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

LOK SABHA
UNSTARRED QUESTION NO. 3062
TO BE ANSWERED ON 9TH AUGUST, 2024

SUPPORT TO PATIENTS OF RARE DISEASES

3062. SHRI ARUN BHARTI :

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

(a) the key features of the National Policy for Rare Diseases;
(b) the total number of rare diseases that have been identified under this policy and support provided to patients;
(c) the budget allocation made and utilized for the treatment of rare diseases during the last three years; and
(d) the number of patients who have benefited from the Policy for Rare Diseases particularly in Bihar?

ANSWER
THE MINISTER OF STATE IN THE MINISTRY OF HEALTH AND FAMILY WELFARE
(SMT. ANUPRIYA PATEL)

(a) Ministry of Health & Family Welfare launched National Policy for Rare Diseases (NPRD) in March 2021. The key features of NPRD, 2021 are as under:

- The rare diseases have been identified and categorized into 3 groups as below:
  Group 1: Disorders amenable to one-time curative treatment.
  Group 2: Diseases requiring long term/lifelong treatment with relatively lower cost of treatment.
  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.
- 12 (Twelve) Centres of Excellence (CoEs) have been identified so far, which are premier Government tertiary hospitals with facilities for diagnosis, prevention and treatment of rare diseases. List of Centres of Excellence (CoEs) is at **Annexure I**.
- In order to receive financial assistance for treatment of rare disease, the patient may approach nearby or any Centre of Excellence to get registered.
- Ministry of Health and Family Welfare has obtained exemption from Department of Expenditure on Goods & Services Tax (GST) and Basic Customs Duty on drugs imported for Rare Diseases for individual use and through CoE.

- As envisaged in the policy, Department of Health Research has established the National Consortium for Research and Development on Therapeutics for Rare Diseases (NCRDTRD) for streamlining the research activities for rare diseases.

(b) Currently, 63 rare diseases are included under National Policy for Rare Diseases on recommendation of Central Technical Committee for Rare Diseases(CTCRD). The list of rare diseases is at **Annexure II.** Financial support of upto Rs. 50 lakhs per patient is provided for the treatment at the notified Centres of Excellence(CoEs) for Rare Diseases.

(c) Budget allocation made and utilized for the treatment of rare diseases during the last three years including the current financial year is as under:

(Rs. in lakhs)

Financial Year	**Grant-in Aid General for Treatment**			**Grant for Creation of Capital Assets**		
	Budget Estimate	**Revised Estimate**	**Expenditure till date**	**Budget Estimate**	**Revised Estimate**	**Expenditure till date**
2022-23	2500	3500	3499	-	-	-
2023-24	9284	7400	7400	5000	3500	3500
2024-25	8241	-	2420	2000	-	-

(d) Since the launch of the policy, a total number of one thousand one hundred and eighteen(1118) patients have benefited under NPRD. State-wise data is not available as the patient can approach any CoE across the country as per their convenience.

Annexure I referred to in reply to part (a) of Lok Sabha Unstarred Q.No. 3062 to be answered on 9.8.2024

List of Centres of Excellence (CoEs)

1. All India Institute of Medical Sciences, New Delhi
2. Maulana Azad Medical College, New Delhi
3. Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow
4. Post Graduate Institute of Medical Education and Research, Chandigarh
5. Centre for DNA Fingerprinting & Diagnostics with Nizam's Institute of Medical Sciences, Hyderabad
6. King Edward Medical Hospital, Mumbai
7. Institute of Post-Graduate Medical Education and Research, Kolkata
8. Center for Human Genetics(CHG) with Indira Gandhi Hospital, Bengaluru
9. Institute of Child Health and Hospital for Children (ICH & HC), Chennai
10. All India Institute of Medical Sciences (AIIMS), Jodhpur
11. Sree Avittam Thirunal Hospital (SAT), Government Medical College, Thiruvananthapuram
12. All India Institute of Medical Sciences, Bhopal

Annexure II referred to in reply to part (b) of Lok Sabha Unstarred Q.No. 3062 to be answered on 9.8.2024

List of Rare Diseases as per National Policy of Rare Diseases, 2021

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

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

1. 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.
2. Adrenoleukodystrophy (early stages), before the onset of hard neurological signs.
3. Immune deficiency disorders like Severe Combined Immunodeficiency (SCID), Chronic Granulomatous disease, Wiskot Aldrich Syndrome etc.
4. Osteopetrosis
5. Fanconi Anemia

(b) Disorders amenable to organ transplantation

1) Liver Transplantation -Metabolic Liver diseases:

(i) Tyrosinemia,

(ii) 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,

(iii) MSUD (Maple Syrup Urine Disease),

(iv) Urea cycle disorders,

(v) Organic acidemias.

2) Renal Transplantation-

(i) Fabry disease

(ii) Autosomal recessive Polycystic Kidney Disease (ARPKD),

(iii) Autosomal dominant Polycystic Kidney Disease (ADPKD) etc.

3) 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.

Newly added diseases

1. Laron Syndrome
2. Glanzmann Thrombasthenia Diseases
3. Congenital Hyperinsulinemic Hypoglycemia (CHI)
4. Familial Homozygous Hypercholesterolemia
5. Mannosidosis
6. XY Disorder of Sex Development due to 5 alpha reductase deficiency, partial androgen insensitivity syndrome
7. Primary Hyperoxaluria- Type 1

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)

1. Phenylketonuria (PKU)
2. Non-PKU hyperphenylalaninemia conditions
3. Maple Syrup Urine Disease (MSUD)
4. Tyrosinemia type 1 and 2
5. Homocystinuria
6. Urea Cycle Enzyme defects
7. Glutaric Aciduria type 1 and 2
8. Methyl Malonic Acidemia
9. Propionic Acidemia
10. Isovaleric Acidemia
11. Leucine sensitive hypoglycemia
12. Galactosemia
13. Glucose galactose malabsorption
14. Severe Food protein allergy

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

1. NTBC for Tyrosinemia Type 1

2. Osteogenesis Imperfecta – Bisphosphonates therapy
3. Growth Hormone therapy for proven GH deficiency, Prader Willi Syndrome, Turner syndrome and Noonan syndrome.
4. Cystic Fibrosis- Pancreatic enzyme supplement
5. Primary Immune deficiency disorders -Intravenous immunoglobulin and sub cutaneous therapy (IVIG) replacement eg. X-linked agammablobulinemia etc.
6. Sodium Benzoate, arginine, citrulline, phenylacetate (Urea Cycle disorders), carbaglu, Megavitamin therapy (Organic acidemias, mitochondrial disorders)
7. Others - Hemin (Panhematin) for Acute Intermittent Porphyria, High dose Hydroxocobalamin injections (30mg/ml formulation – not available in India and hence expensive if imported)
8. Large neutral aminoacids, mitochondrial cocktail therapy, Sapropterin and other such molecules of proven clinical management in a subset of disorders
9. Wilson's disease
10. Congenital Adrenal Hyperplasia (CAH)
11. Neonatal Onset Multisystem Inflammatory Disease (NOMID)

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.

(a) 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 onset diagnosed 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.

(b) 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 (Antesensce oligoneucletides, PTC)
3. Spinal Muscular Atrophy (Antisense oligonucleotides both intravenous & oral & gene therapy)
4. Wolman Disease
5. Hypophosphatasia
6. Neuronal ceroid lipofuschinosis

Newly added diseases

1. Hypophosphatic Rickets
2. Atypical Hemolytic Uremic Syndrome (AHUS)
3. Cystinosis
4. Hereditary Angioedema