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Variant: NM_005629.4(SLC6A8):c.467_469del (p.Phe156del)

Version: 1.0

CA1139667841

941841 (ClinVar)

Gene: SLC6A8 (HGNC:6535)
Condition: creatine transporter deficiency (MONDO:0010305)
Inheritance Mode: X-linked inheritance
UUID: f718bb4d-d807-4b74-b951-9ac606749496
Approved on: 2024/6/25
Published on: 2024/6/25

HGVS expressions

NM_005629.4:c.467_469del
NM_005629.4(SLC6A8):c.467_469del (p.Phe156del)
NC_000023.11:g.153691376_153691378del
CM000685.2:g.153691376_153691378del
NC_000023.10:g.152956831_152956833del
CM000685.1:g.152956831_152956833del
NC_000023.9:g.152610025_152610027del
NG_012016.1:g.8080_8082del
NG_012016.2:g.8080_8082del
ENST00000253122.10:c.467_469del
ENST00000675713.1:n.221_223del
ENST00000253122.9:c.467_469del
ENST00000430077.6:c.122_124del
ENST00000466243.1:n.259_261del
ENST00000467402.1:n.14_16del
NM_001142805.1:c.467_469del
NM_001142806.1:c.122_124del
NM_005629.3:c.467_469del
NM_001142805.2:c.467_469del

Uncertain Significance

Met criteria codes 3

PM2_Supporting PP3 PM4

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.467_469del variant in SLC6A8 is a deletion of 3 nucleotides within exon 3/13 of the SLC6A8 gene and is predicted to lead to an in-frame deletion of 1 amino acid (p.Phe156del) (PM4). This variant has been previously reported (PMID: 22281021); however, no data, including zygosity and/or phenotypic features, regarding the individual/s with this variant is provided. The variant is absent in gnomAD v2.1.1. (PM2_Supporting). The in silico predictors MutPredIndel and MutationTaster predict that the variant impacts SLC6A8 function (PP3) There is a ClinVar entry for this variant (Variation ID: 941841, 1 star review status), with one submitter classifying it as a variant of uncertain significance and one submitter classifying it as likely pathogenic. In summary, this variant meets criteria to be classified as a variant of uncertain significance for creatine transporter deficiency. SLC6A8-specific criteria applied, as specified by the ClinGen CCDS VCEP (Specifications Version 1.1.0): PM4, PP3, PM2_Supporting. (Classification approved by the ClinGen CCDS VCEP on June 25, 2024)

Met criteria codes

PM2_Supporting	✔	The variant is absent in gnomAD v2.1.1. (PM2_Supporting).
PP3	✔	The in silico predictor MutPredIndel gave a score of 0.83337 (0.70 or above yields an false of 5%), and MutationTaster predicted that the variant is "disease-causing" (PP3)
PM4	✔	The NM_005629.4:c.467_469del variant in SLC6A8 is a deletion of 3 amino acids within exon 3/13 of the SLC6A8 gene and is predicted to lead to an in-frame deletion of 1 amino acid (p.Phe156del) (PM4).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_005629.4(SLC6A8):c.467_469del (...	Uncertain Significance	Creatine Transporter Deficiency	2024-06-25	1.0	ClinGen Cerebral Creatine Deficiency Syndromes Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for SLC6A8 Version 1.1.0	SLC6A8

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