# India’s first Pompe disease patient passes away

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## What’s in today’s article?

- Why in news?
- Pompe disease
- What is Pompe Disease?
- How does Pompe disease affect an individual?
- How is Pompe disease diagnosed?

- Is Pompe disease curable?

## Why in news?

- Nidhi Shirol, India's first Pompe disease patient, passed away last month at the age of 24 years after battling the disease.
- She spent the last six years in a semi-comatose state.

## What is Pompe Disease?

- Also known as Glycogen Storage Disease Type II, Pompe disease is a rare genetic disorder.
- It is caused by a deficiency of the enzyme acid alpha-glucosidase (GAA).
    - This enzyme is crucial for breaking down glycogen into glucose within the lysosomes of cells.
- Its prevalence estimates range from 1 in 40,000 to 1 in 300,000 births. It occurs across diverse ethnicities and populations.

## How does Pompe disease affect an individual?

- Muscle weakness:
    - Progressive muscle weakness is a primary feature of Pompe disease.
    - It affects both skeletal and smooth muscles, leading to difficulties in mobility and daily activities.
    - Weakness in the respiratory muscles can result in breathing difficulties, especially during physical exertion or even while lying down.
- Motor skill delay:
    - Children with the disease may experience delays in achieving motor milestones, such as sitting, crawling, and walking.
    - The degree of motor skill delay can vary, and some individuals may never attain certain motor milestones.
- Degenerative impact on bones:
    - Prolonged muscle weakness and reduced mobility can have a degenerative impact on bones, leading to joint contractures and skeletal deformities.
- Respiratory complications:
    - The weakening of respiratory muscles, including the diaphragm, can have an impact.
    - Patients may experience shortness of breath, respiratory infections, and in severe cases, respiratory failure.
- Cardiac involvement:
    - In some cases, Pompe disease can affect the heart muscles, leading to complications.
    - Symptoms such as heart palpitations, fatigue, and chest pain, may manifest.
- Hypertrophic cardiomyopathy:
    - Pompe disease can cause hypertrophic cardiomyopathy, characterised by the thickening of the heart muscle walls.
    - This can lead to impaired heart functions and cardiovascular symptoms.
- Implications for daily living:

    - Patients may face challenges in performing daily activities independently due to muscle weakness and respiratory limitations.
    - Assistive devices such as wheelchairs and respiratory support equipment may become necessary.

## How is Pompe disease diagnosed?

- It involves a multi-faceted approach.
- Enzyme assays are conducted to measure the activity of acid alpha-glucosidase (GAA), the deficient enzyme.
    - Enzyme tests, often performed on blood or skin cells, provide crucial insights into GAA deficiency.
- Genetic testing identifies mutations in the responsible GAA gene.
    - Genetic analysis confirms the presence of specific mutations associated with Pompe Disease.
- The combination of these diagnostic tools enables healthcare professionals to accurately identify and confirm the disease.

## Is Pompe disease curable?

- While there is currently no cure, there are treatment options available to manage symptoms and improve the patient’s quality of life.
- Enzyme Replacement Therapy (ERT) is a standard treatment, involving the infusion of the missing enzyme to alleviate glycogen buildup.
    - ERT is a medical treatment that replaces a missing or deficient enzyme in the body.

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### Q1) What is Glycogen storage disease type II (GSD2)?

Glycogen storage disease type II (GSD2), also known as Pompe disease, is a metabolic disorder that causes glycogen to build up in muscle tissue. It’s caused by a deficiency of the enzyme α-glucosidase.

### Q2) What is Alpha-glucosidase?

Alpha-glucosidase is an enzyme that breaks down carbohydrates into monosaccharides, which can then be absorbed by the small intestine. It’s located in the brush border of the small intestine.

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**Source:** India’s first Pompe disease patient passes away: What is this rare genetic disorder?