Background

Rare diseases usually affect small populations of patients, sometimes fewer than 1 in 2,000 individuals. Most rare conditions are chronic, complex, degenerative, painful, and often fatal. These rare conditions result in significant unmet needs for those impacted along with their caregivers, ranging from quality of life and health, equality, social inclusion, and access to education, as well as employment and other chances to contribute back to society.

Ministry of Health and Family Welfare, India approved a national policy for rare diseases in 2021. The policy aims to reduce the incidence and prevalence of rare diseases through an integrated and comprehensive preventive strategy that includes raising awareness, early screening, counselling programs and enabling patients with rare diseases to access affordable health care within the constraints of resources and competing health care priorities.

Many patients suffering from rare diseases that are still not recognized or covered by the present NPRD face significant challenges in early diagnosis and management. The challenges include not having confirmatory molecular diagnostic facilities, identified and approved treatments, and epidemiological evidence to judge the burden of diseases and treatment costs for the condition. As most conditions require multispecialty interventions, all stakeholders that patients encounter throughout their journey must have their roles and responsibilities defined. A significant challenge is the lack of accelerated regulatory pathways for the treatment of new rare diseases under the NPRD. Concerted efforts and defined actions should be in place and propagated to support the inclusion of new rare disease diagnostic and treatment protocols.

In a follow-up to our first-round table discussion DakshamA Health organized a second-round table discussion on the theme ***" Roadmap for the inclusion of new rare diseases—Discussion on stakeholder responsibilities and accelerated regulatory pathways"***, **30th July 2022, Saturday, 04:00 PM IST**. The round table discussion deliberated about the roles and responsibilities of the stakeholders involved in the identification, treatment, and inclusion of new rare diseases, as well as explored accelerated regulatory pathways for new rare dermatological disease treatment options.

The first panel discussion focused on the opportunities to synergize and facilitate communication between the Centers of Excellence (COE) for Rare Diseases and dermatology departments, strategies to overcome a lack of awareness, information asymmetry, and how professional bodies can contribute to the inclusion process. The second panel discussion focused on the pathways for developing standard treatment protocols for new rare diseases, as well as the role that regulators can play in fast track approvals and registration of new treatemnst

The following list of experts in the discussion:

Panel Discussion 1: Stakeholder Discussion on landscape of Dermatology Rare Diseases in India
Chair: Dr. Sunil Dogra, ***Prof. at PGIMER Chandigarh***
Dr. Madhulika Kabra ***Professor, Division of Genetics, Department of Paediatrics, All India Institute of Medical Sciences, New Delhi***
Dr. Seema Kapoor ***Director Professor Pediatrics and Coordinator of Genetic Unit & Lab, MAMC***
Dr. Rashmi Sarkar ***President IADVL-2022, Director Professor, Dermatology, Lady Hardinge Medical College***
Panel Discussion 2: Accelerated regulatory pathways for rare dermatological disease treatments
Chair: Dr. Y.K.Gupta ***President - AIIMS Bhopal, AIIMS Jammu, Chairman, Board of Directors, BIBCOl, DBT, GOI***
Dr. Vibhu Mendiratta ***Professor, Lady Hardinge Medical College representative of the DGHS***
Dr. Reeta Rasaily ***Ex. Scientist 'G' and Head, Division of RBMCH & Nutrition, Chief Nodal Officer-Northeast, Region, ICMR***
Dr. Alok Bhattacharya ***Prof, Ashoka University and World Without GNE Myopathy***

The following report summarises the discussion points. The actionable points will highlight some of the solutions and recommendations that will set the stage for the inclusion of new rare dermatological diseases in the National Policy for Rare Diseases. This would also support any other new rare disease apart from the dermatology speciality.

Key discussion points:

- **Awareness:**
  - There is a significant lack of awareness among healthcare professionals about the national policy on rare diseases and the benefits available to patients under the policy. Interdepartmental and intradepartmental communication needs to be strengthened to facilitate smoother flow of information. The **Centres of Excellence for rare diseases can come forward and write to the departments within their institutions about the provisions under NPRD.** This would help in increasing awareness and inter-departmental communication.

- **Benefits entitled under Rashtriya Arogya Nidhi (RAN) Scheme:**
  - Under the RAN scheme, **patients receiving day care treatment may have their biologics costs covered.**
  - By **submitting a request to the RAN committee of the respective centres of excellence, funds under the RAN may be requested for new rare diseases.**
  - The funds from the RAN can be used gradually, but they shouldn't go over Rs. 50 lakhs. The **scheme does not necessitate that the funds be spent all at once.**
  - **Information asymmetry on drugs** available for treatment under RAN and other central and state government schemers **can be overcome by writing to ICMR with a list of medicines for consideration for inclusion into Essential list of medicines.**
- **Inclusion of new department as co-opted member under rare disease committee:**
  - Patient needs are addressed on a case-by-case basis. If a **rare disease has a treatment, a member from the treating speciality can be co-opted from the same field and expertise**. For example, under the rare genodermatoses, if a dermatologist is needed, they can be co-opted as one of the members. The treating doctor of the head of the department can write to the Focal point of the Core Committee of the COE for inclusion
- **Support by central technical committee/centres of excellence:**
  - **Dermatologists from various COEs can collaborate to project their needs and the support they may require** for rare dermatological patients and submit it to the central technical committee.
  - The **treating clinician can submit a request for funding or inclusion of a new rare disease (not listed in the NPRD) to the CTC** along with information on the disease epidemiology, cost of treatment, effectiveness of treatment, and likely response to treatment. This will then be reviewed by the technical committee, and **it could be added to the list of diseases.**

