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Variant: NM_000152.5(GAA):c.1824_1828dup (p.Ala610fs)

Version: 1.0

CA16041896

370866 (ClinVar)

Gene: GAA (HGNC:2548)
Condition: glycogen storage disease II (MONDO:0009290)
Inheritance Mode: Autosomal recessive inheritance
UUID: 9dd079f9-ba49-4a93-908f-77d8ea766c37
Approved on: 2020/10/29
Published on: 2020/11/12

HGVS expressions

NM_000152.5:c.1824_1828dup
NM_000152.5(GAA):c.1824_1828dup (p.Ala610fs)
NC_000017.11:g.80112647_80112651dup
CM000679.2:g.80112647_80112651dup
NC_000017.10:g.78086446_78086450dup
CM000679.1:g.78086446_78086450dup
NC_000017.9:g.75701041_75701045dup
NG_009822.1:g.16092_16096dup
ENST00000570803.6:c.1824_1828dup
ENST00000572080.2:c.1824_1828dup
ENST00000577106.6:c.1824_1828dup
ENST00000302262.8:c.1824_1828dup
ENST00000302262.7:c.1824_1828dup
ENST00000390015.7:c.1824_1828dup
ENST00000570716.1:n.264_268dup
ENST00000572080.1:c.212_216dup
ENST00000572803.1:n.438_442dup
NM_000152.3:c.1824_1828dup
NM_001079803.1:c.1824_1828dup
NM_001079804.1:c.1824_1828dup
NM_000152.4:c.1824_1828dup
NM_001079803.2:c.1824_1828dup
NM_001079804.2:c.1824_1828dup
NM_001079803.3:c.1824_1828dup
NM_001079804.3:c.1824_1828dup

Pathogenic

Met criteria codes 4

PP4 PM2 PVS1 PM3_Supporting

Evidence Links 1

Expert Panel

Lysosomal Diseases VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Lysosomal Diseases VCEP

This variant, c.1824_1828dup (p.Ala610AspfsTer88), is a frameshift variant that is predicted to result in a premature termination codon, nonsense mediated decay, and lack of gene product. Therefore, PVS1 can be applied. The variant is absent in gnomAD v2.1.1, meeting PM2. A patient with infantile onset Pompe disease, meeting the ClinGen LSD VCEP's specifications for PP4 has been reported who is homozygous for the variant (PMID 24976573). This data meets PM3_Supporting. There is a ClinVar entry for this variant (Variation ID: 370866, 1 star review status) with one submitter classifying the variant as likely pathogenic. In summary, this variant meets the criteria to be classified as pathogenic for Pompe disease. ACMG/AMP criteria applied, as specified by the ClinGen LSD VCEP: PVS1, PM2, PM3_Supporting, PP4.

Met criteria codes

PP4	✔	One patient with infantile onset Pompe disease has been described with no detectable GAA activity in dried blood spot samples (PMID 24976573), meeting PP4.
PM2	✔	This variant is absent in gnomAD v2.1.1.
PVS1	✔	This is a frameshift variant which is predicted to result in a premature termination codon, nonsense mediated decay, and lack of gene product. Therefore, PVS1 can be applied.
PM3_Supporting	✔	A patient with infantile onset Pompe disease and meeting the Clingen LSD VCEP's specifications for PP4 has been described who is homozygous for the variant (PMID 24976573). 0.5 points were given towards PM3. Therefore, PM3_Supporting is met.

PubMed:24976573

Curation History

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