NATIONAL POLICY FOR RARE DISEASES

2020

Ministry of Health and Family Welfare

Government of India

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1. Background

Ministry of Health and family Welfare, Government of India formulated a National Policy for Treatment of Rare Diseases (NPTRD) in July, 2017. Implementation of the policy was, however, faced with certain challenges. A limiting factor in its implementation was bringing States on board and lack of clarity on how much Government could support in terms of tertiary care. Public Health and Hospitals is a State subject. Stakeholder consultation with the State Governments at the draft stage of formulation of the policy could not be done in an elaborate manner. When the policy was shared with State Governments, issues such as cost effectiveness of interventions for rare disease vis-à-vis other health priorities, the sharing of expenditure between Central and State Governments, flexibility to State Governments to accept the policy or change it according to their situation, were raised by some of the State Governments.

In the circumstances, though framed with best intent, the policy had implementation challenges and gaps, including the issue of cost effectiveness of supporting such health interventions for resources limited situation, which made it not feasible to implement. Given the challenges in implementing the policy, the need for wider consultation and recommendations, a decision was taken to reframe the National Policy for Treatment of Rare Diseases. An Expert Committee was constituted by Ministry of Health and Family Welfare in November, 2018 to review the NPTRD, 2017. The Terms and References of the Expert Committee are given below:

a. To review the national Policy for Treatment of rare Diseases, 2017 and to suggest amendments/changes as may be required.
b. To define rare diseases for India.
c. To draft National Policy for rare Diseases.
d. To suggest vision and strategy in country's context.

Pending reframing the policy, the earlier policy has been kept in abeyance vide a non-statutory Gazette Notification dated 18-12-2018, till the revised policy is issued or till further orders, whichever is earlier.

2. Rare Diseases: Issues & Challenges

The field of rare diseases is complex and heterogeneous. The landscape of rare diseases is constantly changing, as there are new rare diseases and conditions being identified and reported regularly in medical literature. Apart from a few rare diseases, where significant progress has been made, the field is still at a nascent stage. For a long time, doctors, researchers and policy makers were unaware of rare diseases and until very recently there was no real research or public health policy concerning issues related to the field. This poses formidable challenges in development of a comprehensive policy on rare diseases. Nevertheless, it is important to take steps, in the short as well as long term, with the objective of tackling rare diseases in a holistic and comprehensive manner.

2.1 The varying definitions of rare diseases

WHO defines rare disease as often debilitating lifelong disease or disorder condition with a prevalence of 1 or less, per 1000 population. However, different countries have their own definitions to suit their specific requirements and in context of their own population, health care system and resources. In the US, rare diseases are defined as a disease or condition that affects fewer than 200,000 patients in the country (6.4 in 10,000 people). EU defines rare diseases as a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 people. Japan identifies rare diseases as diseases with fewer than 50,000 prevalent cases (0.04%) in the country. A summary of the prevalence based definitions of rare diseases used in various countries is tabulated below:

Table 1: Definitions of Rare Disease in different countries

S No.	Country	Prevalence less than per 10,000 population
1	USA	6.4
2	Europe	5.0
3	Canada	5.0

3	Japan	4.0
4	South Korea	4.0
5	Australia	1.0
6	Taiwan	1.0

Source: The I.C. Verma Sub-Committee Report 'Guidelines for Therapy and Management'

The use of varying definitions and diverse terminology can result in confusion and inconsistencies and have implications for access to treatment and for research and development. According to a study[1,] that reviewed and analysed definitions across jurisdictions, most definitions, as discussed above, appear to consider disease prevalence, but other criteria also apply sometimes, such as - disease severity, whether the disease is life-threatening, whether there are alternative treatment options available, and whether it is heritable. The study found that relatively few definitions included qualifiers relating to disease severity and/or a lack of existing treatments, whereas most definitions included a prevalence threshold. The average prevalence thresholds used to define rare diseases ranges among different jurisdictions from 1 to 6 cases/10,000 people, with WHO recommending a prevalence less than 10/10,000 population for defining rare diseases. The study concluded that attempts at harmonising the differing definitions, should focus on standardizing objective criteria such as prevalence thresholds and avoid qualitative descriptors like severity of the disease.

