DakshamA Health & Education

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Report on Workshop to Demystify the National Policy for Rare Diseases

Supported By:-

Pfizer

Acknowledgements

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We would like to acknowledge our gratitude to the Ministry of Health and Family Welfare, Goverment of India and its representatives for participating in the consultation. We thank the doctors and physicians from the Centers of Excellence and the private sector who participated in the consultation. We would like to thank and acknowledge the insights from the representatives of the patient advocacy groups, patients and families who wholeheartedly participated in the discussions.

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Executive Summary

In India, rare diseases are a significant public health concern, although awareness and understanding of these conditions is still evolving. The exact prevalence of rare diseases in India is not well-documented due to limited data and underreporting. However, it is estimated that there are around 70 million people affected by rare diseases in India. The availability of treatments for rare diseases in India is often limited. Many rare diseases have no specific therapies; supportive care is the primary management approach. The high cost of treatment, particularly for orphan drugs, poses a significant challenge for patients and their families, as most of these medications are expensive and not easily accessible. Diagnosis and treatment of RDs is a complicated process that requires multisystem involvement and complex care by several healthcare providers.

The Government has launched National Policy for Rare Diseases (NPRD), 2021 in March 2021 with a revision in April 2022 and operational guidelines released in August 2022 **to address the public health concern on RD.** Provision for financial support of up to Rs. 50 lakhs to the patients suffering from any category of the Rare Diseases and for treatment in any of the Centre of Excellence (CoE) mentioned in NPRD-2021, outside the Umbrella Scheme of Rashtriya Arogya Nidhi was made.

The launch of NPRD and enhancement of coverage to Rs. 50 lakhs for all rare disease treatments was a huge win for the patients. Now after many months of implementation of NPRD, a Multi stakeholder workshop with policy makers (government officials), Centers of Excellence for Rare Diseases (COEs), treating physicians and (Patient Advocacy Groups (PAGs) was organised by **DakshamA Health**. **Objective of this workshop was demystifying the NPRD policy for Rare Diseases and together find solutions to address key gaps in the implementation of the current policy.** The day long workshop was centred on hearing the government initiatives and progress made so far as well as initiatives by the centres of excellence to identify and treat patients. The patient groups were divided into three groups to deliberate on the following topics-

1.Initiatives to Raise Awareness: How to Scale It Up
2.Establishing effective referral networks and pathways is essential for addressing rare diseases and implementing the National Policy for Rare Diseases (NPRD)
3.Financial access And Other Support Programs - How to Elevate the Reach and Access.

Key challenges identified in operationalizing the policy to improve its benefits for patients with RD were the following.

Lack of Awareness – amongst patients and treating physicians.

- Most patients are unaware of the process for seeking treatments, natural history of diseases and have no knowledge that the condition may require specialist knowledge for diagnosis. Years are often lost before a diagnosis is arrived at, leading to poor treatment outcomes or sometimes even loss of life.
- Most RDs are treated by Geneticists or Paediatricians, specialities other than genetics and paediatrics are often unaware of Rare diseases and their signs and symptoms. Patients presenting to other specialities are often missed or misdiagnosed leading to delays in treatment.

Scope of coverage

- Paediatric vs adult - **adult rare diseases** are not given recognition as most centres of excellence are established out of paediatric units. Adults with rare diseases face challenges for diagnosis and treatment as the policy lacks clarity in this aspect.
- Unlisted rare diseases – The NPRD has a list of rare diseases. Diseases outside this list face challenges for diagnosis and treatment access. Process for inclusion of new rare diseases is unclear and most physicians do not know how and where to report new cases.
- Listed rare diseases - Patients and families are unaware of the provisions for listed rare diseases and may discontinue treatment or not start it due to financial constraints. Not all COEs specialise in treatment of all rare diseases. Patients who arrive at COEs where their disease treatment is not available are often confused and frustrated and may be referred to a center that is too far for them to access. Information on the list of COEs specialising in treatment for specific rare diseases is unavailable. Process for listing new RDs, especially adult-onset ones like cardiomyopathies (ATTRCM), dermatology – (GPP, HAI) rare cancers and neurological conditions to be made simpler and included into the current systems.

