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Variant: NM_001142805.2:c.676G>T

Version: 1.0

CA415079810

3066449 (ClinVar)

Gene: SLC6A8 (HGNC:6535)
Condition: creatine transporter deficiency (MONDO:0010305)
Inheritance Mode: X-linked inheritance
UUID: 70581c7b-0743-4ebc-b2f6-b8c4415011df
Approved on: 2024/3/21
Published on: 2024/3/25

HGVS expressions

NM_001142805.2:c.676G>T
NC_000023.11:g.153692006G>T
CM000685.2:g.153692006G>T
NC_000023.10:g.152957461G>T
CM000685.1:g.152957461G>T
NC_000023.9:g.152610655G>T
NG_012016.1:g.8710G>T
NG_012016.2:g.8710G>T
ENST00000253122.10:c.676G>T
ENST00000675713.1:n.430G>T
ENST00000253122.9:c.676G>T
ENST00000429147.1:c.125G>T
ENST00000430077.6:c.331G>T
ENST00000466243.1:n.468G>T
ENST00000467402.1:n.145+499G>T
NM_001142805.1:c.676G>T
NM_001142806.1:c.331G>T
NM_005629.3:c.676G>T
NM_005629.4:c.676G>T

Pathogenic

Met criteria codes 3

PVS1 PP4 PM2_Supporting

Evidence Links 0

Expert Panel

Cerebral Creatine Deficiency Syndromes VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Cerebral Creatine Deficiency Syndromes Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for SLC6A8 Version 1.1.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Cerebral Creatine Deficiency Syndromes VCEP

The NM_005629.4:c.676G>T variant in SLC6A8 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 4/13 (p.Glu226Ter) leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant has been previously reported in one male individual with elevated urinary creatine/creatinine (PMID: 23644449) (PP4). The variant is absent in gnomAD v2.1.1. (PM2_Supporting). In summary, this variant meets criteria to be classified as pathogenic for creatine transporter deficiency. ACMG/AMP SLC6A8-specific criteria applied, as specified by the ClinGen CCDS VCEP (Specifications Version 1.1.0): PVS1, PM2_Supporting, PP4. (Classification approved by the ClinGen CCDS VCEP on March 21, 2024).

Met criteria codes

PVS1	✔	The NM_005629.4:c.676G>T variant in SLC6A8 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 4/13 (p.Glu226Ter) leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PP4	✔	This variant has been previously reported in one male individual with elevated urinary creatine/creatinine (PMID: 23644449) (PP4).
PM2_Supporting	✔	The variant is absent in gnomAD v2.1.1. (PM2_Supporting).

Curation History

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