However, it has been contested that disease prevalence alone may also not be an accurate basis for defining rare diseases, as it does not take into account changes in population over time. Hence, some have suggested that a more reliable approach to arriving at a definition could be based on the factors of – a) location - a disease which is uncommon in one country may be quite common in other parts of the world; b) levels of rarity - some diseases may be much more rare than other diseases which are

1. [1] *Richter, T., Nestler-Parr, S., Babela, R., Khan, Z. M., Tesoro, T., Molsen, E., & Hughes, D. A. (2015). Rare Disease Terminology and Definitions – A Systematic Global Review: Report of the ISPOR Rare Disease Special Interest Group [Electronic version]. Value in Health, 18, 906-914. Available at: https://www.ispor.org/raredisease-terms-definitions.pdf*

also uncommon; and c) study-ability - whether the prevalence of a disease lends itself to clinical trials and studies.

This underscores the need for further research to better understand the extent of the existing diversity of definitions for rare diseases and to examine the scope of arriving at a definition, which is best suited to conditions in India. It shall be done on a priority basis as soon as sufficient data is available. Steps have already been taken for creation of a registry of rare diseases in India by ICMR.

2.2 Diagnosis of rare diseases

` Early diagnosis of rare diseases is a challenge owing to multiple factors that include lack of awareness among primary care physicians, lack of adequate screening and diagnostic facilities etc.

Traditional genetic testing includes tests that can only address a few diseases. As a result, physicians most often provide their best guess on which tests are to be done. If the test is negative, further testing will be required using next generation sequencing based tests, or chromosomal microarray which are available, but expensive and time- consuming processes with interpretation and counselling issues at times.

There is a lack of awareness about rare diseases in general public as well as in the medical profession. Many doctors lack appropriate training and awareness to be able to correctly and timely diagnose and treat these conditions. According to a recent report[2], it takes patients in United States (US) an average of 7.6 years and patients in United Kingdom (UK) an average of 5.6 years to receive an accurate diagnosis, typically involving as many as eight physicians (four primary care and four specialists). In addition, two to three misdiagnoses are typical before arriving at a final diagnosis. Delay in diagnosis or a wrong diagnosis increases the suffering of the patients exponentially. There is an immediate need to create awareness among general public, patients & their families and doctors, training of doctors for better diagnosis, standardization of diagnostic modalities and development of newer diagnostic & therapeutic tools.

[2] *Rare Disease Impact Report: Insights from patients and the medical community available at: https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf*

2.3 Challenges in research and development

A fundamental challenge in research and development for the majority of rare diseases is that there is relatively little known about the pathophysiology or the natural history of these diseases. Rare diseases are difficult to research upon as the patient pool is very small and it often results in inadequate clinical experience. Therefore, the clinical explanation of rare diseases may be skewed or partial. The challenge becomes even greater as rare diseases are chronic in nature, where long term follow up is particularly important. As a result, rare diseases lack published data on long-term treatment outcomes and are often incompletely characterised.

This makes it necessary to explore international and regional collaborations for research, collaborations with the physicians who work on any rare disease and with patient groups and families dealing with the consequences of these disorders. This will help gain a better understanding of the pathophysiology of these diseases, and the therapeutic effects that would have a meaningful impact on the lives of patients. There is also a need to review and where possible modify, clinical trial norms keeping in mind the particular challenges in rare diseases, without compromising on the safety and quality of the drugs or diagnostic tools.

2.4 Challenges in treatment

2.4.1 Unavailability of treatment

Availability and access to medicines are important to reduce morbidity and mortality associated with rare diseases. Despite progress in recent years, effective or safe treatment is not available for most of the rare diseases. Hence, even when a correct diagnosis is made, there may not be an available therapy to treat the rare disease. There are between 7000 - 8000 rare diseases, but less than 5% have therapies available to treat them. About 95% rare diseases have no approved treatment[3] and less than 1 in 10 patients receive disease specific treatment. Where drugs are available, they are prohibitively expensive, placing immense strain on resources.

[3] https://www.thelancet.com/journals/landia/article/PIIS2213-8587(19)30006-3/fulltext

2.4.2 Prohibitive cost of treatment

As the number of persons suffering from individual rare diseases is small, they do not constitute a significant market for drug manufacturers to develop and bring to market drugs for them. For this reason, rare diseases are also called 'orphan diseases' and drugs to treat them are called "orphan drugs". Where, they do make drugs to treat rare diseases, the prices are extremely high apparently to recoup the cost of research and development. At present very few pharmaceutical companies are manufacturing drugs for rare diseases globally and there are no domestic manufacturers in India. Due to the high cost, the government has not been able to provide these drugs for free. It is estimated that for a child weighing 10 kg, the annual cost of treatment for some rare diseases, may vary from Rupees 10 Lakhs to more than 1 crore per year with treatment being lifelong and drug dose and cost increasing with age.

