National Policy on Rare Diseases

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What are rare diseases?

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. It is estimated that globally around 6000 to 8000 rare diseases exist with new rare diseases being reported in the medical literature regularly.

WHO defines a rare disease as often debilitating lifelong disease or disorder condition with a prevalence of 1 or less, per 1000 population. However, different countries have their own definitions to suit their specific requirements and in the context of their own population, the health care system and resources.

Rare Diseases as a public health issue in India

- The field of rare diseases is complex, heterogeneous, continuously evolving and suffers from a deficit of medical and scientific knowledge. So far about 450 rare diseases have been recorded in India. Globally as well as in India, rare diseases pose a significant challenge to public health systems in terms of – difficulty in collecting epidemiological data, which in turn impedes arriving at burden of diseases and cost estimations, difficulty in research and development, making correct and timely diagnosis, complex tertiary level management involving long term care and rehabilitation and unavailability and prohibitive cost of treatment.
- So far only about 450 diseases have been recorded in India from tertiary care hospitals that are globally

considered as rare diseases.

- **The most commonly reported diseases include** Haemophilia, Thalassemia, Sickle-cell Anaemia and Primary Immuno Deficiency in children, auto-immune diseases, Lysosomal storage disorders such as Pompe disease, Hirschsprung disease, Gaucher's disease, Cystic Fibrosis, Hemangiomas and certain forms of muscular dystrophies

Committees appointed by the government to make recommendations for framing a policy on rare diseases

Pursuant to the orders of the Hon'ble Court, the GOI had constituted committees with the objective to make suggestions towards the framing of a 'national policy on the treatment of rare diseases'. Similarly, the Government of NCT of Delhi had also appointed a high powered interdisciplinary committee on rare diseases. **The various committees that were appointed were:**

- The committee under Professor V.K. Paul, Head, Department of Pediatrics, AIIMS, New Delhi– 'Prioritisation of Therapy for Rare Genetic Disorders'
- Sub-committee on rare diseases in India, under Prof. I.C. Verma, Director, Institute of Medical Genetics Genomics, Sir Ganga Ram Hospital – 'Guidelines for Therapy and Management'
- A high powered interdisciplinary Committee on rare diseases under the Chairpersonship of Dr. Deepak K. Tempe, Dean, Maulana Azad Medical College(MAMC), New Delhi

Policy Direction

- The policy **aims at lowering the incidence and prevalence of rare diseases based on an integrated and comprehensive preventive strategy** encompassing awareness generation and screening programmes to prevent births of children with rare diseases, and, within the constraints on resources and competing health care priorities, enable access to affordable health care to patients of rare diseases which are amenable to one-time treatment.

- Considering the limited data available on rare diseases, and in the light of competing health priorities, the focus shall be on the **prevention of rare diseases as a priority** for all the three groups of rare diseases identified by Experts.
- Public Health and hospitals being a State subject, the Central Government shall encourage & support the States in their endeavour towards screening and prevention of rare diseases.
- In addition, the **following initiatives shall be taken for patients of Rare Diseases:**
    1. **Financial support up to Rs. 15 lakh under the Umbrella Scheme of Rashtriya Arogaya Nidhi** shall be provided by the Central Government for treatment, of those rare diseases that require a one-time treatment (diseases listed under Group I). **Beneficiaries for such financial assistance would not be limited to BPL families, but extended to 40% of the population** who are eligible **as per norms of Pradhan Mantri Jan Arogya Yojana**, for their treatment in Government tertiary hospitals only.
    2. State Governments can consider supporting patients of such rare diseases that can be managed with **special diets or hormonal supplements** or other relatively low-cost interventions (Diseases listed under Group II).
    3. Keeping in view the resource constraints, and a compelling need to prioritize the available resources to get maximum health gains for the community/population, the Government will endeavour to **create alternate funding mechanism through setting up a digital platform for voluntary individual and corporate donors** to

contribute to the treatment cost of patients of rare diseases.

4. The Government will have a **National Registry for Rare Diseases at ICMR with the objective of creating a database** of various rare diseases. Steps have already been taken in this direction by ICMR. Over a period of time, the registry is expected to yield enough information to be able to arrive at a definition of rare diseases best suited to the Country
5. State Governments will be supported for undertaking **screening & diagnosis amongst high-risk pregnant women as well as amongst suspected cases of rare diseases** reporting to Government facilities as a result of the awareness building activities
6. The Government shall notify selected Centres of Excellence at premier Government hospitals for comprehensive management of rare diseases.