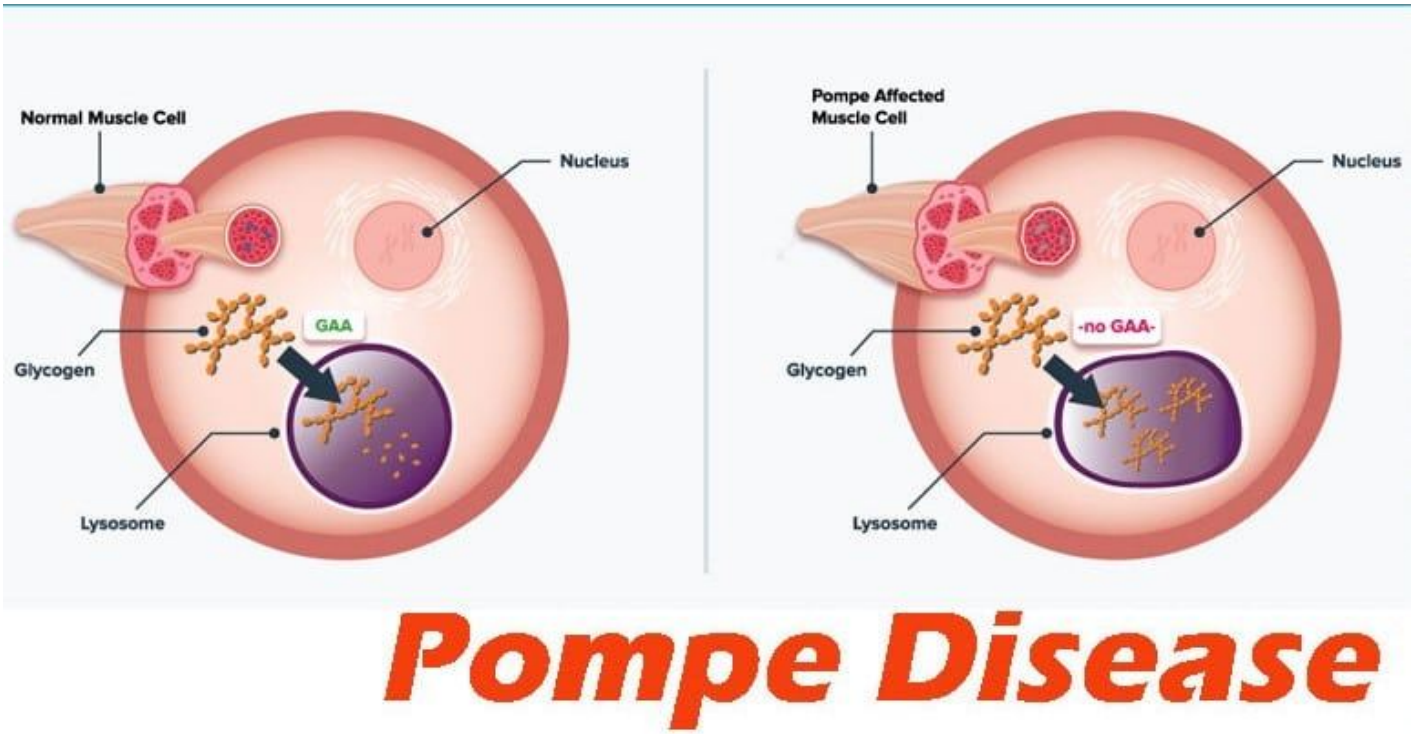
# What is Pompe disease?

December 6, 2023 • vajiramandravi.com




## About Pompe disease

- It is a rare inherited disorder that affects one child per million.
- Causes
    - Mutations in the GAA gene cause Pompe disease.
    - The GAA gene provides instructions for producing an enzyme called acid alpha-glucosidase (also known as acid maltase).
    - This enzyme is active in lysosomes, which are structures that serve as recycling centers within cells.
    - The enzyme normally breaks down glycogen into a simpler sugar called glucose, which is the main energy source for most cells.
    - Mutations in the GAA gene prevent acid alpha-glucosidase from breaking down glycogen effectively, which allows this sugar to build up to toxic levels in lysosomes.
    - This buildup damages organs and tissues throughout the body, particularly the muscles, leading to the progressive signs and symptoms of Pompe disease.
- Some common side effects and symptoms include muscle weakness, respiratory issues, heart problems and difficulty swallowing.

- This disease can be:
    - Infantile-onset: symptoms begin in the first few months after birth.
    - Late-onset or delayed-onset: symptoms appear later in childhood or in adulthood.
- It affects males and females equally.
- Treatment: The treatment includes enzyme replacement therapy (ERT).

---

## Q1) What is enzyme?

An enzyme is a biological catalyst and is almost always a protein. It speeds up the rate of a specific chemical reaction in the cell. The enzyme is not destroyed during the reaction and is used over and over.

**Source:** Being Nidhi's parents: A 24-year journey of joy and struggles with India's 'first' Pompe disease patient