Countries have dealt with this unique problem of high cost through various means that were suited to their local needs. Instruments like the Orphan Drug Act (ODA) in US & Canada, provide incentives to drug manufacturers to encourage them to manufacture drugs for rare diseases. The economic incentives & safeguards offered under the Act ensure benefits to the local patients. However, the exorbitant prices of drugs for rare diseases has led to concerns even in the developed countries about maintaining sustainability of the rare diseases funding/reimbursement programmes. The exorbitant prices have led to calls for transparency in setting prices of drugs and for price control and have even prompted scrutiny and congressional inquiries.
Considering the above, Government shall promote R &D for improving availability of affordable therapies for rare diseases with adequate safeguards in place.

3. The Indian Scenario

Data on how many people suffer from different diseases that are considered rare globally, is lacking in India. The cases identified so far have been diagnosed at tertiary hospitals. The lack of epidemiological data on incidence and prevalence of rare diseases impedes understanding of the extent of the burden of rare diseases and development of a definition. It also hampers efforts to arrive at correct estimation of the number of persons suffering from these diseases and describe their associated morbidity and mortality. In

such a scenario, the economic burden of most rare diseases is unknown and cannot be adequately estimated from the existing data sets.

Although extremely challenging, considering the complexity of various diseases and the difficulty in diagnosis, there is a clear need to undertake systematic epidemiological studies to ascertain the number of people suffering from rare diseases in India.

So far only about 450 diseases have been recorded in India from tertiary care hospitals that are globally considered as rare diseases. The most commonly reported diseases include Haemophilia, Thalassemia, Sickle-cell Anaemia and Primary Immuno Deficiency in children, auto-immune diseases, Lysosomal storage disorders such as Pompe disease, Hirschsprung disease, Gaucher's disease, Cystic Fibrosis, Hemangiomas and certain forms of muscular dystrophies

4. Experiences from other countries:

While preparing the policy on rare diseases for India, policies of other countries have been reviewed. In United States of America, development of drugs for rare disease is sought to be encouraged through the Orphan Drugs Act, which incentivises industry by way of market exclusivity, grants to researchers and tax incentives on expenditure incurred during evaluation of drugs for their therapeutic potential. However critics have pointed out that pharmaceutical companies have taken advantage of this arrangement and 'gamed the system' to maximise profits. The European Joint Programme on Rare Disease mostly focuses on research. National Health Service (NHS) England, for example, provides that the treatment for Spinal Muscular Atrophy (SMA) will be made available to the youngest and most severely-affected (SMA Type 1) patients immediately by Biogen (The pharmaceutical company that manufactures treatment for SMA), with NHS England offering funding on National Institute for Health and Care Excellence (NICE's) publication of final guidance. In Singapore, a fund - Rare Disease Fund – has been created to fund five medicines to treat three rare disease conditions. In Malaysia and Australia subsidised access for eligible patients is provided to expensive and lifesaving drugs.

5. Need to balance competing priorities of public health in resource constrained settings

Rare diseases place a major economic burden on any Country and especially in resource-constrained settings. The financial capacity to support exorbitant cost of treatment, is an important consideration in public health policy development with reference to treatment for rare diseases. In resource-constrained settings, it is pertinent to balance competing interests of public health for achieving optimal outcome for the resources allocated. As resources are limited and have multiple uses, the policy makers have to make choice of prioritizing certain set of interventions over others- the appropriate choice is then to support those interventions that would provide more number of healthy life years for given sum of money while simultaneously looking at the equity i.e. interventions that benefit poor who cannot afford healthcare are prioritised. Thus, interventions that address health problems of a much larger number of persons by allocating a relatively smaller amount are prioritized over others such as funding treatment of rare diseases where much greater resources will be required for addressing health problems of a far smaller number of persons.

Hence any policy on rare diseases needs to be informed by the available scarce resources and the need for their utmost judicious utilization for maximizing the overall health outcomes for the whole of society measured in terms of increase in terms of healthy life years.

6. Definition of Rare Diseases:

6.1 There is no universal or standard definition of rare disease. A disease that occurs infrequently is generally considered a rare disease, and it has been defined by different countries in terms of prevalence – either in absolute terms or in terms of prevalence per 10,000 population. A country defines a rare disease most appropriate in the context of its own population, health care system and resources.

