

List of Rare Diseases as per National Policy for Rare Diseases (NPRD) 2021

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)
- 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.

c) Other

- i. Familial Homozygous Hypercholesterolemia
- ii. Mannosidosis
- iii. XY Disorder of Sex Development due to 5 alpha reductase deficiency, partial androgen insensitivity syndrome
- iv. Primary Hyperoxaluria (only for transplants)
- v. Laron's Syndrome
- vi. Glanzmann Thrombasthenia Disease
- vii. Congenital Hyperinsulinemic Hypoglycemia (CHI)
- viii. Laron's Syndrome

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 I
- 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 subcutaneous therapy (IVIG) replacement eg. X-linked agammaglobulinemia etc.
- VI. Sodium Benzoate, arginine, citrulline, phenylacetate (Urea Cycle disorders), carbamglut, Megavitamin therapy (Organic acidemias, mitochondrial disorders).
- VII. Others - Hemin (Panhematin) for Acute Intermittent Porphyrin, 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.
- IX. Wilson's Disease
- X. Congenital Adrenal Hyperplasia (CAH)
- XI. 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.

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

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 (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 complication.
8. DNAase for Cystic Fibrosis.
9. Cystinosis.
10. Hereditary Angioedema.

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 (Antesence oligoneucleotides, PTC)
3. Spinal Muscular Atrophy (Antisense oligonucleotides both intravenous & oral & gene therapy).
4. Wolman Disease
5. Hypophosphatasia
6. Neuronal ceroid lipofuscinosis
7. Hypophosphatic Rickets
8. Atypical Hemolytic Uremic Syndrome (AHUS).