Pompe disease

What is Pompe disease?

Pompe disease is a progressive disease that involves the accumulation of a complex sugar molecule (called glycogen) in the various muscles of the body, particularly respiratory and heart muscles.

Why does it occur?

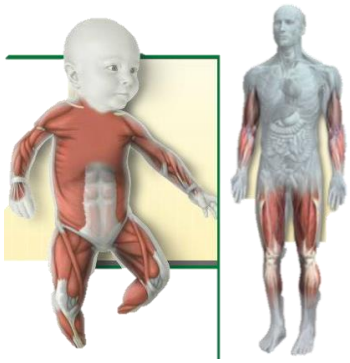
It occurs due to a defect in a GAA gene that encodes for an enzyme called acid alpha-glucosidase. This enzyme normally breaks the complex sugar into simple sugars such as glucose. Glucose is used by the cells including muscle cells to produce energy.

What are the symptoms of Pompe disease?

Depending upon the severity and the onset it can be classic Infantile, non-classic infantile Pompe disease, and late-onset Pompe disease. Infantile onset shows symptoms during the first year of life in the form of

- Feeding difficulties
- Loosening of the body
- Difficulty in achieving motor milestones
- Breathing difficulties
- Repeated respiratory infection
- Increased heart size
- Mild liver enlargement

Late-onset Pompe disease is less severe and is apparent in later childhood, adolescence, or even adulthood. It results in difficulty in climbing stairs, frequent falls, breathing problems and may require a ventilator.



Muscles involvement in infantile onset and Late-onset Pompe disease
(Source-modified from https://memorang.com/flashcards/193283/Rapid+Review+-+Classic+Presentations+4)

Does Pompe disease affect survival?

Yes! As it can involve major muscles of respiration and heart, it can be fatal if not given immediate care upon diagnosis.

What are the treatment options available for Pompe disease?

At present, there is NO cure for Pompe disease. However, Enzyme replacement therapy (ERT) in addition to physical therapy, breathing exercises as per requirement can be helpful in the improvement of the above-mentioned symptoms.

What is the response to ERT?

The overall response to therapy for Pompe disease is satisfactory. This therapy slows the progression of muscle and heart related problems . Early initiation of therapy is the key . The response is variable and depends upon the antibody status to ERT and requires constant follow-up.

What are the approximate cost of ERT for a 10 kg child and a 50 kg adult?

Currently, Myozyme, Sanofi Genzyme provides enzyme replacement therapy.
For a 10 kg child will be around 45-50 lakhs per year
For a 50 kg person – 2.7 crores per year

Do the patients with Pompe disease require regular follow-up for better care?

Yes! Patients with Pompe disease require 4-6 monthly follow up for their clinical and lab assessment depending upon the severity.

How frequently the ERT is given?

It is given intravenously every two weeks for lifelong.

How can Pompe disease be prevented?

Families having affected children are encouraged to consult a clinical geneticist for counseling and prevention of recurrence in future pregnancies.