Operational aspects for seamless utilization of the policy

- **Referrals** - referral mechanisms are weak with no clear guidance on referral from other public and private hospitals to the Centers of Excellence.
- **Patient navigation and counseling within the CoE** - The Centers of Excellence are large medical institutions and patients find it extremely challenging to navigate the campuses. Adequate information, guidance and signages are missing to help patients understand the complex process of application submission, follow up and release of funds status.
- **Listed diseases -** Treatment access for listed diseases is a challenge as processes for completing applications are long and complicated. There is no process for monitoring the progress of an application once submitted to the COE. Fund disbursement is a challenge for busy physicians who do not have the time to track applications.
- **Selection of cases for submission -** There are criteria for selection listed in NPRD and physicians use their clinical acumen to judge whether a case is clearing eligibility criteria for receipt of financial support. Patients often feel that the process was not explained well to them and in case of rejection do not have the means to understand the reasons for not qualifying.
- **Fund disbursal, treatment procurement -** The process from application to receipt of funds, procurement of medicines, and initiation of treatment is often long and arduous leading to frustrations at the patient level. There is no mechanism to track whether the application is successful or understand the timeline for fund disbursement. Even after fund disbursement, the procurement processes at CoEs are long and cumbersome leading to delays in actual start of treatment and care.
- **Capacity building at CoEs** - CoEs are understaffed and overburdened, struggling to manage a long list of patients within the limited financial capacity provided to them.

Conclusion and key takeaways

While there is a lot of progress made in operationalising the National policy for Rare diseases, much work still needs to be done. Some of the key takeaways are as follows:-

Awareness

1. A communication and mass awareness program led by the government but supported by multiple stakeholders to increase awareness and outreach would help dispel myths and improve access to authentic information.
2. Improve awareness of RDs amongst physicians of all specialities through physician training, CMEs and accreditation programs to maximise impact of the policy and provide benefits to a larger pool of patients
3. Create an interactive National information hub integrated into the existing fundraising portal for use of all stakeholders.
4. Improve inter and intra departmental communication for faster dissemination of policy decisions to enhance uptake of existing provisions under NPRD.

Scope of coverage

1. Consider the lifecycle approach for inclusion of all Rare diseases which have treatment options available including adult Rare Diseases. This will help the policy to attain a comprehensive character as envisaged.
2. Create district and subdistrict Rare Disease clinics and establish a robust referral mechanism.
3. As there are only 11 CoEs to date in a country as huge as India where private sector plays a crucial role in healthcare service delivery, a strong referral and linkage mechanism needs to be developed from private facilities as well. It is suggested that private physicians closer to the location of the patient be allowed to continue to provide treatment to the patients under the guidance of physicians at CoEs in a hub and spoke model.
4. Provide impetus to strengthening of rare disease registries to include all public and private diagnosing institutions on an urgent basis.

Operational aspects for seamless utilization of the policy

1. Create transparent systems for application and tracking using digital technology so that treating physicians and patients can have real time updates.
2. Streamline funds and procurement mechanisms with standard operating protocols to reduce turnaround time.
3. Promote research and indigenous manufacturing of medicines and supportive therapies.
4. Work closely with other ministries to raise funds and spread awareness on RDs.
5. Strengthen COEs with techno- managerial support to improve fundraising capabilities with PSUs and CSR
6. Create a national working group that includes representatives of PAGs.
7. In order to make the treatment more affordable, it was suggested to have a blanket exemption of Custom duty and GST for rare disease drugs, irrespective of whether they are provided by the CoEs. This will enable greater access of treatment to more patients.

