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NATIONAL POLICY ON RARE DISEASES

Relevant for: Developmental Issues | Topic: Health & Sanitation and related issues

Ministry of Health and Family Welfare

National Policy on Rare Diseases

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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.

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 goup of disorders 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 dradt policy is on prevention of rare diseases as a priority for all the three groups of rare diseases identified by experts.

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) – 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.Adrenoleukodystrophy (early stages), before the onset of hard neurological signs.Immune deficiency disorders like Severe Combined Immunodeficiency (SCID), Chronic Granulomatous disease, Wiskot Aldrich Syndrome,etcOsteopetrosisFanconi AnemiaOthers 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: Tyrosinemia,Glycogen storage disorders (GSD) I, III and IV due to poor metabolic control, multiple liver adenomas, or high risk for Hepatocellualr carcinoma or evidence of substantial cirrhosis or liver dysfunction or progressive liver failure,MSUD (Maple Syrup Urine Disease),Urea cycle disorders,Organic

acidemias

ii. Renal Transplantation- Fabry's diseaseAutosomal recessive Polycystic Kidney Disease (ARPKD),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 malabsorbtion
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 agammablobulinemia 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 [Mucopolysaccharisosis (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

The Minister of State (Health and Family Welfare), Sh Ashwini Kumar Choubey stated this in a written reply in the Lok Sabha here today.

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MV/LK

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