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**Editorial**

# Rare diseases are not rare in India: Government must provide more research and screening

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A rare disease is a health condition of the infrequency of their occurrence in general population. It is also referred as an orphan disease. The occurrence of these diseases is marginally higher in India. The number of factors influencing the high prevalence of these diseases included high birth rate, consanguineous marriages, scarcity of expertise, poor government support, lack of early diagnosis and counselling services. However, additional disadvantages are unawareness and paucity of dedicated healthcare policies. These diseases are chronic, enfeebling, and life-threatening nature due to this the treatment of rare diseases is long, specialized, and expensive.

The world health organization (WHO) has reported rare disease with a prevalence of 1 or less per 1000 population, but many countries define rare diseases differently as per their suitability and requirement. It is estimated that globally there are around 6000-8000 rare diseases exist. Out of these about 350 rare diseases are more common in population[1]. The most frequently occur rare diseases are Thalassemia, Haemophilia, Sickle-cell Anaemia, Lysosomal storage disorders, Primary Immuno-deficiency syndrome, Auto-immune diseases, Cystic Fibrosis, Muscular Dystrophies, and Haemangiomas.

Some reports have indicated that over 350 million people are affected globally, whereas, nearly 60 million people affected in the United States and Europe. Currently, more than 70 million people are living with rare diseases alone in India. These reports clearly showed that the prevalence of rare diseases is high in India as compared to the United States and Europe[2]. As per the national policy for the treatment of rare diseases, rare diseases excessively impact children. About 50 per cent of rare diseases onset at birth and 35 per cent deaths happen before the age of one. Due to inaction and apathy cases are increasing day by day and, we are losing children every day with significant numbers. These diseases are important causes of morbidity and mortality in developing countries like India[3].

In 2016, the Judicial system took the wake-up call and directed the Indian government for implementing a rare disease policy at the earliest and guided to different states, including Union Territory governments about the establishment of district-level committees to give treatment and diagnosis of rare diseases. But still, these patients are struggling to avail of treatment and care. Sadly, we are losing many patients every month in the absence of timely treatment. It is estimated that about 90 per cent rare diseases have no approved treatment and approximately 1 in 10 patients receive disease-specific treatment[4]. The challenge becomes even greater as some rare disease treatment excessively expensive. At present very limited pharmaceutical companies are producing drugs for these disorders globally and there are no Indian manufacturers. Due to the high cost of drugs, the Indian government is unable to provide these drugs for free. The annual recurrent spending of one patient (enzyme replacement therapy) could range from 0.18 to 1.7 million per kilogram of body weight. If we apply this calculation for a child weighing 10 kgs, the estimated cost would be between 1.8 million to 17 million[1,5].

The correct incidence and prevalence of rare diseases are still unknown in India due to the paucity of epidemiological data. India should have arrived at its definition based on prevalence, disease severity, and population. Although all known rare diseases have not been reported in India, and regional distribution of rare diseases, prevalence data and mutation profiles are not well known. There is a great need to undertake systematic epidemiological studies in the Indian population. Implementation of national policy for the rare diseases will help in the diagnosis, management, and counselling for subsequent pregnancies with rare disorders. The research and screening programs are mandatory for the development of guidelines and policies, treatment advancement and cost estimation. Inadequate clinical experience and relatively little knowledge of pathophysiology are the major challenges in research and development of rare diseases.

Presently there is no cure and complete treatment for rare diseases except gene therapy and genome editing which are under clinical trials.

Rare diseases should pay more attention at a priority level. Because, these diseases are generally serious, chronic and life-threatening in nature which are requiring long term and specialized treatment. We should focus on the multipronged and multisectoral approach to tackle rare diseases. The diagnosis and treatment for rare diseases should get the same priority as other diseases. We should establish or improve genetic services including neonatal screening, networking and international and regional collaboration of genetic centres. There is a need for active collaboration between a clinical geneticist and basic scientists for research advancement. Systematic, effective, and affordable approaches are the need of the hour for these dreaded disorders. The government should also develop and update guidelines or management policies, promote quality control programs, support research in these areas and provide technical and financial support for prevention and disease control.

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## REFERENCES

[1] Commission of the European Communities. Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges. 2008. Available from: *https://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf.* [Accessed November 3, 2019].

[2] Rajasimha HK, Shirol PB, Ramamoorthy P, Hegde M, Barde S, Chandru V, *et al*. Organization for rare diseases India (ORDI) - addressing the challenges and opportunities for the Indian rare diseases' community. Genet Res. 2014;96:e009.

[3] National Policy for Treatment of Rare Diseases. 2017. Available from: *https://mohfw.gov.in/sites/default/files/Rare%20Diseases%20Policy%20FINAL.pdf.* (Accessed October 5, 2019).

[4] Rahme E, Joseph L. Estimating the prevalence of a rare disease: adjusted maximum likelihood. J R Stat SocSer D 1998;47:149–58.

[5] Martinez-Pardo M. Orphan drugs and metabolic disorders. Rev Neurol 2001; 1-15; 33:220-5.