Landscape of Rare Diseases

In India, rare diseases are a significant public health concern, although awareness and understanding of these conditions is still evolving. Patients with rare diseases often lack adequate access to treatment due to a lack of awareness and medicines. About 80% of rare diseases have a genetic cause. There is no standard global definition of a rare or orphan disease. In the U.S., a disease is defined as rare if it affects fewer than 200,000 people, while the threshold is 50,000 in Korea and 2,000 in Australia. The World Health Organization defines a rare disease with a frequency of 6.5–10 per 10,000 persons. In India, there is still no national definition of a rare disease, as the country lacks sufficient epidemiological data to define a rare disease . However, the National Policy for Rare Diseases has identified three groups of rare diseases based on their treatability, treatment availability, and status. India has a dedicated National Policy for Rare Diseases and allocated funding to strengthen Centres of Excellences (CoEs). Under the policy, eleven Centres of Excellences (CoEs) have been notified with fund allocation for infrastructure development, and patient care services for screening, diagnosis and prevention of rare diseases. A portal is linked to these eleven CoEs for uploading data of eligible patients and generate funds through crowdsourcing for treatment of the patients is available, in case the treatment requires more than Rs. 50 lakhs for the patient.

The need for the consultation

The Government has launched the National Policy for Rare Diseases (NPRD), 2021 in March 2021 with a revision in April 2022 and operational guidelines released in August 2022 to address the public health concern on RD. Provision for financial support of up to Rs. 50 lakhs to the patients suffering from any category of the Rare Diseases and for treatment in any of the Centre of Excellence (CoE) mentioned in NPRD-2021, outside the Umbrella Scheme of Rashtriya Arogaya Nidhi

The governments initiatives to support patients with rare diseases have raised hope in many, however the steps to access and the provisions in NPRD are not clear. Patients struggle to access the right information and navigate the complex medical institutions housing the designated COEs for treatment of Rare diseases. Physicians and health care providers are often unaware of the provisions of the NPRD and express inability to treat patients thereby creating missed opportunities for patients who can benefit from the program.

Considering the complicated healthcare systems in India, many of these suggestions still managed to find their way into the policy. Thus, it was felt that in order to make the policy more effective, a platform was required where the Patient advocacy groups, government officials, treating physicans and CoE administrator come together to deliberate on what more can be done within the policy, however very few suggestions have seen the light of the day due to complicated structures in our healthcare systems.

A Workshop with Rare Disease Patient advocacy groups and other rare disease stakeholders on National Policy for Rare Disease 2021 was conceptualised with the objective of **demystifying the NPRD policy for RD and together find solutions to address key gaps in the implementation of the current policy.**

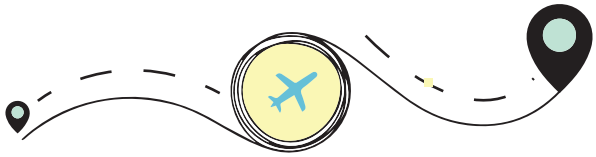
The following topics were considered for discussion

- **Awareness –** at CoEs, amongst patients and families, amongst treating physicians and amongst other specialities
- **Scope of coverage**
  - Paediatric conditions and as well as adult-onset rare diseases and access barriers
  - Listed diseases – treatment and diagnostic access, monitoring applications for patients, access to funds and medicines.
  - Unlisted rare diseases – whether a process exists for funding under the current policy. What could be a future pathway?
- **Operational aspects for seamless utilization of the policy**
  - Referrals- within COEs, from public health institutions to the COE, from private sector to the COE
  - Patient navigation and counselling within the CoE
  - Selection of cases for submission
  - Approval process for treatable rare diseases, supportive treatment for Rd's with no definitive treatment
  - Fund disbursal, treatment procurement at COEs
  - Capacity building at CoEs

The journey so far:-

The Ministry of Health has considered the need for treatment and diagnosis for rare disease patients and has formulated the National Policy for Rare Diseases. Of the 11 COEs currently operating some have specialized in certain diseases. Each COE needs to be upgraded as a Center of Excellence for a particular spectrum of Rare diseases. These can then handhold other COEs in that specific disease area. A proposal for supporting the COE's with a functional unit and Human Resources is also being considered to expand their ability to function more optimally. Technical guidelines series for Rare Diseases is being drafted in consultation with the specialists. Enhancing treatment access is being explored through day care centers identified in District hospitals. New COEs are being identified and will help increase access to many more patients.

