



Rare Disease Day 2024

For Prelims: Rare Disease Day, [Rare Disease](#), [World Health Organization](#), [National Policy for Rare Diseases 2021](#)

For Mains: Initiatives Related to Universal Health Coverage in India, Health, Rare Diseases

[Source: TH](#)

Why in News?

Recently, [Rare Disease Day](#) was observed on the **last day of February**. This international awareness day is dedicated to raising awareness of rare diseases and their significant impact on patients and their families.

What is Rare Disease Day?

- Rare Disease Day is a globally coordinated movement dedicated to advocating for **equity in social opportunity, healthcare, and access to diagnosis** and therapies for individuals with rare diseases.
- The theme for Rare Disease Day 2024 is "**Share Your Colours**," which emphasizes collaboration and support.
- It was established in 2008 and observed annually on **28th February (or 29 in leap years)**. Rare Disease Day is coordinated by the **European Organisation for Rare Diseases (EURORDIS)** and over 65 national alliance patient organization partners.
- It serves as a focal point for advocacy work on local, national, and international levels, engaging individuals, families, caregivers, healthcare professionals, researchers, policymakers, industry representatives, and the general public.

What is a Rare Disease?

- **About:**
 - Rare diseases are loosely defined as **occurring infrequently in the population**, with prevalence varying between nations.
 - The [World Health Organization](#) defines rare diseases as often debilitating lifelong conditions with a prevalence of **1 or less per 1000 population**.
 - Different countries have their own definitions; for instance, the US considers diseases affecting fewer than 200,000 patients rare, while the European Union sets the limit at no more than 5 in 10,000 people.
 - **India currently does not have a standard definition**, but the **Organisation of Rare Diseases India (ORDI)** has suggested that a disease is to be defined as rare if it affects 1 in 5,000 people or less.
- **Global Rare Diseases Burden:**
 - **300 million people** worldwide are affected by rare diseases.
 - Rare diseases affect approximately **3.5% to 5.9% of the population**.

- 72% of rare diseases are **genetic**, with over 7000 characterized by diverse disorders and symptoms.
- **75% of rare diseases affect children.** 70% of rare diseases have their onset during childhood.
- **Characteristics and Impact of Rare Diseases:**
 - Rare diseases present with a **broad diversity of disorders and symptoms**, varying not only between diseases but also among patients with the same disease.
 - The **chronic, progressive, degenerative, and often life-threatening nature** of rare diseases significantly affects patients' quality of life.
 - Lack of effective cures exacerbates the pain and suffering endured by patients and their families.
- **Challenges Faced by Individuals with Rare Diseases:**
 - Delayed diagnosis due to lack of **scientific knowledge and quality information.**
 - Inequalities in **access to treatment** and care leading to **social and financial burdens.**
 - **Common symptoms** can mask underlying rare diseases, leading to initial misdiagnosis.
 - According to EURORDIS, 5 years is the time it takes on average for rare disease patients to get a diagnosis.
 - 70% of people with rare diseases wait more than 1 year to get a confirmed diagnosis after coming to medical attention.
 - Physicians' **lack of awareness and training in interpreting rare disease** signs and symptoms contributes to diagnostic challenges.

What is the Landscape of Rare Diseases in India?

- **Impact:**
 - India represents **one-third of global rare disease cases**, encompassing over 450 identified diseases.
 - Despite this significant prevalence, rare diseases remain largely overlooked in India, with limited awareness, diagnosis, and drug development.
 - Over **8 to 10 crore Indians are estimated to suffer from rare diseases**, with **over 75% being children.**
- **Challenges in Policy and Implementation:**
 - The Ministry of Health and Family Welfare formulated a national policy for rare diseases in 2017 but withdrew it in 2018 due to implementation challenges.
 - The revised **first [National Policy for Rare Diseases \(NPRD\)](#) was announced in 2021**, but problems persist, including the **absence of a clear definition for rare diseases.**
- **Treatment Accessibility and Funding:**
 - Less than **50% of identified rare diseases in India are treatable**, with approved treatments available for only about 20 diseases.
 - Access to approved treatments is limited to designated **[Centres of Excellence \(CoEs\)](#)**, which are few (12) in number, unevenly distributed, and often lack coordination.
 - The NPRD guidelines provide limited financial assistance per patient, insufficient for lifelong management and therapy of chronic rare diseases.
- **Challenges in Fund Utilization:**
 - Budget allocations for **rare diseases have increased but remain low**, with Rs 93 crore allocated for 2023-2024.
 - Confusion and disparities in **fund utilization among CoEs** highlight inefficiencies in resource allocation.
 - A staggering 51.3% of allocated funds remain unutilized, despite patients urgently needing treatment.
 - Some CoEs struggle with **underutilization of allocated funds**, while others **exhaust their budgets quickly**, leading to unequal access to treatment.
 - For instance, **Mumbai exhausted all its funds** while treating only 20 out of 107 patients, **Delhi utilized less than 20% of its funds.**
 - The burden of funding treatment often falls on patients and their families, with governmental support falling short.
 - Patients and advocacy groups call for sustainable funding from both the central and state governments to support rare disease treatment.
 - Sustainable funding is crucial for patients, especially those who have exhausted

their allotted funds and are struggling to continue treatment.

National Policy for Rare Diseases (NPRD), 2021

- The NPRD, 2021 aims to reduce the prevalence and incidence of rare diseases.
- Rare diseases are categorized into three groups: **Group 1, Group 2, and Group 3** based on treatment requirements.
 - **Group 1:** Disorders amenable to one-time curative treatment.
 - **Group 2:** Diseases requiring long-term/lifelong treatment with relatively lower treatment costs.
 - **Group 3:** Diseases with definitive treatment available, but challenges exist in patient selection and **high treatment costs**.
- NPRD, 2021 has the provision for financial support of up to Rs. 50 lakhs for patients suffering from any category of rare diseases and for treatment in any of the CoE mentioned in NPRD-2021, outside the Umbrella Scheme of [Rashtriya Arogya Nidhi\(RAN\)](#).
 - **RAN offers a maximum financial assistance of Rs 20 lakh for patients suffering from specified rare diseases.**

Way Forward

- Frame a **standard definition of rare diseases** to provide clarity and consistency in policy implementation.
- Increase **budgetary outlays** dedicated to rare diseases to support drug development, therapy, and research.
 - Expand the number of CoEs for rare diseases and ensure better coordination among them.
 - Develop satellite centres under CoEs to improve accessibility and outreach in underserved regions.
 - Enhance responsible utilization of funds to maximize impact and address disparities in fund utilization.
- There is a need for a **national registry for rare diseases to report** and clarify the list of rare diseases, as well as a centralized laboratory for the detection of rare diseases.
- Incentivize **domestic drug manufacturers** under the [Production-Linked Incentive Scheme](#) to encourage the production of affordable drugs.
- Implement the **Comprehensive Rare Disease Care (CRDC) model**, it aims to bridge the gap between patients and families suspected or affected by genetic etiology (gene abnormality).
 - The CRDC model establishes a technical and administrative roadmap for hospitals.
- Ensure affordable access to rare disease medicines, reduce tax on commercially available drugs, expanding access for patients.

UPSC Civil Services Examination, Previous Year Questions (PYQs)

Mains

Q. Appropriate local community-level healthcare intervention is a prerequisite to achieve 'Health for All' in India. Explain. (2018)