

Variant: NM_002055.5(GFAP):c.1140C>T (p.Pro380=)

Version: 1.0

CA217126 [↗](#)

66446 (ClinVar) [↗](#)

Gene: GFAP ([HGNC:2670](#))

Condition: Alexander disease ([MONDO:0008752](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 2381c78a-d14e-425b-993d-f0ef29f0d9e1

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HGVS expressions

NM_002055.5(GFAP):c.1140C>T (p.Pro380=)

NM_002055.5:c.1140C>T

NC_000017.11:g.44910646G>A

CM000679.2:g.44910646G>A

NC_000017.10:g.42988014G>A

CM000679.1:g.42988014G>A

NC_000017.9:g.40343540G>A

NG_008401.1:g.9901C>T

ENST00000253408.11:c.1140C>T

ENST00000435360.8:c.1140C>T

ENST00000253408.10:c.1140C>T

ENST00000435360.7:c.1140C>T

ENST00000585543.6:n.293C>T

ENST00000586125.2:c.75C>T

ENST00000586127.6:n.1669C>T

ENST00000588735.3:c.1140C>T

ENST00000591327.2:n.2294C>T

ENST00000591880.2:c.70C>T

ENST00000592320.6:c.717C>T

ENST00000638281.1:c.1140C>T

ENST00000638304.1:c.59C>T

ENST00000638488.1:n.81C>T

ENST00000638618.1:c.795C>T

ENST00000639042.1:c.77C>T

ENST00000639277.1:c.1140C>T

ENST00000639921.1:c.313C>T

ENST00000640552.1:n.1154C>T

ENST00000253408.9:c.1140C>T

ENST00000435360.6:c.1140C>T

ENST00000585543.5:n.293C>T

ENST00000586793.5:c.1140C>T

ENST00000588640.5:n.520C>T

ENST00000588735.1:c.83-2530C>T

ENST00000591880.1:c.6C>T

ENST00000592320.5:c.717C>T

NM_001131019.2:c.1140C>T

NM_001242376.1:c.1140C>T

NM_002055.4:c.1140C>T
NM_001363846.1:c.1140C>T
NM_001131019.3:c.1140C>T
NM_001242376.2:c.1140C>T
NM_001242376.3:c.1140C>T
NM_001363846.2:c.1140C>T

Benign

Met criteria codes 2

BP7 BA1

Evidence Links 0

Expert Panel

Leukodystrophy and Leukoencephalopathy VCEP

Criteria Specification Information

[Criteria Specification:](#) ClinGen Leukodystrophy and Leukoencephalopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GFAP Version 1.0

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Leukodystrophy and Leukoencephalopathy VCEP

The c.1140C>T (NM_002055.5(GFAP)) variant in GFAP is a synonymous (silent) variant (p.Pro380=) that is not predicted to impact the splice consensus sequence, nor the creation of a new splice site, and the nucleotide is not highly conserved with a phyloP score of <2.27 (BP7). The GrpMax filtering allele frequency in gnomAD v4.1.0 is 0.0001188 from the African/African American genetic ancestry group (the lower threshold of the 95% CI of 21/74738 alleles), which is higher than the ClinGen Leukodystrophy and Leukoencephalopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GFAP Version 1.0.0 threshold $\geq 0.01\%$ for BA1, and therefore meets this criterion (BA1). In summary, this variant meets the criteria to be classified as likely benign based on the ACMG/AMP criteria applied, as specified by the ClinGen Leukodystrophy and Leukoencephalopathy VCEP: BA1, BP7 (ClinGen Leukodystrophy and Leukoencephalopathy VCEP Specifications Version 1.0.0).

Met criteria codes

BP7



The c.1140C>T (p.Pro380=) variant is a synonymous (silent) variant that is not predicted to impact the splice consensus sequence, nor the creation of a new splice site, and the nucleotide is not highly conserved with a phyloP score of <2.27 (BP7).

BA1



The GrpMax filtering allele frequency in gnomAD v4.1.0 is 0.0001188 from the African/African American genetic ancestry group (the lower threshold of the 95% CI of 21/74738 alleles), which is higher than the ClinGen Leukodystrophy and Leukoencephalopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GFAP Version 1.0.0 threshold $\geq 0.01\%$ for BA1, and therefore meets this criterion (BA1).

Curation History

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