



## Rare Diseases in India

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### Why in News

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Recently, it was reported that the application of the majority of patients suffering from **Lysosomal Storage Disorders (a rare disease)** has been pending with the Union Ministry of Health and Family Welfare for several months.

- There are more than **2,000 children infected with rare diseases** across the country. Many of them require **Enzyme Replacement Therapy (ERT)**.  
ERT is a medical treatment which replaces an enzyme that is deficient or absent in the body.
- There is also a demand for the **reformulation of National Policy for Treatment of Rare Diseases, 2017**.

### Lysosomal Storage Disorders

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- Lysosomal storage disorder is an **inherited metabolic disease** that is characterized by **an abnormal build-up of various toxic materials** in the **body's cells** as a result of **enzyme deficiencies**.
- It may affect different parts of the body, including the skeleton, brain, skin, heart, and central nervous system.
- There is currently **no approved treatment** for many lysosomal storage diseases.

### Rare Diseases

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- A rare disease is a health condition of low prevalence that affects a **small number of people** compared with other prevalent diseases in the general population.  
There is **no universally accepted definition** of rare diseases and the definitions usually vary across different countries.
- Though rare diseases are of **low prevalence and individually rare**, collectively they affect a **considerable proportion of the population**.

- **80%** of rare diseases are **genetic in origin** and hence disproportionately **impact children**.
- In India there are **56-72 million people** affected by rare diseases.

## National Policy for Treatment of Rare Diseases, 2017

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- The policy highlights the **measures and steps**, both in the **short as well as in the long term**, that need to be taken to deal comprehensively with rare diseases.
- The policy intends to constitute an **Inter-ministerial Consultative Committee** to coordinate and steer the initiatives of different ministries and departments on rare diseases.
- It also mentions for the **creation of a corpus fund at Central and State** level for funding treatment of rare diseases.
- The policy aims to create a **patient registry for diseases** housed in Indian Council of Medical Research (ICMR).
- However, recognizing the higher cost of treatment for rare diseases, the policy also seeks to **strike a balance between access to treatment with health system sustainability**.
- It also aims to create awareness among health professionals, families of patients and the public in general, about rare diseases.

**Source: TH**