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Product datasheet for AM32801PU-N

GAA Mouse Monoclonal Antibody [Clone ID: 43G7]

Product data:

Product Type:	Primary Antibodies
Clone Name:	43G7
Applications:	IHC, WB
Recommended Dilution:	**Western Blot.** **Immunohistochemistry on Paraffin Sections.**
Reactivity:	Human
Host:	Mouse
Isotype:	IgG2a
Clonality:	Monoclonal
Immunogen:	Derived by fusion of Mouse myeloma cells with spleen cells from a Balb/cHeA mouse hyperimmunized with purified acid Alpha-glucosidase from Human placenta.
Specificity:	Reacts with the two major bands with apparent molecular weights of 76 and 70 kDa, the band with a molecular weight of about 94 kDa and minor bands with apparent molecular weights of less than 67 kDa, when analyzing Alpha-Glucosidase isolated from placenta by Polyacrylamide Gel Electrophoresis in the presence of SDS and a reducing agent.
Formulation:	PBS State: Purified State: Liquid purified IgG fraction Preservative: 0.09% Sodium Azide
Concentration:	lot specific
Conjugation:	Unconjugated
Storage:	Store undiluted at 2-8°C for one month or (in aliquots) at -20°C for longer. Avoid repeated freezing and thawing.
Stability:	Shelf life: one year from despatch.
Predicted Protein Size:	105 kDa (Predicted)
Gene Name:	glucosidase alpha, acid
Database Link:	Entrez Gene 2548 Human P10253

View online »

Background:

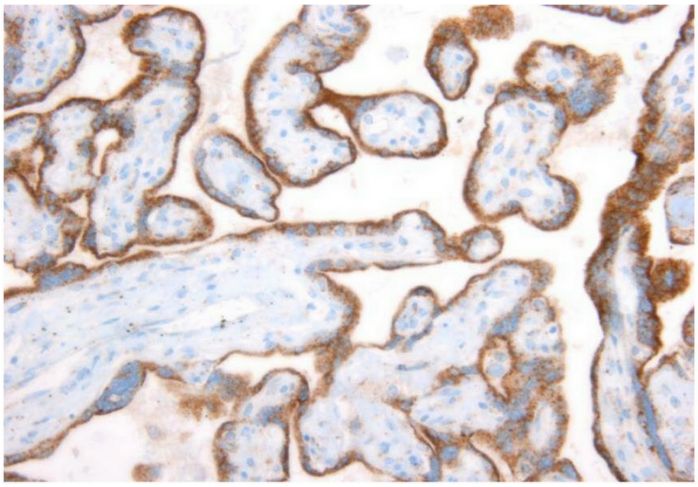
Lysosomal α -glucosidase, like all other lysosomal enzymes of which the 'life-cycle' has been studied, is synthesized as a large precursor that is processed to mature forms of lower molecular mass.
Acid α -glucosidase catalyzes the hydrolysis of the α1 -> 4 and α1 -> 6 glucosidic linkages in glycogen and the α1 -> 4 glucosidic linkage in maltose and artificial substrates likep-nitrophenyl- α -glucoside. The enzyme is deficient in patients with Glycogenosis Type II (Pompe's disease).
Pompe disease (also called Glycogen storage disease type II (GSD II) or acid maltase deficiency) is an autosomal recessive metabolic disorder which damages muscle and nerve cells throughout the body. It is caused by an accumulation of glycogen in the lysosome due to deficiency of the lysosomal acid alpha-glucosidase enzyme. It is the only glycogen storage disease with a defect in lysosomal metabolism.
The build-up of glycogen causes progressive muscle weakness (myopathy) throughout the body and affects various body tissues, particularly in the heart, skeletal muscles, liver and nervous system.
There are exceptions but levels of alpha-glucosidase determines the type of GSD II an individual may have. More alpha glucosidase present in the individuals muscles means symptoms occur later in life and progress more slowly. GSD II is broadly divided into two onset forms based on the age symptoms occur:
• Infantile-onset form is usually diagnosed at 4-8 months; muscles appear normal but are limp and weak preventing them from lifting their head or rolling over. As the disease progresses heart muscles thicken and progressively fail. Without treatment death usually occurs due to heart failure and respiratory weakness.
• Late/later onset form occurs later than one to two years and progresses more slowly than Infantile-onset form. One of the first symptoms is a progressive decrease in muscle strength starting with the legs and moving to smaller muscles in the trunk and arms, such as the diaphragm and other muscles required for breathing. Respiratory failure is the most common cause of death. Enlargement of the heart muscles and rhythm disturbances are not significant features but do occur in some cases.

The disease is caused by a mutation in a gene (acid alpha-glucosidase: also known as acid maltase) on long arm of chromosome 17 at 17q25.2-q25.3 (base pair 75,689,876 to 75,708,272). The number of mutations described is currently (in 2010) 289 with 67 being non-pathogenic mutations and 197 pathogenic mutations. The remainder are still being evaluated for their association with disease.

Synonyms:

Acid maltase, Aglucosidase alfa, GAA

Product images:



Formalin-Fixed, Paraffin-Embedded Human Placenta tissue stained with GAA Antibody AM32801