



Pompe Disease

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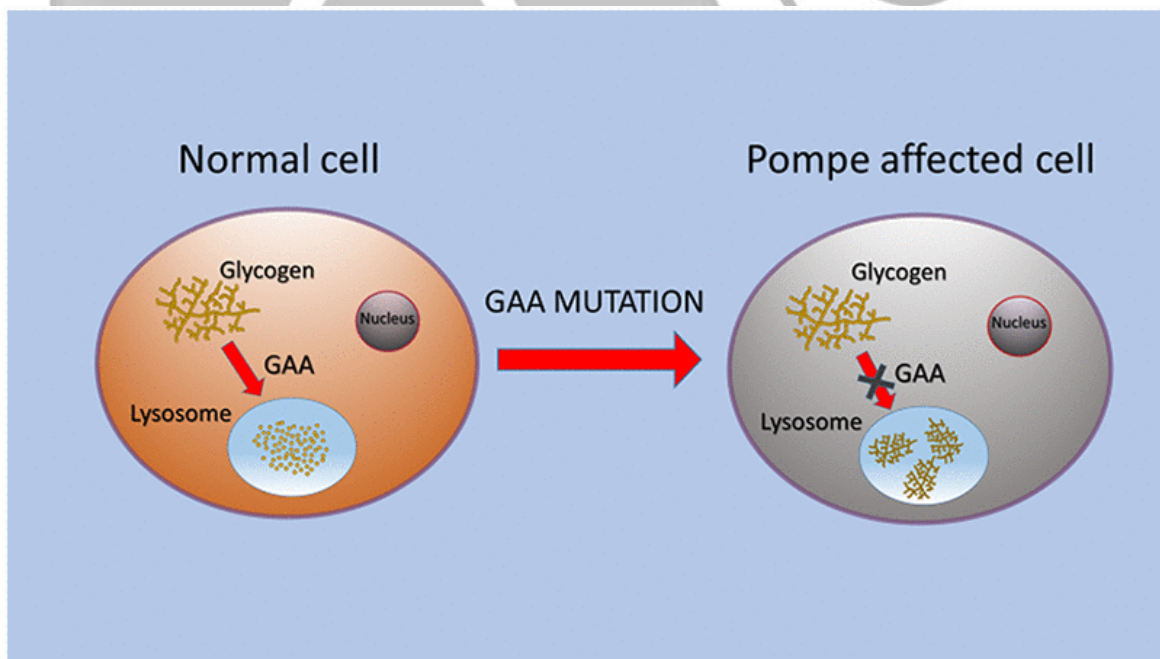
Why in News?

India's **first Pompe disease patient**, passed away at the age of 24 years after battling the disease in a **semi-comatose state**.

- A **semi-comatose state** is characterized by **partial coma**, manifesting as **disorientation** and **stupor** without reaching a complete coma. Individuals in a **semi-comatose state** may exhibit responsiveness to stimuli, such as groaning and mumbling.

What is Pompe Disease?

- **About:**
 - Pompe Disease (also known as Glycogen Storage Disease Type II) is characterized by the **buildup of glycogen in the lysosomes of the body's cells**.
 - This disease is a rare **genetic disorder** 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.
 - Lysosomes are membrane-enclosed organelles that contain an array of enzymes capable of breaking down all types of biological polymers—proteins, nucleic acids, carbohydrates, and lipids.
 - Its prevalence estimates range from **1 in 40,000 to 1 in 300,000 births**.



- **Symptoms:**

- Muscle weakness, Motor skill delay, Degenerative impact on bones, Respiratory complications, Cardiac involvement, Implications for daily living.

▪ **Diagnosis:**

- **Enzyme assays** are conducted to measure the activity of GAA, the deficient enzyme.
- Genetic testing identifies mutations in the responsible **GAA gene**. Genetic analysis confirms the presence of specific mutations associated with Pompe Disease.

▪ **Treatment:**

- Although there is presently no cure for **Pompe disease**, there are treatment alternatives accessible to address symptoms and enhance the patient's quality of life.
- **Enzyme Replacement Therapy (ERT)** is a common treatment method that entails infusing the deficient enzyme to mitigate glycogen accumulation.

PDF Reference URL: <https://www.drishtiias.com/printpdf/pompe-disease>

