To,

22 March 2017
New Delhi

Committees on Rare Diseases
Government of India

Reference: **"ORDI's points of view and recommendations for rare diseases policy framework for India"**

About the Organization for rare diseases India (ORDI):
ORDI is a national non-profit organization with offices at Bengaluru, India and at Herndon, VA USA. ORDI was founded to create an umbrella organization unifying and representing the collective voices of all patients and other stakeholders of rare diseases in India. ORDI mission is to raise awareness about the more than 7000 rare diseases among patients, caregivers and doctors in India. We facilitate and foster national and international collaborative research programs to advance our understanding of rare diseases, discovery of novel diagnostics and orphan therapeutics. ORDI is a member of the Global Alliance for Genomics and Health (GA4GH), International collaboration on rare diseases (ICORD), EURORDIS, IRDiRC, and collaborates with Rare Genomics Institute (RGI), George Mason University, RareDR, NIH office of rare diseases, Global genes and other international initiatives. ORDI and RGI received the Sanofi Genzyme's Patient Advocacy Leadership Award in 2016. ORDI sponsors include numerous multinational companies such as Quintiles, Genzyme, Shire, and Centogene.

National Agenda on Rare Diseases:

(1) To review the current situation of rare diseases in India and define rare diseases in the Indian context

The first and most recent review of challenges and opportunities for rare diseases in India was published in 2014 in *Genetics Research* (see attached article).

ORDI's points of view:

- India is lagging behind by at least 34 years (USA adopted Orphan drugs Act in 1983) in recognizing the needs of rare diseases compared to less populous advanced countries. Given the population size and consanguinity prevalent in Indian communities, it is an **urgent need to start off with adopting a national policy on rare diseases** suitable to the Indian context.
- While there is no standard definition for a rare disease, different countries have adopted a single and consistent definition to facilitate research, development and policy making at the national level. Currently, the only proposed definition published in a peer-reviewed journal is '**any disease or condition that affects less than 1 in 5,000 persons**' (see attached). It is easy to argue for alternative definitions but **since there cannot be a "perfect definition for a rare disease", its an informed yet heuristic based executive decision to be made by an esteemed committee** appointed by the Government of India at the earliest. This can then kick-start the process of listing out all rare diseases in India – which cannot exist without a standardized consistent national definition.

(2) To establish norms for laboratories/ centers for diagnosis of rare diseases.

With the estimated number of patients with rare disorders in India at 7 crore, a **national initiative to accelerate diagnosis with advanced pre-natal, neonatal and adult-onset assays is critical to identify and account for patients** with these diseases as most patients remain undiagnosed. Numerous genomics based tests have been launched recently in India to diagnose hundreds of rare diseases in a single assay. We recommend the **appointment of a consortium of expert medical geneticists representing all India to formulate best practices for diagnosis of rare diseases. ORDI is willing and able to convene such a forum and distribute the published best**

practices to all pediatric and genetics centers across India. It is important to consider and include international best practices from standards organizations such as American College of Medical Genetics (ACMG), Institute of Medicine (IOM), Association for Molecular Pathology (AMP), etc. which renowned experts in the field can contribute**. USA has a similar initiative such as ClinGen led by board certified medical geneticists that might serve as a model for us to consider.**

(3) To recommend the design for creation of a national registry system for rare diseases.

Creation of a nationally integrated patient registries can immediately follow the publication of a national definition of rare disease. This could combine existing newborn registries and other accurate population health databases. ORDI is able and willing to implement this program with support from the central government and in collaboration with major medical genetics centers across India. While there is no single off-the-shelf software or data framework for this purpose, the National Institutes of Health has published common data elements and tools to help in this regard. A collaborative program with NIH could also be explored rather than re-inventing the wheel.

(4) To suggest funding mechanisms for dealing with rare diseases.