6.2 As mentioned above, 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 National Registry for Rare Diseases shall be initiated by ICMR by involving centers across the Country that are involved in diagnosis and management of Rare Diseases. Steps have already been taken in this direction by ICMR. This will yield much needed epidemiological data for arriving at a prevalence based definition of rare diseases. In the absence of epidemiological data on

diseases considered as rare in other countries, it is not possible to prescribe threshold prevalence rates to define a disease condition as rare.

Till the time such data is available and the Country arrives at a definition of a rare disease based on prevalence data, the term rare diseases, for the purpose of this policy, shall construe the following groups of disorders 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) –

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 Hepatocellualr 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 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

7. Policy Direction

The policy aims at lowering the incidence and prevalence of rare diseases based on an integrated and comprehensive preventive strategy encompassing awareness generation and screening 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 which are amenable to one-time treatment.

Considering the limited data available on rare diseases, and in the light of competing health priorities, the focus shall be on prevention of rare diseases as a priority for all the three groups of rare diseases identified by Experts. Public Health and hospitals being a State subject, the Central Government shall encourage & support the States in their endeavour towards screening and prevention of rare diseases.

In addition, the following initiatives shall be taken for patients of Rare Diseases:

i. Financial support upto Rs. 15 lakh under the Umbrella Scheme of Rashtriya Arogaya Nidhi shall be provided by the Central Government for treatment, of those rare diseases that require a one-time treatment (diseases listed under Group I). 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).

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.

8. Prevention & Control of Rare Diseases:

Though in the last two decades, due to advancement in technologies, understanding of the pathophysiological mechanisms of rare genetic disorders has somewhat improved, yet the treatment modalities are few and the available therapies may not lead to "cure'. More importantly these are exorbitantly costly and not universally available & accessible. Accordingly, prevention needs to be the focus for all genetic disorders. The prevention of genetic disorders can be done at multiple levels. For application of these strategies, the first step is increasing awareness among the health professionals and population at large.

8.1 Education & Awareness

Education and awareness are the pillars for strengthening healthcare system. The education for suspicion, diagnosis and management of genetic disorders is limited among healthcare professionals at all levels. This acts as a barrier against early diagnosis of genetic disorders. The Central Government will work with the State governments to increase the awareness and knowledge of all levels of health care functionaries in this regard.

8.2 Prevention at different levels

Preventive strategies start with the education of general public and health professionals at large but more specifically in the preconception, antenatal, neonatal

and even after disease identification at various levels. The prevention of genetic diseases should be based on an integrated and comprehensive strategy incorporating:

8.2.1: Primary Prevention: This aims at preventing the occurrence of the disease. This strategy yields the highest returns in terms of decreasing the incidence & prevalence of rare disorders in the population in the long run. Some of the interventions under this strategy include:

a. Identification of high-risk families and couples in the reproductive age group based on family history of any rare disease and counseling for preventive measures.

b. High risk screening: Screening the pregnant women in the high risk families on a priority basis

c. If screening indicates a suspected case, counselling of the pregnant woman and families about the challenges of bringing up a child with rare disease and offer prenatal testing for checking the status of the fetus to enable them to make an informed decision about continuing the pregnancy in case fetus is affected.

d. Education and counselling about need to avoid marriages between carriers of genetic traits that might result in birth of children with rare diseases

8.2.2: Secondary prevention refers to the use of various interventions for the management of rare disease patients. This is primarily done to prevent the onset of complications associated with delayed diagnosis and treatment. Newborn screening is the best example of secondary prevention in which the babies are screened within few days of birth before symptoms of the disease manifest and treatment is initiated which prevents morbidity and mortality. Secondary prevention also includes early post natal diagnosis and treatment (before complications manifest) of disorders amenable to therapy which would require increasing awareness and better availability of diagnostics.Timely referral of the suspected patients & their families to appropriate facilities that are equipped to make a correct diagnosis and where indicated, initiate treatment is the key.

8.2.3: Tertiary prevention refers to provision of better care and medical rehabilitation to those rare disease patients who present at an advanced stage of the

disease. It encompasses providing best supportive care to the affected patients with various rare disorders including the ones for which no specific treatment is available. This would improve quality of life of affected individuals and families. Supportive care includes developmental assessment and intervention including early stimulation and behavioural intervention, physical therapy and rehabilitation, provision of visual and hearing aids and above all emotional and psychological support to affected individuals and families.

