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PPHF
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# Brief on East Region Rare Disease Consultation

**December 12, 2020**

Strategic Partners

Organization for Rare Diseases India

LSDSS
Lysosomal Storage Disorders Support Society

Cure SMA India
Hope Beyond Boundaries

Research Partner

GRID Council

Knowledge Partner

Institute of Child Health, Calcutta, Founded 1952

Almost 80% rare diseases are genetic in nature, non-dependent to the environment and out of these, almost 70% manifest during the early years of human life majorly affecting children. This underscores the need for early diagnosis, newborn screening, and large-scale awareness. For a diverse country like India, working on such complex, time-sensitive plan calls for multi-sectoral engagement of stakeholders for discussing, working in tandem and paving the clear path in an integrated manner.

**People to People Health Foundation (PPHF), jointly with the State Government of West Bengal organized the virtual consultation on rare diseases for eastern states of India. The consultation was the first in the series - "The Blueprint for Rare Disease in India initiative", an attempt to enhance policy commitment to prioritize access, investment, and partnerships on rare diseases in India. The initiative plans to organize two more regional consultations in the South and North regions and one international consultation on 28th February, the international rare disease day. The initiative has been co-powered by Takeda Pharmaceutical. Initiatives associate partners include ORDI, LSDSS, Cure SMA Foundation of India (Strategic partners), GRID council (Research partner) and Institute of Child Health, Kolkata (Knowledge partner).**

The objective of the consultations through this initiative is to discuss and address the key issues that come in way of tackling rare diseases including management of rare diseases, list priority actions, explore partnership opportunities across the stakeholders and beyond for the development of better infrastructure, diagnosis, and management systems. Emphasis will also be on increasing collaboration and commitment to address Rare Diseases challenges. Apart from that, the high-level recommendations that come out from the consultations will be shared with the Government of India and Ministry of Health and Family Welfare to further strengthen the upcoming “Rare Diseases – National Policy”.

The eastern event opened up with an inaugural session where Government leaders including Dr. Saumitra Mohan, Mission Director NHM, West Bengal, Ms. Kriti Garg, Mission Director NHM, Andaman & Nicobar Islands and Dr. Pempa Tshering, Health Secretary and DG, Sikkim set the context of the consultation followed by panel discussions on subjects **– 1)'Key unmet needs in the management of rare diseases at the State level & Priority actions for states and best practices’, 2) 'Collaborations for better infrastructure, diagnosis & management', and;3) 'Indispensable role of peer support groups in prioritizing rare diseases'.** During the inaugural, a special rare disease awareness video had been released that aims at familiarizing key decision makers, implementers, healthcare professionals, resource providers and the whole community about rare diseases through the patient’s lens and make them more aware of the rare disease space and its challenges. You can watch the full video by clicking on the link: https://www.youtube.com/watch?v=zHye8nFusyQ&t=10s

The sessions were chaired by esteemed experts - Dr. Madhulika Kabra, Professor, Division of Genetics, Dept. of Pediatrics, AIIMS, Dr. Archisman Mohapatra, Executive Director, GRID Council, New Delhi, and Dr. Tanushree Mondal, Associate Professor & Assistant Director of Medical Education, Govt. of West Bengal. The respected panelists included Dr Debasis Bhattacharyya, Director, Dept. of Medical Education, West Bengal, Dr. Pradip Mitra, Co-ordinator COVID 19 & Former Director of Medical Education, West Bengal, Dr. Prabin Pradhan, Epidemiologist, Govt. of Sikkim, Dr. Apurba Ghosh- Paediatrician, Institute of Child Health, Kolkata, Mr. Koki Sato, General Manager, Takeda India, Mr. Pramod K. Sinha, Formerly with Dept. of Public Enterprises, Top-Management Member of MNCS & SCOPE, New Delhi, Mr. Naresh Kumar, GM, Power Grid Corporation Ltd., Mr. Prasanna Shirol, Co-Founder & Executive Director, ORDI, Mr. Manjit Singh, President, LSDSS, and Ms. Moumita Ghosh, Co-Founder & Director, Cure SMA Foundation of India.

