

Acid Maltase Deficiency (AMD)

(Also known as Pompe Disease)

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What is Pompe disease?

Pompe disease is a metabolic muscle disorder first described in 1932 by Dr JC Pompe. It is a rare neuromuscular genetic disorder that occurs in babies, children, and adults who inherit a defective gene from each of their parents. The disorder has a number of synonyms, the most common are:

Acid Maltase Deficiency

Glycogen Storage Disease Type II (GSD II)

What are the symptoms of Pompe disease?

Progressive muscle weakness is the most common symptom of both the infantile-onset and the late-onset forms of Pompe disease. The muscles most often affected are those used for breathing and mobility. In infants, the heart muscle is often severely affected. Where breathing muscles are affected the patient might suffer from headaches in the morning and an inability to concentrate during the day.

What causes Pompe disease?

In people with Pompe disease, there is a defect in a gene that is responsible for making an enzyme called acid alpha-glucosidase (GAA). Enzymes are proteins that do specific jobs to help keep the cells in the body working normally.

In people with Pompe disease, the GAA enzyme is either missing or in short supply. The job of acid alpha-glucosidase is to break down excess glycogen, a form of sugar stored in muscle cells throughout the body. The missing enzyme leads to a build up of glycogen in the lysosomes of cells. This is why Pompe disease is also referred to as a Lysosomal Storage Disease (LSD).

How is Pompe disease diagnosed?

A conclusive diagnosis of Pompe disease generally requires an enzyme assay test that demonstrates that the patient has deficient

enzyme (acid alpha-glucosidase, "GAA") activity. This is determined by performing tests on a tissue (muscle, skin) or blood sample to verify that the patient's GAA enzyme activity is low or absent. Infantile-onset patients generally display less than 1% of the normal enzyme activity levels in skin cells; late-onset patients generally display less than 40% of the normal enzyme activity levels in skin cells.

Is there any treatment?

An Enzyme Replacement Therapy (ERT), developed by The Genzyme Corporation (www.pompe.com) is now available to patients around the world.

Can Pompe disease be passed on to my children?

The condition is caused by a recessive gene. This means that one abnormal copy of the gene is passed from each parent to the affected child, who in turn will have inherited two abnormal copies of the gene. The risk for both carrier parents to have an affected child is 1 in 4 for each pregnancy.

People who have Pompe Disease do not usually pass the condition onto their own children because one normal copy of the gene will have been provided by their partner. However, all of their children will be carriers. Generally speaking carriers do not have symptoms. Prenatal screening is available for parents who think they may be carriers of Pompe disease or who become pregnant following the diagnosis of the disease in one of their other children. Those families affected by Pompe disease may want to consult with a genetic counsellor to discuss issues of genetic inheritance and family planning.

Information for this fact sheet has been sourced from the Association of Glycogen Storage Disease UK.

For more information about Pompe Disease you can go to the following websites:

The International Pompe Association (IPA) is a federation of Pompe disease patient groups worldwide; currently around 39 countries are represented. It seeks to coordinate activities and share experience and knowledge between the different groups. Its web pages are updated regularly and can be viewed at www.worldpompe.org