The 20 lacs in the RAN pool was moved to an exclusive fund for NPRD and increased to 50 Lacs for improving treatment access. Processes for using the funds were also put in place and freedom was given to decide the beneficiaries to treating physicians. Challenges in decision for identification of beneficiaries is brought to CTRD under the chairmanship of DGHS for appropriate decisions. In 2022, 35 crores were released and in 2023, till date 42 crores have been released benefitting 203 patients. The ministry is trying to park a small unit within the COEs to support them to screen and conduct due diligence for offering the financial support to beneficiaries and after submission of Statement of Expenditure, request for more funds. The funds are untied and provide a lot of flexibility to the COEs to spend as per requirement within the guidelines provided by the ministry. WHO and UNDP is supporting the ministry for human resources. Ministry is exploring MOUs with private hospitals for transplants etc. Exemption of basic custom duty and GST is also being pursued.



While there has been a lot of distance covered, decisions and policy making has not been inclusive and patient voice has been missing in policy discussions. Patient and family inputs are necessary to guide the policy making and collective and collaborative approach is the need of the hour. The PAGs can bring forth the ground realities into a consultative process of policy making. A list of patient support groups working on various rare diseases exists with the ministry, this list needs to include more patient groups and be proactive in reaching out to new groups. The process for inclusion of rare diseases that have no definitive treatment, needs a representation from the PAGs. An institutional mechanism where all PAGs can meet with DGHS once on six months so that feedback can be received needs to be established. A rare disease advocacy group was created in 2019. Only one meeting took place and needs reinvigoration. Representatives from other groups like CSR, other departments can also be invited to some of these meetings.

Many patients do not reach the COEs and are treated by private hospitals. A discussion can be initiated with IRDAI for making a product for rare disease patients to support treatment costs by providing insurance cover. ICMR can be requested register all rare diseases in their registry irrespective of private or public sector.

Discussion points with PAGs

Recommendations from group discussions

Policy & Provisions

1

Streamlining Application Processes

Funds for support are available through various means and programs. However, there is no consolidated information that is readily available for patients as well as Health Care providers as a ready reference. The NPRD fund has guidelines and steps for approval process, however once submitted the patient or physician is unable to keep a track of the progress of the application. A common web portal with the ability to track applications would be useful for the patient as well as the physician.

Training and Education

Hub should contain information on diseases and specifics of treatment and diagnosis as an educational portal for physicians. Telemedicine and webinars can be held for training physicians in rural areas. Curriculum should have chapters on Rare Diseases in MBBS and other degrees. Ongoing certification and education programs for upgrading knowledge and skills for specialities should be made available on a rolling basis.

Centralized Information Hub

(Well advertised, constant updates, languages, awareness programs) It will help reduce load on doctors, bring more transparency and autonomy for patients. Patients can search for themselves and track their applications. Physicians can refer to it when treating a potential rare disease patient. The hub should be dynamic, comprehensive, option of being moderated, patient centric and focused on verified information which are of relevance to different patient categories with or without diagnosis.

Dr. Shalima highlighted the need of having on-line portal, where application/case filed can be uploaded by referring cardiologist to CoE Lead Cardiologist. This will help in on-line tracking and will avoid multiple visits of patients to RD CoE and will make the system transparent.

2

Patient Navigation Support

Patients who reach a COE can be referred to Patient groups for further support and navigation in the hospital. Patient navigators can be present in every COE as an integral part of the health system and offered an honorarium for their time and effort. Information booklets can be provided in regional language in an easy-to-understand format.

3

Establishing a National Coordinating Mechanism

Establishing a National Coordinating mechanism which brings together all PAGs, professional associations, civil society and organizing six monthly meets. Listening to and seeking inputs from PAGS and families to strengthen the ongoing program. This can meet twice a year to review the policy and add value by bringing inputs from stakeholders.

4

Procurement of medicines.

Since RD medicines are not listed in NLEM and are required in very small quantities for a select number of patients, procurement is a challenge in COEs and sometimes creates long delays which adversely affect the treatment outcomes of the patient. SOPs for procurement of medicines should be created with clear guidelines to the CEOs. Supportive therapies, nutrition and diagnostics should be included in the treatment support for the patient apart from the specific medicines required for the condition.

