Acid Maltase Deficiency Association

What is Pompe Disease?

Pompe disease is a rare condition inherited from both birth parents. The disease effects about 1 in 40,000 people. However, as newborn screening increases, the understanding of the incidence rate is changing.

Pompe disease mainly affects the skeletal, breathing, and in some cases heart muscles. It is a spectrum disease that can range from being severe to mild.

Pompe disease is named after the doctor who first described it (https://amda-pompe.org/wp-content/uploads/2020/06/Joseph_Walter.pdf). Some other names for Pompe disease are:

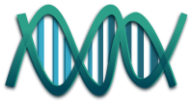
- acid maltase deficiency disease
- glycogen storage disease type 2
- alpha-1,4-glucosidase deficiency

Learn more about inheritance and Pompe disease at Medline Plus:

- Understanding Inheritance
  https://medlineplus.gov/genetics/understanding/inheritance
- Pompe Disease
  https://medlineplus.gov/genetics/condition/pompe-disease/

What Causes Pompe Disease?

The human body is made of many cells. Inside cells are genes that tell cells what to do. The GAA gene tells cells to make a substance called GAA enzyme. It has other names like acid maltase.



GAA helps break down a type of sugar in cells called glycogen. This sugar is stored in lysosomes in the cells. When broken down by GAA, it gives our bodies energy.

When the GAA gene is not working right, there may be no GAA enzyme or only a small amount. This causes Pompe disease. A copy of the non-working GAA gene must be inherited from both parents to cause Pompe.

Without enough GAA enzyme, glycogen builds up in the body's lysosomes. This buildup damages cells in muscles.

Learn more about genes and Pompe:

- GenePossibilities
  https://www.genepossibilities.com/build-understanding-genetics
- National Organization for Rare Disorders
  http://rarediseases.org/videos/pompe-disease/

Types of Pompe Disease

When Pompe disease appears and how severe it is depend, in part, on the mutations to the GAA gene. Some are more severe than others, and cause onset to occur earlier. In general, the younger the age when signs appear, the more severe the progression. There are two main categories of Pompe, but it is a spectrum disease:

1) *Infantile-onset Pompe disease (IOPD).* This type has two forms:
   - Classic – appears in the first few months of life
   - Non-classic – appears by the first year of life

   Learn more about IOPD at the AMDA website: https://amda-pompe.org/iopd
2) *Late-onset Pompe disease (LOPD).* This type appears after the first year of life. It may occur in childhood, teen years, or adulthood. There is a wide range in how LOPD presents and affects a person. Learn more about LOPD at the AMDA website: https://amda-pompe.org/lopd

There are treatments and therapies that can improve the course of Pompe disease.
Babies and all people with the disease can live longer, healthier lives.

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