Pompe Disease

Source: IE

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.**

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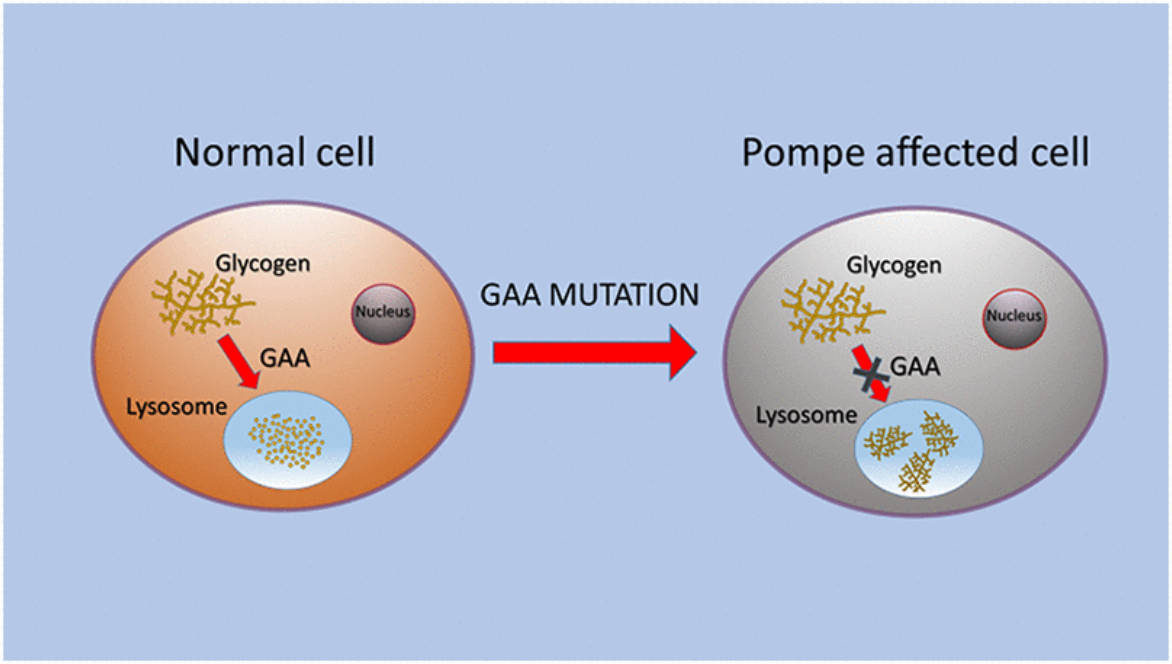

- **Symptoms:**
  - Muscle weakness, Motor skill delay, Degenerative impact on bones, Respiratory complications, Cardiac involvement, Implications for daily living.
- **Diagnosis:**
  - **Enzyme assays** are conducted to measure the activity of GAA, the deficient enzyme.
  - Genetic testing identifies 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.