

Commentary: Rare eye diseases: More than meets the eye!

The authors have made an honest effort under the aegis of the Women Ophthalmologists Society of India to create awareness on the need of having an eye registry for rare diseases affecting the eye.^[1]

There is no universal or standard definition of a rare disease. A condition that occurs infrequently is generally considered a rare disease, and has been described in terms of prevalence—either in absolute terms or in terms of prevalence per 10,000 population. A country or region defines a rare disease most appropriate in the context of its own population, race, health care system, and resources. In 2019, the European list of rare diseases included 53 ophthalmological diseases, which were classified as rare diseases and another 103 systemic diseases with ophthalmological involvement, out of a total of 7,000 rare diseases.^[2]

A research registry of individual case reports when compiled becomes a useful case series. By using rigorous study designs, a case-control study can be designed based on the exposure or outcome between the patients of the cohort.^[3] Before starting a registry, ample planning and discussion with experts is essential. Any selection or data bias can impact the study outcome. We are in the era of big data, which can be from electronic medical records, sources like bio-bank, and administrative databases, in different formats—unstructured or structured—and for different

purposes (administration, clinical care, or research). This big data would be helpful in studying clinical outcomes, bringing about improvements in the work process and in fueling research to improve the quality of life for those affected with rare diseases.^[4] Electronic medical record–based analytics of big data for uveitis has been recently published, and a similar exercise for rare eye diseases would be the first step in collating data, analyzing them and bringing out meaningful insights.^[5]

The Ministry of Health and Family Welfare, Government of India, formulated a National Policy for Treatment of Rare Diseases (NPTRD) in July 2017. Unfortunately, there were huge gaps in implementing the policy. The reasons are multi-fold and included financial aspects, concurrence from different states in the country, public and private partnership, and diverting funds from common treatable diseases that affect the masses like cataract to rarer eye diseases that affect only a few.^[6] Hence, a decision was taken to reframe the NPTRD and an expert committee was constituted by the Ministry of Health and Family Welfare in November 2018. Based on this report and with the approval of the competent authority, the draft National Policy for Rare Diseases was finalized and placed in the public domain on 13 January 2020, inviting comments and views from all the stakeholders, general public, organizations, and states and union territories.^[6]

To take this task forward, ophthalmologists can work with Centers of Excellence (COE) for rare diseases designated by the Government of India in major cities of the country. There is also a proposal to further expand the reach, if necessary. The role of COE would be to provide education and training

to personnel involved in managing these diseases, antenatal and neonatal screening, cytogenetics, molecular and metabolic diagnostics. The scope also extends to prevention of the rare diseases by pre- and ante-natal counselling and research for low-cost diagnostics and therapeutics for rare diseases.^[6] Center of Excellence for Rare Eye Diseases (CERED), which is functioning at LV Prasad Eye Institute, Hyderabad is the first-of-its-kind center focused exclusively on advancing eye care and treatment of patients who are diagnosed with rare eye diseases or eye conditions related to rare systemic diseases.^[7] Leading eye institutes in India can collaborate to initiate a rare eye disease consortium and set guidelines on collection, dissemination, and publication of data.

Rare diseases place a huge economic burden and more so in resource-constrained settings. The financial capacity to support the exorbitant cost of treatment is an important consideration in public health policy development with reference to rare diseases. It is pertinent to balance competing interests of public health for achieving optimal outcome for resource allocation. Financial support is available from the Government of India up to Rs 20 lakh under the Umbrella Scheme of Rashtriya Arogya Nidhi for rare diseases that require a one-time treatment. Beneficiaries for such financial assistance would be extended to 40% of the population, who are eligible as per 23 norms of the Pradhan Mantri Jan Arogya Yojana, for treatment in government tertiary hospitals only. Individual hospitals may also have a Rare Disease Fund to provide long-term financial support for patients with rare diseases requiring treatment with high cost medicines.^[8] Crowd-sourced funding is another option to be explored for the treatment of rare diseases.

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