5

Relief from taxation

Medicines for Rare diseases including supplements can be considered for customs duty and GST waivers. An Orphan Drug act could go a long way in incentivising manufacturers to invest in new drug research or repurposing of existing molecules for the benefit of RD patients.

Spreading Awareness

Content to spread awareness including infotainment, patient stories, blogs and other means can be used to reach all sections of the society. Targeted awareness by defining a strategy and implementing it to reach specific audiences can be beneficial and raise awareness amongst families. Specific programs can be aired on national television like Doordarshan, Rajya Sabha TV etc. Finding influencing "voices" from the society who have a family member with rare diseases and using them to raise awareness can have a large impact. Specific communities with high risk can be targeted with focused messaging to reduce risk of having more than one child with the same disease in the family. Public Speaking engagements for patient advocates can help spread awareness by listening to the lived experience and patient stories.

Community Events

Schools and colleges and educational institutions can be approached to spread awareness on rare diseases. National and other Day Specific events like UHC day or World Health Day can be used as platforms to speak about inclusion of rare diseases in all policies. Partnerships with healthcare professionals, researchers and social workers to join in their events could bring support from professional associations to spread awareness

Standardized Referral Processes

Referral process streamlined and easily accessible from the hub should be built as a one stop solution to provide support for uploading information on new patients as well as access information on submitted applications. Referrals from rural areas and peripheries as well as private sector institutions to be enabled and a standard referral protocols put in place that is linked to the hub.. Creation of District and subdistrict RD clinics will help easy and early access to patients.

Inclusion of adult-onset rare diseases

COEs need to have a multidisciplinary approach so that patients are not lost in the hospitals. Dedicated clinics for adult rare diseases with specific days can be established within the COEs. Since adult onset RDs can present to other specialities, adequate information on the provision of RD policy should be made available in all departments. COEs can conduct physician training and CMEs to help young doctors and other specialities understand the provisions of RD policy.

Financial Access and Other Support Programs - How to Elevate the Reach and Access

Increasing Awareness about fund disbursement process

Fund disbursement applications and process to be made transparent and easy through use of technology. Both the patients and physicians today have very limited visibility on progress of applications for funding which causes mistrust. A transparent and easily accessible system for tracking applications needs to be developed such that the status and reasons for approval/rejection are understood clearly.

Techno managerial support to COEs

Providing techno managerial support to COEs can help improve the systems within the COEs to address the patient load, upload data on the portal and track applications. It can also support in writing proposals for raising funds. Operational challenges can be addressed to a large extent by providing nursing, data entry and administrative support to COEs.

Inclusion of all Rare Diseases including ones with adult onset

Currently only pediatric RDs are covered and listed. Process for listing new RDs, especially adult-onset ones like cardiomyopathies (ATTRCM), dermatology – (GPP, HAI) rare cancers and neurological conditions to be made simpler and included into the current systems. Adult rare diseases are not given recognition as most centres of excellence are established out of paediatric units. Adults with rare diseases face challenges for diagnosis and treatment as the policy lacks clarity in this aspect.

The NPRD has a list of rare diseases. Diseases outside this list face challenges for diagnosis and treatment access. Process for inclusion of new Rare diseases is unclear and most physicians do not how and where to report new cases. An information hub and application process should be initiated.

Conclusion

Governments initiatives to support patients with rare diseases have shown a positive trend with increase in financial outlay through the policy, increase in Centers of Excellence and better access to information around changes in policy. The National Consultation has created a platform and explored key areas for collaboration. Each stakeholder can bring unique skills to the table and with everyone's participation, faster reach and better access can be a reality for RDs patients and families. The recommendations of this report can be taken up by various stakeholders to strengthen the existing policy. Patient advocacy groups can play a critical role in spreading awareness, educating and supporting patients and connecting with treating physicians to enable faster uptake and earlier treatment access for those who need it most. The private sector can play a role by bringing in skilled clinicians and supporting the COEs in delivering high quality care, exchanging best practices and learnings as well as referring patients who need financial support. Tech companies can be approached to help set up the hub and support collection of data and analysis. Media and journalists can support raising awareness and dispel myths around rare diseases. The government can act as an aggregator to all these stakeholders and drive the agenda around increased uptake of the benefits of the policy and better treatment and diagnostic access to patients and families with rare diseases.

