

**GOVERNMENT OF INDIA  
MINISTRY OF HEALTH AND FAMILY WELFARE  
DEPARTMENT OF HEALTH AND FAMILY WELFARE**

**LOK SABHA  
UNSTARRED QUESTION NO. 936  
TO BE ANSWERED ON 07<sup>TH</sup> FEBRUARY, 2020**

**NATIONAL POLICY ON RARE DISEASES**

**936. SHRIMATI MEENAKASHI LEKHI:  
SHRI SYED IMTIAZ JALEEL:  
SHRI SANJAY SADASHIV RAO MANDLIK:  
SHRI SHRIRANG APPA BARNE:  
SHRI ASADUDDIN OWAISI:  
SHRI BIDYUT BARAN MAHATO:  
SHRI GAJANAN KIRTIKAR:  
SHRI SUDHEER GUPTA:  
SHRI M. SELVARAJ:**

Will the Minister of **HEALTH AND FAMILY WELFARE** be pleased to state:

(a) whether the Government is working on a Draft National Policy for treatment of Rare Diseases in the country and invited views of all the stakeholders on the said policy, if so, the details thereof along with the aims and objective and salient features thereon and the timeline for its finalization, notification and implementation;

(b) whether the policy provides financial assistance to children only till the age of 2 years for lysosomal disorders which is likely to be inadequate for poor and lower income group families, if so, the details thereof and the manner in which the policy would ensure safety nets for lower income group families, treatment of patients for rare diseases with no approved treatment or requiring import of drugs;

(c) whether there is scarcity of funds for rare diseases and a separate fund is proposed for treatment of poor people, if so, the details thereof along with Centre and States share therein and the funds made available for the treatment of rare diseases during each of the last three years and the current year;

(d) whether the Government does not maintain any comprehensive data on prevalence of rare diseases and the associated morbidity/mortality, if so, the reasons therefor; and

(e) whether ICMR has been proposed to create a data base to provide Rs. 15 lakh assistance for treatment of rare diseases under different schemes of the Government, if so, the details thereof?

**ANSWER**

**THE MINISTER OF STATE IN THE MINISTRY OF HEALTH AND  
FAMILY WELFARE**

**(SHRI ASHWINI KUMAR CHOUBEY)**

(a) to (e): A draft National Policy for Rare Diseases has been finalized and placed on the website of the Ministry of Health and Family Welfare with a view to elicit comments / views of the stakeholders, including the States/UTs and the general public by 15-02-2020.

Contd.....

The draft policy provides for lowering the incidence of rare diseases based on an integrated preventive strategy encompassing awareness generation and screening programmes 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.

The draft policy has noted that number of persons suffering from diseases considered rare globally, is lacking in India and accordingly provides that for the purpose of the policy the term rare diseases shall construe three group of disorders (**Annexure**) identified and categorised by experts based on their clinical experience. Considering the limited data available on rare diseases, and in the light of competing health priorities, the focus of the draft policy is on prevention of rare diseases as a priority for all the three groups of rare diseases identified by experts.

The draft policy further provides for:

- i. Financial support upto Rs. 15 lakh under the Umbrella Scheme of Rashtriya Arogya Nidhi by the Central Government for treatment of those rare diseases that require one-time treatment (diseases listed under Group I of **Annexure**). 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.
- ii. 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 of **Annexure**).
- iii. Keeping in view the resource constraints, and a compelling need to prioritize the available resources to get maximum health gains for the community/population, policy also provides for endeavour by the Government 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.
- iv. Creating a National Registry for rare diseases at ICMR with the objective of preparing a database of various rare diseases, for which action has already been initiated by ICMR.

In the meanwhile, under the Umbrella Scheme of Rashtriya Arogya Nidhi, a component of rare disease has been included w.e.f. 01.01.2019 for providing one-time financial assistance upto Rs.15 lakh to patients belonging to families living below threshold poverty line for treatment of identified Rare Diseases amenable to one-time treatment in Government hospitals. An amount of Rs.100 crore has been allocated for Rare Disease component of Umbrella Scheme of Rashtriya Arogya Nidhi during 2019-20.

.....

