM_130838.4:c.532G>T
NR_148916.2:n.1108G>T

Uncertain Significance

Met criteria codes 1

BP4

Not Met criteria codes 1

BS1

Evidence Links 0

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 c.532G>T p.Ala178Ser variant in UBE3A (NM_130838.2) is present in gnomAD v2.1.1 at a frequency of 0.0023% in the European (non-Finnish) sub population (no criteria met). Computational analysis prediction tools suggest that the p.Ala178Ser variant does not have a deleterious impact; however this information does not predict clinical significance on its own. In summary, the c.926A>G p.Asn309Ser variant in UBE3A is classified as a Variant of Uncertain Significance based on the ACMG/AMP criteria (BP4).

Met criteria codes

BP4	✔	Computational analysis prediction tools suggest that the p.(Ala178Ser) variant does not have a deleterious impact; however this information does not predict clinical significance on its own.

Not Met criteria codes

BS1	✖	The c.532G>T p.(Ala178Ser) variant in UBE3A (NM_130838.2) is present in gnomAD v2.1.1 at a frequency of 0.0023% in the European (non-Finnish) sub population (no criteria met).

Curation History

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