The event witnessed approx. 300 participants from across multiple sectors- Government bodies from eastern states & international, corporates, NGOs, academia & healthcare professionals, PSUs, patient advocacy bodies etc. Watch the full consultation recording by clicking on: https://www.youtube.com/watch?v=dhvqr1ZG45w&t=19s

## Key Challenges Highlighted:

- Lack of awareness about rare diseases at the system, healthcare professionals and community level.
- Lack of proper training of undergraduate and postgraduate medical students on rare disease screening and management.
- Lack of systematic approach of diagnosis and treatment in rare disease space like other disease areas.
- Unlike other priority diseases that affect major number of populations, rare diseases affect few people because of which it falls behind when it comes to prioritizing within the health system. A simple example is COVID 19 which has affected masses and so, the world is busy looking for solutions and a lot of work has already been done in this area.
- At the State level, for many rare diseases diagnostic facilities are not available and these suspected cases are often referred to outside the state centers which increases the time of diagnosis and delays treatment.
- Many a times, challenges from remote territories of the country are given less weightage while drafting policies
- Diagnostic and treatment services available for rare diseases are quite expensive.
- Treatment for many rare diseases is available in India with long term benefits. Access of this treatment to diagnosed patients is challenging in current situation in liu of no national policy in force.
- Registration of Rare disease cases through ICMR and Nidan Kendra is still picking up and needs to be escalated by active participation of hospitals that have rare disease cases

## High-level Recommendations:

### Awareness Generation & Trainings:

- Provisions should be made for training of the health professionals on managing few common rare diseases at identified health facilities
- Training on rare diseases diagnosis and management should be made a part of the Undergraduate and Postgraduate medical curriculum and taught extensively
- Provisions for overall increase in the awareness of Health Care Providers involved as touchpoints in Lifecycle of the patient's journey

### Planning:

- Documentation of long-term and short-term actionable measure and best practices for clear guidance should be done at the State level and submitted to the center for strengthening the rare disease policy
- Define clear cut roadmap for different departments including healthcare, administrators and policy makers considering the current constraints they face like other health priorities, lack of awareness etc. The specifics should cover how can they evolve treatment protocols, healthcare management system and policy achieving the short-term goals to start with gradually leading to long-term goals

- Each region/state should come up with a committee/ team for taking rare disease cause forward. To begin with a group of clinicians like physicians, neonatologist, neurologists, obstetricians etc. should try to find out within their settings what kind of rare diseases patients they see more commonly and then State Government can be chipped in on how to plan a program at the State level and then club the center into it.

**Early Identification, Diagnosis & Treatment:**

- Define Diagnostic Algorithm for life threatening Rare Disease
- Identification and spread of multiple centers of excellence across the state-level with help from state and center to ensure patients must travel less to get diagnosis and treatment
- Early detection of Rare Disease in infants and adolescents (Newborn screening) should be prioritized at the State level and the policy level
- Critical role of ASHA/ ANM workers in early identification should be explored, defined, and implemented
- Development of app for early detection and referral to CoE can be explored for real-time data transfer to reduce the time of treatment
- Research & Development on gene therapy to eradicate rare diseases should be encouraged
- Early screening and prevention should be given equal attention as treatment aspects of rare diseases
- Insurance companies should include at least common rare disease in reimbursement panel
- Hospitals and institutes can be appraised and encouraged on the use of compassionate drug program that can be implemented for critically ill children with rare disease. The processes within the healthcare centers must be streamlined for smooth application of the program for enrolled patients as per the amendment in the drug and clinical trial rules 2019.