Given the early stages at which we are in India, the central government can create financial grants for advocacy, field work, research and development with four-fold objectives.

a. Administrative grants to convene national and international workshops, conferences and committee discussions with clearly defined goals and objectives. This should include travel expenses for consultants involved. This will include supporting **operations of committees** on diagnostics best practices, creation and maintenance of Patient registries, Evolving national and state level policy frameworks for rare diseases, scalable hub-spoke model to establish centers of excellence for rare diseases nationally to foster patient data aggregation, etc.
b. Funding programs for research innovations of low-cost alternatives for diagnosing, treating or supporting devices for patients with rare diseases. Even **mobile phone apps, wearable devices, natural or ayurvedic medicines, dietary supplements or drugs** with demonstrated success to aide patients could be **"made affordably in India"** aligned with Prime Minister's Make in India campaign**. This needs to be with an emphasis on development of viable products for immediate benefit to patients** and not simply conducting basic research.
c. **Incentives for biopharmaceutical and medical devices** companies with availability of FDA approved medicines for rare diseases to make them immediately available and accessible to patients in India.
d. **Budget to enable free treatment of patients with rare diseases where approved treatments are available anywhere in the world.** While Government hospitals in India are setup to give free treatment to patients with common diseases including hospitalization for surgeries, they have been refusing to treat patients with rare diseases even when approved treatments are available due to expenses involved. **Recent high court decisions in New Delhi and Karnataka** have resulted in free treatments to patients with lysosomal storage diseases. **ORDI, in collaboration with the center for human genetics has setup a center of excellence for rare diseases at the Indira Gandhi Institute for Child Health to utilize the state government's budget of 3 crore rupees** to make available free treatment for LSD patients. A similar **program in every state of India with sufficient budget to treat all patients would be a major breakthrough in addressing the patients' immediate unmet needs**.

ORDI would be interested in applying for grants of type (a) and facilitate the supplemental dissemination of grants of type (b) and (c) to grant applications selected by eminent committees appointed by the Government.

(5) To recommend measures to create awareness among the general public regarding prevention and management of rare diseases.

Since its conception just 4 years ago, ORDI has created enormous momentum through mass media, social media and press to create national level awareness about rare diseases – a majority of which tend to be genetic in origin. See http://www.ordindia.org and www.racefor7.com.

The awareness needs to be created at multiple levels – general public, educated public, medical and scientific fraternity, diagnostic laboratories, policy makers and data/informatics community. Of these, all but the general public understand and consume information in English. **ORDI received PAL grant award from Sanofi Genzyme** for the creation of informational audio/visual content on Lysosomal Storage Disorders in English and **translation into major regional Indian languages** to reach out to masses. ORDI also has various rare diseases information dissemination in regional Indian languages such as Kannada. We are certainly setup to do a lot more with the support of the central and state governments.

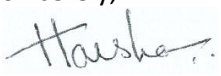
Creation of high-quality genetic counselling programs is recommended to produce sufficient number of **next-generation genetic counselors to counsel patients, parents and inform family planning activities in India**. A **collaboration with George Mason University and health systems at Fairfax, Virginia, USA could be facilitated by ORDI**.

(6) To frame a draft national policy for rare diseases in India.

Please see **attached ORDI's draft policy** for your consideration. The policy needs to most importantly address patients' needs of accessibility and affordability of diagnostics/treatments. The policy needs to be holistic in nature to also **incentivize stakeholders such as biopharmaceutical R&D organizations** to **encourage investment in therapies without obvious economic returns.** The US Orphan Drugs Act of 1983 and its amendments could serve as initial guide of major elements. Doctors are equated to Gods in Indian culture due to their selfless service to mankind – however, from a practical standpoint, **even Doctors have to be incentivized to justify their additional effort to learn, understand and implement best practices for diagnosis and treatment of rare diseases** in India. Enhancing curriculum for medical education and **incorporation of Indian Ayurvedic medicine for certain rare diseases** which have no cure in allopathy could be considered. The initial policy framework and budget allocation needs to be **with a long-term view (say by 2030) to make rare diseases as easily diagnosable and treatable as common diseases** in India.

ORDI team will be extremely obliged to participate in and contribute towards your committee's future activities, programs and events. I have been informed by the international committee on rare diseases policy of my inclusion in their planning committee for India. Hence, **ORDI is in a fortuitous and privileged position to bridge the international best practices** that have evolved over the last 34 years **for the benefit of patients with rare diseases in India**.

Sincerely,



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