

Variant: *NM_014297.5(ETHE1):c.6G>A (p.Ala2=)*

Version: 1.0

CA9487981 [↗](#)

260385 (ClinVar) [↗](#)

Gene: ETHE1 ([HGNC:23474](#))

Condition: ethylmalonic encephalopathy ([MONDO:0011229](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 1931d802-2e7e-48dd-b06f-68f862cfb491

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HGVS expressions

NM_014297.5:c.6G>A

NM_014297.5(ETHE1):c.6G>A (p.Ala2=)

NC_000019.10:g.43527172C>T

CM000681.2:g.43527172C>T

NC_000019.9:g.44031324C>T

CM000681.1:g.44031324C>T

NC_000019.8:g.48723164C>T

NG_008141.1:g.5073G>A

ENST00000292147.7:c.6G>A

ENST00000292147.6:c.6G>A

ENST00000458714.2:c.135+546C>T

ENST00000594342.5:c.6G>A

ENST00000595115.1:n.59G>A

ENST00000598330.1:c.6G>A

ENST00000600651.5:c.6G>A

ENST00000602138.1:c.6G>A

NM_014297.3:c.6G>A

NM_001320867.1:c.6G>A

NM_001320868.1:c.-215G>A

NM_001320869.1:c.6G>A

NM_014297.4:c.6G>A

NM_001320867.2:c.6G>A

NM_001320868.2:c.-215G>A

NM_001320869.2:c.6G>A

Benign

Met criteria codes 3

BS2 BP7 BA1

Not Met criteria codes 4

BS1 PS1 PM5 PM2

Evidence Links 0

Expert Panel

Mitochondrial Diseases VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Mitochondrial Diseases VCEP

The allele frequency of the c.6G>A variant in the ETHE1 gene is reported as >16% in gnomAD, including >2,000 homozygotes, which is high enough to be classified as benign based on thresholds defined by the ClinGen ETHE1 Variant Curation Expert Panel (>0.1% in gnomAD- BA1 and BS2). In silico splicing predictors (Splice AI) do not predict a deleterious effect (BP7). In summary, this variant meets criteria to be classified as benign for ETHE1-related ethylmalonic encephalopathy. ETHE1-specific ACMG/AMP criteria applied: (BA1, BS2, BP7).

Met criteria codes

BS2			Allele frequency is reported as >16% in gnomAD, including >2,000 homozygotes
BP7			This is a synonymous change that is not predicted to affect splicing. Splice AI score is 0.0 for acceptor loss and gain and 0.0 for donor loss and gain
BA1			Allele frequency is reported as >16% in gnomAD, including >2,000 homozygotes, which is higher than the cutoff proposed for ETHE1 variants at >0.1%

Not Met criteria codes

BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			Multiple individuals are homozygotes for this mutation in ExAC

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