

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

**LOK SABHA
UNSTARRED QUESTION NO.728
TO BE ANSWERED ON 23RD JULY, 2021**

RARE DISEASES POLICY

**728. SHRI PRATHAP SIMHA:
SHRI ANNASAHEB SHANKAR JOLLE:
SHRI D.M. KATHIR ANAND:
DR. UMESH G. JADHAV:
SHRI V.K.SREEKANDAN:
SHRI B.Y. RAGHAVENDRA:
SHRI L.S. TEJASVI SURYA:**

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

- (a) whether the Government is considering to finalise and notify the National Health Policy for Rare diseases, if so, the details thereof;
- (b) the objective and current status of the same;
- (c) the total number of suggestions received for the draft policy, State/UT-wise;
- (d) the number of rare diseases recognized by the Government and its proposal to assist the people suffering from such diseases particularly the low income groups;
- (e) whether the Government proposes to allocate increased budget for this policy and also considering to provide assistance to poor patients through Ayushman Bharat, if so, the details thereof and the feasibility of the assistance likely to be provided through Ayushman Bharat; and
- (f) whether Government is considering to set up a National Consortium for Research and Development and Therapeutics for Rare Diseases, if so, the details thereof?

**ANSWER
THE MINISTER OF STATE IN THE MINISTRY OF HEALTH AND
FAMILY WELFARE
(DR. BHARATI PRAVIN PAWAR)**

(a): National Policy for Rare Diseases, 2021(the Policy) has been finalized and put in public domain. The Policy can be accessed at website-

<https://main.mohfw.gov.in/documents/policy>.

(b): The policy aims at lowering the incidence and prevalence of rare diseases based on an integrated and comprehensive preventive strategy encompassing awareness generation, premarital, post-marital, pre-conception and post-conception screening and counselling 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.

(c): The total number of suggestions received for the draft policy is 279. State/UT-wise data is not maintained.

(d): The number of rare diseases categorized as per the Policy are as per Annexure-A.

Initiatives for treatment support for patients of rare diseases under the Policy are as follows:-

i. Financial support upto Rs. 20 lakh under the Umbrella Scheme of Rashtriya Arogya Nidhi shall be provided by the Central Government for treatment, of those rare diseases that require a one-time treatment (diseases listed under Group 1). Beneficiaries for such financial assistance would not be limited to BPL families, but extended to about 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 2).

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, 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.

iv. Voluntary crowd-funding for treatment: Keeping in view the resource constraint and competing health priorities, it will be difficult for the Government to fully finance treatment of high cost rare diseases. The gap can however be filled by creating a digital platform for bringing together notified hospitals where such patients are receiving treatment or come for treatment, on the one hand, and prospective individual or corporate donors willing to support treatment of such patients. The notified hospitals will share information relating to the patients, diseases from which they are suffering, estimated cost of treatment and details of bank accounts for donation/contribution through online system. Donors will be able to view the details of patients and donate funds to a particular hospital. This will enable donors from various sections of the society to donate funds, which will be utilized for treatment of patients suffering from rare diseases, especially those under Group 3. Conferences will be organised with corporate sector companies to motivate them to donate generously through digital platform. Ministry of Corporate Affairs will be requested to encourage PSUs and corporate houses to contribute as per the Companies Act as well as the provisions of the Companies (Corporate Social Responsibility Policy) Rules, 2014 (CSR Rules). Promoting health care including preventive health care is included in the list in the Schedule for CSR activities.

Treatment cost of the patient will be first charge on this fund. Any leftover fund after meeting treatment cost can be utilized for research purpose also.

(e): At present financial assistance to poor patients, living below threshold poverty line and also to the population, who are eligible as per norms of Pradhan Mantri Jan Arogya Yojana under Ayushman Bharat, suffering from specified rare diseases for their treatment at Government Hospitals or Institutes having super specialty facilities / Government tertiary hospitals is being provided under the Umbrella Scheme of Rashtriya Arogya Nidhi (RAN). The budget allocation for the current financial year 2021-2022 for rare diseases is Rupees 25 Crore.

(f): National Policy for Rare Diseases, 2021 provides for National Consortium for Research and Development on therapeutics for Rare Diseases with an expanded mandate to include research & development, technology transfer and indigenization of therapeutics for rare diseases. It will be convened by Department of Health Research (DHR) with ICMR as a member.

India faces the limitation of lack of epidemiological data to be able to define rare diseases in terms of prevalence or prevalence rate, which has been used by other countries. To overcome this, a hospital based National Registry for Rare Diseases has been initiated by ICMR by involving centers across the country that are involved in diagnosis and management of Rare Diseases. This will yield much needed epidemiological data for rare diseases.

Till the time such data is available, for the purpose of National Policy for Rare Diseases, 2021, the following groups of disorders have been identified and categorized by experts based on their clinical experience:

Group 1: Disorders amenable to one-time curative treatment:

a) Disorders amenable to treatment with Hematopoietic Stem Cell Transplantation (HSCT) –

- i. 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.*
- ii. Adrenoleukodystrophy (early stages), before the onset of hard neurological signs.*
- iii. Immune deficiency disorders like Severe Combined Immunodeficiency (SCID), Chronic Granulomatous disease, Wiskot Aldrich Syndrome etc.*
- iv. Osteopetrosis*
- v. Fanconi Anemia*

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 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 Malonicaciduria 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, Turner syndrome and Noonan syndrome.**
- iv) Cystic Fibrosis- Pancreatic enzyme supplement**
- v) Primary Immune deficiency disorders -Intravenous immunoglobulin and sub cutaneous 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) Large neutral aminoacids, mitochondrial cocktail therapy, Sapropterin and other such molecules of proven clinical management in a subset of disorders

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 [Mucopolysaccharisosis (MPS) Type I] (attenuated forms)*
- 3. Hunter syndrome (MPS II) (attenuated form)*
- 4. Pompe Disease (Both infantile & late onsetdiagnosed early before development of complications)*
- 5. Fabry Disease diagnosed before significant end organ damage.*
- 6. MPS IVA before development of disease complications.*
- 7. MPS VI before development of disease complications.*
- 8. DNAase for Cystic Fibrosis.*

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. Cystic Fibrosis (Potentiators)*
- 2. Duchenne Muscular Dystrophy (Antesense oligoneucletides, PTC)*
- 3. Spinal Muscular Atrophy (Antisense oligonucleotides both intravenous & oral & gene therapy)*
- 4. Wolman Disease*
- 5. Hypophosphatasia*
- 6. Neuronal ceroid lipofuschinosis*

The list of diseases under Group 1, Group 2 and Group 3 are not exhaustive and will be reviewed periodically based on updated scientific data by the Technical Committee.