Agenda

Workshop on National Policy for Rare Diseases

Venue: India International Center, Kamla Devi Complex, Seminar Hall 3
Date: June 13, 2023
Time: 11:00 AM to 4:00 PM

Time	Topic	Speakers	
10:30-11:00	Tea and registration		
11:00- 11:05	Introduction and Welcome	Dr. Ratna Devi	DakshamA Health
11:20- 11:35	Initiatives by government to increase reach and access for Rare Diseases	Dr. L Swasticharan. Add DDG	DGHS
11:35-11:50	Financial support programs - CSR	Dr. Pramod Kumar Sinha - PhD	CSR expert
11:50-12:00	Challenges faced by PAGs and patients	Madan Gopal	ORDI
12:00-12:40	Panel discussion COEs- roles and responsibilities. Process for referral and treatment , procurement	Dr. Madhulika Kabra Dr Mamata Muranjan Dr. Ashwin Dalal Dr. Suresh Kumar COEs present	Centers of Excellence for Rare Diseases
	Group photo		
12:50-13:00	Introduction to group work	Dr. Ratna Devi	
	Group – 1- initiatives to raise awareness- how to scale it up	Madan Gopal (moderator) Manjit Singh, LSSDS (co-moderator)	
	Group- 2 establishing referral networks and pathways. Adult and pediatric	Dr. Sudha Bhattacharya, GNE Myopathy moderator Dr. Shalima Gautam, Jupiter Hospital, Co moderator	
	Group – 3 RAN and other support programs- how to elevate reach and access	Dr. Neerja Gupta AIIMS, Sameer, Indian Rhett moderator Syndrome (co-moderator)	
13:00-13:45	Lunch		
13:45-14:45	Group work		
14:45-15:30	Open discussion		
15:30-15:40	Closure and wrap up	Dr. Ratna Devi	DakshamA Health

Participants

Dr Astha Chug	Consultant, MoHFW
Dr.L Swasticharan	DDG, DGHS
Dr. Mamta Muranjan	Geneticist KEM Mumbai (COE)
Dr.Neerja Gupta	AIIMS DELHI (COE)
Dr Madhulika Kabra (online)	AIIMS Delhi (COE)
Dr. Ashwin Dalal (online)	Head, Diagnostics Division at Centre for DNA Fingerprinting and Diagnostics (COE)
Dr. Pramod kumar Sinha	Independent consultant CSR
Dr. Shalima Gautam	Independent consultant, Cardiomyopathies
Dr. Parul	Prader Willi Syndrome
Dr. Vandana	
Dr. Shudha Bhattacharaya	GNE Myopathy
Gitanjali Sehagal	Friends of SMA
Javed Hasan	Multiple Sclerosis Delhi
Madan Gopal	ORDI
Manjit Singh	LSSDS
Nisha Venugopal	INDO-US RARE
Samir	Indian Rett Syndrome Foundation
Sukhvinder Kaur	ALS support group
Vikas Bhatia	MERD
Clarinda Cerejo	MOGAD
Dr Ratna Devi	Dakshama Health
Chitransh Saxena	Dakshama Health
Amita Singh Chauhan	Dakshama Health
Vikash Prasad	Dakshama Health
Sunil Sharma	Dakshama Health
Mridula Kapil Bhargava	Dakshama Health
Khushboo Sharma	Pair Academy
Pravalika(online)	HAE
Nishant (online)	IBD
Representatives from Pfizer	

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https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7044965/
MOHFW, New drugs and clinical trial rules. Ministry of Health and Family Welfare, Government of India (GoI), 2019, pp. 149–264; https://cdsco.gov.in/opencms/opencms/system/modules/CDSCO . WEB/elements/download_file_division.jsp?num_id=NDU0Mg.