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Variant: NM_000277.1(PAH):c.208_210delTCT (p.Ser70del)

CA229490

102632 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: ea84cf1d-348b-4e04-a809-550ce2b8d111
Approved on: 2018-08-05
Published on: 2018-12-10

HGVS expressions

NM_000277.1:c.207_209delTTC
NM_000277.1(PAH):c.208_210delTCT (p.Ser70del)
NM_000277.1:c.208_210delTCT
NM_000277.1:c.206_208delCTT
NM_000277.1:c.208_210del
NM_000277.2:c.208_210del
NM_001354304.1:c.208_210del
NM_000277.3:c.208_210del
ENST00000307000.7:c.193_195del
ENST00000546844.1:c.208_210del
ENST00000548677.2:n.295_297del
ENST00000548928.1:n.130_132del
ENST00000549111.5:n.304_306del
ENST00000550978.6:n.192_194del
ENST00000551337.5:c.208_210del
ENST00000551988.5:n.297_299del
ENST00000553106.5:c.208_210del
ENST00000635500.1:n.176_178del
NC_000012.12:g.102894879_102894881del
CM000674.2:g.102894879_102894881del
NC_000012.11:g.103288657_103288659del
CM000674.1:g.103288657_103288659del
NC_000012.10:g.101812787_101812789del
NG_008690.1:g.27724_27726del
NG_008690.2:g.68532_68534del

Pathogenic

Met criteria codes 5

PS3 PP4 PM2 PM3 PM4

Evidence Links 1

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

PAH-specific ACMG/AMP criteria applied: PP4: Phe>120 umol/L with PKU (PMID:25456745); PM3: In trans with: c.842+2T>A (P, ClinGen) (PMID:25456745); PS3: 0% in BioPKU; PM2: Extremely low frequency. ExAC MAF=0.00012; PM4: In frame deletion. In summary this variant meets criteria to be classified as pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PP4, PM3, PS3, PM2, PM4).

Met criteria codes

PS3		✔	0% in BioPKU
PP4	ⓘ	✔	Phe>120 umol/L with PKU Phe>120 umol/L with PKU **PubMed:25456745**
PM2		✔	Extremely low frequency. ExAC MAF=0.00012
PM3	ⓘ	✔	In trans with: c.842+2T>A (P, ClinGen) In trans with: c.1045T>G, p.Ser349Ala (not provided, ClinGen) c.842+2T>A, p.IVS7+2T>A (P, ClinGen) **PubMed:25456745**
PM4	ⓘ	✔	In frame deletion

Curation History

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