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Variant: NM_130839.5(UBE3A):c.1269C>T (p.Asp423=)

Version: 1.0

CA149398

96256 (ClinVar)

Gene: UBE3A (HGNC:7337)
Condition: Angelman syndrome (MONDO:0007113)
Inheritance Mode: Autosomal dominant inheritance
UUID: 4c851045-b14e-47ce-9c9a-38d49f63e863
Approved on: 2022/9/1
Published on: 2022/9/6

HGVS expressions

NM_130839.5(UBE3A):c.1269C>T (p.Asp423=)
NM_130839.5:c.1269C>T
NC_000015.10:g.25370905G>A
CM000677.2:g.25370905G>A
NC_000015.9:g.25616052G>A
CM000677.1:g.25616052G>A
NC_000015.8:g.23167145G>A
NG_009268.1:g.73077C>T
ENST00000438097.6:c.1209C>T
ENST00000625778.3:c.1209C>T
ENST00000635914.1:c.1209C>T
ENST00000637886.1:c.1269C>T
ENST00000638011.1:c.1209C>T
ENST00000638155.1:c.1209C>T
ENST00000648336.2:c.1269C>T
ENST00000649550.1:c.1209C>T
ENST00000650110.1:c.1278C>T
ENST00000675000.1:n.1944C>T
ENST00000675177.1:c.1092C>T
ENST00000675593.1:n.3965C>T
ENST00000232165.7:c.1209C>T
ENST00000397954.6:c.1278C>T
ENST00000428984.6:c.1209C>T
ENST00000438097.5:c.1209C>T
ENST00000566215.5:c.1209C>T
ENST00000614096.4:c.1269C>T
ENST00000625778.2:c.1209C>T
ENST00000630424.2:c.1209C>T
NM_000462.3:c.1278C>T
NM_130838.1:c.1209C>T
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NM_001354506.1:c.1209C>T
NM_001354507.1:c.1209C>T
NM_001354508.1:c.1209C>T
NM_001354509.1:c.1209C>T

NM_001354511.1:c.1209C>T
NM_001354512.1:c.1209C>T
NM_001354513.1:c.1209C>T
NM_001354523.1:c.1209C>T
NM_001354526.1:c.1209C>T
NM_001354538.1:c.1269C>T
NM_001354539.1:c.1209C>T
NM_001354540.1:c.1209C>T
NM_001354541.1:c.1209C>T
NM_001354542.1:c.1209C>T
NM_001354543.1:c.1209C>T
NM_001354544.1:c.1209C>T
NM_001354545.1:c.1269C>T
NM_001354546.1:c.1092C>T
NM_001354547.1:c.1209C>T
NM_001354548.1:c.1209C>T
NM_001354549.1:c.1209C>T
NM_001354550.1:c.361+4560C>T
NM_001354551.1:c.301+4560C>T
NM_130838.3:c.1209C>T
NM_130839.4:c.1269C>T
NR_146177.1:n.18393-20691G>A
NR_148916.1:n.1817C>T
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NM_001354508.2:c.1209C>T
NM_001354509.2:c.1209C>T
NM_001354511.2:c.1209C>T
NM_001354512.2:c.1209C>T
NM_001354513.2:c.1209C>T
NM_001354523.2:c.1209C>T
NM_001354538.2:c.1269C>T
NM_001354539.2:c.1209C>T
NM_001354540.2:c.1209C>T
NM_001354541.2:c.1209C>T
NM_001354542.2:c.1209C>T
NM_001354543.2:c.1209C>T
NM_001354544.2:c.1209C>T
NM_001354545.2:c.1269C>T
NM_001354546.2:c.1092C>T
NM_001354547.2:c.1209C>T
NM_001354548.2:c.1209C>T
NM_001354549.2:c.1209C>T
NM_001354550.2:c.361+4560C>T
NM_001354551.2:c.301+4560C>T
NM_001374461.1:c.1209C>T
NM_130838.4:c.1209C>T
NR_148916.2:n.1785C>T

Met criteria codes 2

Expert Panel

Benign

BP7 BA1

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 allele frequency of the p.Asp423= variant in UBE3A is 0.18% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). The silent p.Asp423= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Asp423= variant in UBE3A is classified as benign based on the ACMG/AMP criteria (BA1, BP7).

Met criteria codes

BP7	✔	The silent p.Asp423= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7).
BA1	✔	The allele frequency of the p.Asp423= variant in UBE3A is 0.18% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions

Curation History

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