

Rare Diseases [UPSC Notes]

The Indian Government notified the National Policy for Rare Diseases, 2021 in March 2021. Some groups have expressed dissatisfaction over the policy. Read on to know why.

The topic of rare diseases comes under the health segment of the [UPSC syllabus](#).

Rare Diseases

A rare disease is a disease that affects a smaller percentage of the population compared to other diseases. Its prevalence is very low.

- There is no universally accepted definition of a rare disease.
- The definition accepted in the United States is that it is a disease that afflicts fewer than 2,00,000 people.
- This definition is also accepted by the National Organisation for Rare Disorders (NORD) in India.
- Rare diseases came to be known as orphan diseases because pharmaceutical companies were not ready to adopt them and develop drugs for them because of the low prevalence.
- That is why the drugs used to treat or manage rare diseases are known as [orphan drugs](#).
- Please note that every country might have a different definition of what can be called a rare disease. In the European Union, a rare disease is one that affects less than 1 person out of 2000 people.
- The [World Health Organisation](#) defines a rare disease as an often debilitating lifelong disease or disorder condition with a prevalence of 1 or less, per 1000 population.
- It is estimated that there are about 7000 rare diseases.
- Most of the rare diseases (about 80%) are genetic and hence affects a large number of children.
- Some rare diseases are not inherited such as some rare cancers, some autoimmune diseases, infectious tropical diseases, etc.
- Unfortunately, there are no treatments for many rare diseases.

Rare Diseases in India

In India, there are 56-72 million people affected by rare diseases.

- About 450 rare diseases have been recorded in India. Some of the common rare disease affecting people in India are auto-immune diseases, Gaucher's disease, Haemophilia, Sickle Cell Anaemia, Thalassemia, Primary Immuno-deficiency in children, Lysosomal storage disorders such as Pompe disease, Cystic Fibrosis, Hirschsprung disease, certain kinds of muscular dystrophies, Hemangiomas, Kyasanur Forest disease, etc.
- Identification and treatment of rare diseases is a challenge because of the general lack of awareness. Also, the field is a complex and heterogeneous one and one where research is still in the nascent stage. For many rare diseases, there are no diagnostic tests and even if there are, they are largely unavailable.

Why are there so many rare diseases in India?

- According to some experts, there are higher incidences of rare diseases in India because of the practice of consanguineous marriages in some communities. The most common rare diseases in India are Lysosomal Storage Disorders (LSD).

- Rare diseases make lives difficult not only because treatments are not defined for many of them, but also because of the fact that many of them cause a huge financial burden on patients' families. On average, the cost of treatment is exorbitant ranging from Rs 40 lakh to 1.60 crore per year.

National Policy for Treatment of Rare Diseases, 2021

The government notified the National Policy for Treatment of Rare Diseases, 2021 recently. Although the policy was formulated in 2017, it was not notified until now.

- It is proposed to have a national registry for a database of rare diseases.
- The document classifies rare diseases into three groups:
 - Group 1 has disorders controllable by one-time curative treatment, including osteopetrosis and Fanconi anaemia.
 - Group 2 has diseases requiring long-term or lifelong treatment with a relatively lower cost of treatment and benefit has been documented in literature, including galactosemia, severe food protein allergy, and homocystinuria.
 - Group 3 has diseases for which definitive treatment is available, but challenges are to make optimal patient selection for benefit, and very high cost and lifelong therapy, covering diseases such as spinal muscular atrophy (SMA), Pompe disease, and Hunter syndrome.
- The government has decided to provide Rs.20 lakh to poor patients and patients covered under the [Ayushman Bharat](#) for Group 1 diseases.
- For diseases classified under Group 3, which require life-long expensive treatments, the government would create a digital platform to bring together Centres for Excellence, patients undergoing treatment and corporate donors or prospective voluntary individuals who could help fund treatment.
 - The government has identified eight Centres of Excellence for the treatment of rare diseases.
- The state governments would be asked to undertake treatment of diseases covered under Group 2, which largely include disorders managed with special dietary formulae or food for special medical purposes (FSMP) and disorders that are manageable to other forms of therapy.

One of the criticisms of the National Policy for Treatment of Rare Diseases, 2021 is that Rs.20 lakh seems to be quite less and does not cover the actual costs of treatment, according to many caregivers of patients with rare diseases. Another criticism is that more centres of excellence should be started to cater to the needs of people suffering from rare diseases.
