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Variant: NM_000152.5(GAA):c.1156C>T (p.Gln386Ter)

Version: 1.0

CA274049

188858 (ClinVar)

Gene: GAA (HGNC:2548)
Condition: glycogen storage disease II (MONDO:0009290)
Inheritance Mode: Autosomal recessive inheritance
UUID: 1a79a341-f719-450a-8ae2-b5e03aef8015
Approved on: 2020/4/6
Published on: 2020/5/27

HGVS expressions

NM_000152.5(GAA):c.1156C>T (p.Gln386Ter)
NM_000152.5:c.1156C>T
NC_000017.11:g.80108569C>T
CM000679.2:g.80108569C>T
NC_000017.10:g.78082368C>T
CM000679.1:g.78082368C>T
NC_000017.9:g.75696963C>T
NG_009822.1:g.12014C>T
ENST00000570803.6:c.1156C>T
ENST00000572080.2:c.1156C>T
ENST00000577106.6:c.1156C>T
ENST00000302262.8:c.1156C>T
ENST00000302262.7:c.1156C>T
ENST00000390015.7:c.1156C>T
NM_000152.3:c.1156C>T
NM_001079803.1:c.1156C>T
NM_001079804.1:c.1156C>T
NM_000152.4:c.1156C>T
NM_001079803.2:c.1156C>T
NM_001079804.2:c.1156C>T
NM_001079803.3:c.1156C>T
NM_001079804.3:c.1156C>T

Pathogenic

Met criteria codes 3

PVS1 PM2 PP4

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.1156C>T (p.Gln386Ter), is a nonsense variant that is predicted to result in nonsense mediated decay and lack of gene product, meeting PVS1. This variant is absent in gnomAD v2.1.1, meeting PM3. A Korean patient with infantile onset Pompe disease, meeting the specifications for PP4, has been reported. This patient is compound heterozygous for the variant and c.1857C>G (p.Ser619Arg). This in trans data has been used in the assessment of p.Ser619Arg and was not be included here to avoid circular logic. There is a ClinVar entry for this variant (Variation ID: 188858; 1 star review status) with one submitter classifying the variant as likely pathogenic. In summary, this variant meets criteria to be classified as pathogenic for Pompe disease. ACMG/AMP criteria met, as specified by the ClinGen LSD VCEP: PVS1, PM2, PP4.

Met criteria codes

PVS1	✔	This is a nonsense variant which is predicted to cause nonsense mediated decay resulting in no gene product. Therefore, PVS1 can be applied.
PM2	✔	This variant is absent in gnomAD v2.1.1.
PP4	✔	A Korean patient with GAA deficiency in white blood cells meeting the specifications for PP4 has been reported (PMID 23884227).

PubMed:23884227

Curation History

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