- **Standard treatment protocols:**
  - An **expert group consisting of experts from diverse backgrounds** (Regulator-ICMR, Health economist) and treating physicians can be formed to conduct evidence-based research, **which can then support development of standard diagnostic and treatment guidelines.**
  - A Special interest group will be formed with interested dermatologists to lead customization of diagnostic and treatment protocols in the context of India. **IAVDL will lead this activity under the guidance of the President IAVDL**

- Molecular diagnosis is required to establish natural history and epidemiological data. **Centers with molecular diagnostics can be identified for confirming clinical diagnosis.**
- A **special working group consisting of patients, regulators and experts to be constituted to fast-track availability of specified medicines approved by FDA or EMA** and not available in the country on a named patient basis.

❖ **Rare disease registry**

- If data from patient/academic driven registries is to be extracted and included in national rare patient registries**, data quality must be ensured. A few experts can work together to establish fields and format and write to ICMR for inclusion. IAVDL will help establish the group and lead the activity**
- If we are to include any rare disease in the registry, we must emphasize the following points: the **epidemiology of the disease, the treatment available and its effectiveness, the reason why the recommended drug is effective, symptoms of the disease that can be controlled with locally available medicines, symptomatic and supportive treatment options and mechanisms for pooled procurement.**
- **Healthcare professionals can record information epidemiological and clinical data of patients with rare diseases receiving treatment or visiting hospitals after obtaining due consent from patient party** in order to keep records on the severity of the diseases. Guidelines for this can be developed and distributed
- A **mechanism can be developed to pool all dermatological rare conditions data on a single platform**, which can then be presented to policymakers so that the required action can be taken.
- If the treatment is still not available for any rare disease and the **data of the patients is built into the registry; it gives the patients an opportunity to be part of the clinical trial for the development of the drugs.**
- To ensure uniform data collection across the centers, standard proforma / format can be developed. However data quality must be ensured. One of the centers may take the responsibility for coordination.
- The **coordinating institution / center / centers may approach ICMR** expressing desire to participate in the national registry
- Data collected under patient registry / national registry can provide valuable platform for generating information on natural history of disease, availability and response to treatment and also an opportunity for treatment by participating in clinical drug evaluation trial if any.
- Data pooled together from different institutions could be a valuable resource for the policy maker for consideration of rare diseases in rare disease policy.

❖ **Role of professional bodies:**

- **Dermatological associations can disseminate information about advances In treatments and diagnostics, information on policy changes, availability of funds for treatment etc. through their platform**
- **Special interest groups within associations can assist in developing treatment referral pathways for patients.**
- They can ensure that **dermatologists continue to receive medical education and training** to stay tuned to the latest treatment protocols for dermatological rare diseases.

- Professional bodies/dermatologists can **work together to create a standard treatment protocol for rare dermatological diseases** and submit it to the ICMR to take further action.

Conclusion:

The list of the new rare diseases is still evolving. As a result, it is critical to have standard protocols or mechanisms in the public domain that supports inclusion for new rare diseases under the policy. Some opportunities for accessing services exist, but clarity on pathways for utilization and mechanisms by which patients can benefit are still lacking. There is still a significant gap in understanding and awareness of the national policy for rare diseases and the benefits that patients can receive under it. This can be overcome if experts and policymakers come under one umbrella to develop a standard roadmap for availing the current services that are available.

If a patient with a new rare disease (not on the list) turns up at a healthcare facility, and if possible, treatments and diagnostics are available, they must be translated into a standard format with the help of a pool of experts in the field and regulators to make them available to patients. The next critical step in developing these standard guidelines and roadmaps is to take a holistic approach that does not limit them to treatment and diagnosis but also extends their support for managing a specific rare disease.

Another important aspect is the research and development of rare diseases. This is possible if a consortium of academic institutions, professional bodies, experts, and researchers work together to build evidence and data. This would aid policymakers and regulators in taking the required action. Some of the rare disease inclusion's learnings can be adopted and applied to the evolving rare diseases.

This discussion not only provides a possibility and hope to a single vertical of specialty, but it also provides hope to other new rare disease patients beyond the dermatology specialty.