**Supportive Treatment:**

- Protocols for providing care of psychological aspect of patient and family by regular counseling should be specified and implemented
- Supportive care like physiotherapy, orthopedic procedures, behavioral therapies etc of rare diseases is the mainstay since treatment for very few rare diseases has been established till date. So, it is especially important for clinicians to understand which rare disease requires what type of supportive treatment. Also, few specialized institutes should be identified at the district and state level for providing supportive care.
- It is important to develop exclusive counseling centers for patients and their caregivers to support them psychologically.

**Funding & Collaborations:**

- Multi-sectoral approach to manage rare diseases is required where Government partners with private sector like pharma's, associations, PSUs and NGOs to manage rare diseases effectively
- Collaboration is intended to be holistic, both in financial terms (human resources, materials, and budgetary contributions) and in expertise-related terms (knowledge, networks, and capabilities). The strategy for collaboration for India could include i) to establish a mechanism for gathering collaborators, ii) to define and implement standards of care, iii) to enhance outreach of diagnosis and screening, iv) to ensure seamless supply of drugs, v) to set up and enrich registry, and vi) to enhance clinical research.
- Strategic collaboration is also highly essential to pave the path for advancing rare disease management in India in a futuristic manner. This is to build greatly on evidence-based knowledge, innovation in therapies, research and innovative access based on real-world data

and patient experiences. Innovative access models are the next level of collaboration wherein the industry and government health systems come together to build a pathway for patients based on value of therapies and quality of life in long term.

- PSU being one potential sector for making CSR contribution towards health, various innovative funding models can be explored between Government and PSU/ Government, PSU, and private sector under CSR for supporting specific area of rare diseases under health. Provisions under rare disease policy can be made for PSUs to specifically support rare disease management locally and nationally.

## Way Forward:

With all the listed challenges and possible solutions, it is also important to highlight the work that is already happening in the rare disease space in India and some potential collaborations that must be explored. ICMR has initiated a hospital based rare disease registry. ICMR has released an open call for hospitals to participate and around 40 institutes have already collaborated for the registry program. This will help generate some data around the burden of rare diseases in India for helping in further planning. MoH has a rare disease central technical committee which is working on central diagnostic procedures for relatively common rare diseases and the document will be available for experts soon.

In the last financial year, over 150 Public sector units contributed Rs 3876 Cr towards CSR including health, education, skill development and other areas under Section 7 of CSR act. This clearly highlights the significance of PSUs when it comes to heavy cost CSR funding like that of rare diseases. PSUs are divided based on the level of autonomy when it comes to financial and management aspects and hold greater potential for supporting and contributing to the country's wellbeing. The top 10 PSUs which are categorized as the most autonomous and with highest potential to fund include: Bharat Heavy Electricals Ltd, Bharat Petroleum Corporation Ltd, Coal India Ltd, GAIL India Ltd, Indian Oil Corporation Ltd, NTPC Ltd, Oil & Natural Gas Corporation Ltd, Steel Authority of India Ltd, Hindustan Petroleum Corporation Ltd, and Power Grid Corporation Ltd. Last year Rs. 5686.75 Cr was available as CSR funding out of which Rs. 3876 Cr was spent by PSUs under CSR with unspent corpus amount standing. Out of the amount spent, 34% was contributed to health, sanitation and eradicating poverty and hunger. This indicates the kind of potential that lies with PSUs when it comes to exploring possible funding mechanisms and collaborations to support specific areas like rare diseases under health. Amendment opportunities in the clause VII of the CSR laws can be explored for PSUs and pharma industry to support treatment of rare diseases.

Acknowledging the critical role of peer support groups as awareness generators and mobilizers amongst the community, interface between health system and rare disease population, facilitators for developing highly technical committees at the local levels and working on the spectrum of requirements under the rare disease space is of utmost importance.
The consultation clearly states the huge possibilities that lies with various sectors whether private, PSUs or Government. Need of the hour is to evidently list down the challenges, workout clear cut solutions and then decide which solution will be owned by which sector or sectors in collaboration. Policy needs to clearly state these roles and call for partnerships. Policy should also consider making exceptional protocols since we are talking about managing rare diseases which will have rarer solutions.

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