8.2.4: **Optimal screening and diagnosis strategy:** Considering the competing priorities within available resources, universal screening of all pregnancies and/or all newborns in the Country for all rare disorders is neither feasible nor justified. The policy recommends a screening and diagnostic strategy wherein those pregnant women in whom there is a history of a child born with a rare disease and that rare disease diagnosis has been confirmed, would be offered prenatal screening test(s) through amniocentesis and / or chorionic villi sampling. This strategy is in sync with the policy direction of reducing the incidence of rare diseases in the population. In case, the diagnosis could not be established during the prenatal period, it would be offered to the newborn or the infant as the case may be and would include newborn screening for (a) small molecule Inborn Errors of Metabolism by liquid chromatography – tandem mass spectrometry (LC-MS/MS), (b) diagnosis of SCID by T cell receptor excision circles (TREC) and (c) diagnosis of lysosomal storage disorders (LSDs) by microfluids / LC-MS/ MS.

9. Voluntary crowd-funding for treatment

9.1 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, prognosis, 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 category III.

9.2 The Government will notify selected Centres of Excellence, which will be premier Government tertiary hospitals with facilities for treatment of rare diseases. To begin with the following institutes would be notified as Centers of Excellence for Rare Diseases:

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 , 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

The cost of treatment of patient in these centres of excellence will be met out of donations received through the online digital platform, mentioned in Para 8.1 above.

10. Implementation strategy

Keeping in view lack of availability of epidemiological data on rare diseases, constraints on resources and competing health priorities, the focus of the Government will be on the following :

i. The Government will have a National Registry for Rare Diseases at ICMR with the objective of creating a database of various rare diseases. Steps have already been taken in this direction by ICMR. Over a period of time, the registry is expected to yield enough information to be able to arrive at a definition of rare diseases best suited to the Country.

ii. The Government shall take steps to create awareness amongst all levels of health care personnel as well as general public towards the rare diseases. This will encourage people to seek pre-marital genetic counselling, identification of high-risk couples & families and also result in prevention of births as well as early detection of cases of rare diseases.

iii. Public Health and hospitals being a State subject, the Central Government shall encourage and support the State Governments in implementation of a targeted preventive strategy .

iv. State Governments will be supported for undertaking screening & diagnosis amongst high-risk pregnant women as well as amongst suspected cases of rare diseases reporting to Government facilities as a result of the awareness building activities.

   a. For prenatal diagnosis: Amniocentecis and/or CVS testing would be offerred to those pregnant women in whom there is a history of a child born with a rare disease and that rare disease diagnosis has been confirmed diagnosis. The samples would be sent to testing and confirmation at Centers of Excellence for Rare Diseases.

   b. For newborns / Children: Neonatal screening amongst newborns with a family history of a rare disease (with confirmed diagnosis). Such newborns / children would be offerred screening and diagnosis, if required, at the centres of excellence and would include newborn screening for (a) small molecule Inborn Errors of Metabolism by liquid chromatography – tandem mass spectrometry (LC-MS/MS), (b) diagnosis of Severe Combined Immunodeficiency (SCID) by T cell receptor excision circles (TREC) and (c) diagnosis of lysosomal storage disorders (LSDs) by microfluids / LC-MS/ MS.

v. The Government shall provide financial assistance upto Rs. 15.0 lakh (under the Umbrella Scheme of Rashtriya Arogya Nidhi) to (upto 40%) the entitled population, as per PMJAY norms, for their treatment in Government tertiary hospital, for rare diseases amenable to one-time treatment (identified under Category I).

vi. The State Governments may undertake treatment of disorders managed with special dietary formulae or food for special medical purposes (FSMP) and Disorders that are amenable to other forms of therapy (hormone/ specific drugs)- diseases covered under Category II.

vii. The Government shall notify selected Centres of Excellence at premier Government hospitals for comprehensive management of rare diseases.

viii. The Government shall create a digital platform for bringing together notified Centres of Excellence where patients of rare diseases can receive treatment or come for treatment, on the one hand, and prospective voluntary individual or corporate donors willing to support treatment of such patients. Funds received through this mechanism will be utilized for treatment of patients suffering from rare diseases.

ix. The Government shall facilitate the creation of an enabling environment that promotes research & development of diagnostic and therapeutic modalities within the Country.