**List of identified rare diseases covered for treatment under the Umbrella Scheme of RashtriyaArogyaNidhi**

Group 1: Disorders amenable to one time curative treatment:

- a) Disorders amenable to treatment with Hematopoietic Stem Cell Transplantation (HSCT) –
  - a. Such Lysosomal Storage Disorders (LSDs) for which Enzyme replacement Therapy (ERT) is presently not available and severe form of Mucopolysaccharoidosis (MPS) type I within first 2 years of age.
  - b. Adrenoleukodystrophy (early stages), before the onset of hard neurological signs.
  - c. Immune deficiency disorders like Severe Combined Immunodeficiency (SCID), Chronic Granulomatous disease, Wiskot Aldrich Syndrome, etc
  - d. Osteopetrosis
  - e. Fanconi Anemia
  - f. Others if any to be decided on case to case basis by a technical committee
- b) Disorders amenable to organ transplantation
  - i) Liver Transplantation -Metabolic Liver diseases:
    - a. Tyrosinemia,
    - b. Glycogen storage disorders (GSD) I, III and IV due to poor metabolic control, multiple liver adenomas, or high risk for Hepatocellular carcinoma or evidence of substantial cirrhosis or liver dysfunction or progressive liver failure,
    - c. MSUD (Maple Syrup Urine Disease),
    - d. Urea cycle disorders,
    - e. Organic acidemias
  - ii) Renal Transplantation-
    - a. Fabry's disease
    - b. Autosomal recessive Polycystic Kidney Disease (ARPKD),
    - c. Autosomal dominant Polycystic Kidney Disease (ADPKD) etc
  - iii) Patients requiring combined liver and kidney transplants can also be considered if the same ceiling of funds is maintained. ( Rarely Methyl Malonic aciduria may require combined liver & Kidney transplant) etc

Group 2: Diseases requiring long term / lifelong treatment having relatively lower cost of treatment and benefit has been documented in literature and annual or more frequent surveillance is required:

- a) Disorders managed with special dietary formulae or Food for special medical purposes (FSMP)
  - i) Phenylketonuria (PKU)
  - ii) Non-PKU hyperphenylalaninemia conditions
  - iii) Maple Syrup Urine Disease (MSUD)
  - iv) Tyrosinemia type 1 and 2
  - v) Homocystinuria
  - vi) Urea Cycle Enzyme defects
  - vii) Glutaric Aciduria type 1 and 2
  - viii) Methyl Malonic Acidemia
  - ix) Propionic Acidemia

- x) Isovaleric Acidemia
- xi) Leucine sensitive hypoglycemia
- xii) Galactosemia
- xiii) Glucose galactose malabsorption
- xiv) Severe Food protein allergy

b) Disorders that are amenable to other forms of therapy (hormone/ specific drugs)

- i) NTBC for Tyrosinemia Type 1
- ii) Osteogenesis Imperfecta – Bisphosphonates therapy
- iii) Growth Hormone therapy for proven GH deficiency , Prader Willi Syndrome and Turner syndrome, others (to be decided on case to case basis by technical committee)
- iv) Cystic Fibrosis- Pancreatic enzyme supplement
- v) Primary Immune deficiency disorders -Intravenous immunoglobulin therapy (IVIG) replacement eg. X-linked agammaglobulinemia etc.
- vi) Sodium Benzoate, arginine, ,citrulline ,phenylacetate (Urea Cycle disorders), carbaglu, Megavitamin therapy (Organic acidemias, mitochondrial disorders)
- vii) Others - Hemin (Panhematin) for Acute intermittent Porphyria, High dose Hydroxocobalamin injections (30mg/ml formulation – not available in India and hence expensive if imported)
- viii) Others (if any) to be decided on case-to-case basis, by a technical committee.

Group 3: Diseases for which definitive treatment is available but challenges are to make optimal patient selection for benefit, very high cost and lifelong therapy.

3a) Based on the literature sufficient evidence for good long-term outcomes exists for the following disorders

1. Gaucher Disease (Type I & III {without significant neurological impairment})
2. Hurler Syndrome [Mucopolysaccharidosis (MPS) Type I] (attenuated forms)
3. Hunter syndrome (MPS II) (attenuated form)
4. Pompe Disease diagnosed early (Both infantile & late onset)
5. Fabry Disease diagnosed before significant end organ damage.
6. Spinal Muscular Atrophy
7. MPS IVA
8. MPS VI

3b) For the following disorders for which the cost of treatment is very high and either long term follow up literature is awaited or has been done on small number of patients

1. Wolman Disease
2. Hypophosphatasia
3. Neuronal ceroid lipofuscinosis
4. Cystic Fibrosis
5. Duchenne Muscular Dystrophy