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Rare Diseases Need Not Be Neglected Anymore

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India has over 450 types of rare diseases in India. Lack of awareness and stigma attached to rare diseases have added to the woes of patients suffering from them. The exorbitant cost of therapies makes many patients opt out altogether. Experts opine that the road ahead to make notable progress in this area of healthcare is long and the right policy measures will determine how this neglected space fares.



The death of a 13-year-old Ghaziabad boy suffering from Hunter Syndrome or MPS-II due to the non-availability of medicine and the right treatment is something to be pondered on. The boy was suffering from what is known as a rare disease or an orphan disease.

Rare diseases are nothing new to us and according to the Ministry of Health and Family Welfare, there are about 450 diseases recorded so far in India. If we go by the estimates, 50 per cent of new cases are in children and are responsible for 35 per cent of deaths before the age of one year, 10 per cent between the ages of one and five years and 12 per cent between five and 15 years.

The commonly reported rare diseases in India are Haemophilia, Thalassemia, Sickle-cell Anaemia and Primary Immuno Deficiency in children, auto-immune diseases, Lysosomal storage disorders such as Pompe disease, Hirschsprung disease, Gaucher's disease, Cystic Fibrosis, Hemangiomas and certain forms of muscular dystrophies.

According to the World Health Organisation (WHO), these diseases are debilitating, lifelong disorders whose prevalence is less than one per 1,000 persons. They include autoimmune disorders, congenital malformations, inherited cancers, and certain endemic infectious diseases that have very low prevalence.

Hurdles

Lack of awareness and the stigma attached are the two major concerns for rare disease patients and their families. Though the Ministry of Health and Family Welfare formulated a National Policy for Treatment of Rare Diseases (NPTRD) in 2017, the implementation was faced with a lot of challenges. Lack of clarity on the exact number of patients, cost-effectiveness of interventions for rare diseases vis-a-vis other health priorities etc. took a toll on those who suffered from the disease. Poor awareness among the healthcare staff and lack of proper diagnostic facilities also compound the woes of the patients.

The other major concern is the cost of treatment. The exorbitant cost of therapies prevents many patients from going in for the right treatment and many leave halfway.

Ramaiah Muthyala, President and CEO, Indian Organisation for Rare Diseases opines, "Rare diseases are genetic disorders; for them, there are no cures, only treatments. Generic orphan drugs, about 350 APIs are manufactured by Indian pharma and all are being exported. They are formulated elsewhere and come back into India at an exorbitant price and import license requirement. As a result, none of them is easily accessible for Indian rare disease patients. Modern therapies for rare diseases, including gene editing, gene therapy, ASOs, RNAi, mRNA, and ERT are highly promising but unaffordable and inaccessible to the Indian patient community. Although a small percentage of rare disease patients need expensive drugs, others can be managed by alternate therapies such as diet, medical foods, the Indian system of medicines (AYUSH), social support, and medical devices, which improves the quality of life."

Indian Organisation for Rare Diseases has been promoting collaborations with patient organisations, e.g., Rare Disease International (RDI), European Organization for Rare Diseases (EURORDIS), National Organization for Rare Diseases (NORD), the International Rare Diseases Research Consortium (IRDiRC), the International Collaborations on Rare Diseases and Orphan Drugs (ICORD).

Creating awareness

Advancements in medical research and technology will be the key drivers for the development of innovative drugs for the treatment of rare diseases. AI is also being used to improve the diagnostic process. Apart from this, developments in the precision of genetic testing and the clinical use of genome sequencing are now also accelerating the diagnosis of rare diseases.

The Department of Biotechnology has set up Nidan Kendras under the Unique Methods of Management and Treatment of Inherited Disorders (UMMID) project for genetic testing and counselling services. These Nidan Kendras perform screening, genetic testing and counselling for rare diseases.

Recently, the Government of Karnataka launched a research and training unit for rare diseases. The unit will strengthen diagnosis, counselling and treatment, including developing the service facility for prenatal diagnosis for early identification of genetic disorders in pregnancy. The activities to be undertaken at the Research and Training Unit for Rare Diseases include

comprehensive genetic counselling facilities for around 2,500 families with rare disorders, to be evaluated annually; increasing the capacity of diagnostic testing, including exome sequencing; and developing training and service facility for prenatal diagnosis for early identification of genetic disorders in pregnancy. It will also provide hands-on training in laboratory genetics and incubate startups in developing low-cost therapies for rare disorders along with conducting regular educational modules in Kannada, English and Hindi for raising awareness of human genetic disorders, their early recognition, and treatments available, among pre-university college students in the state. The unit will be around 40,000 sq ft with three floors.

Dr Sheela Nampoothiri, Clinical Professor, Department of Pediatric Genetics, Amrita Hospital, who recently joined ICMR's Rare Diseases National Consortium's expert committee as co-chair says, “It is really important if we are starting treatment from a very early stage. Hence, it is extremely important to have such medical treatment facilities to be made available in India. Regarding the rare disease policy, as the cost of therapy is exorbitant, the policy statement is suggesting crowdfunding as the model to raise funds for patients.”

Amrita Institute is currently treating seven patients with Pompe disease apart from treating 17 patients with five types of treatable Lysosomal Storage Disorders and these patients are being treated through a charitable access programme. Recently, the institute has started enzyme replacement therapy (ERT) for a patient with Niemann Pick B disease and this is the first patient to be started with ERT from India.

According to the institute, before 2008, there was no availability of treatment and these babies were all dying before the age of one year. ERT needs to be started at the earliest in the newborn period itself and treatment is given as an intravenous infusion once every two weeks. The therapy should be given for life because they lack the enzyme alpha-glucosidase in their body.

The SAT Hospital in Thiruvananthapuram Medical College has started the registration process for treating diseases as part of the centre of excellence programme. All rare disease cases will be registered with SAT hospital even if the disease was diagnosed in other hospitals. The registered patients will be provided the benefits of the centre of excellence programme at the treating hospital.

In partnership with the All India Institute of Medical Sciences (AIIMS) Jodhpur and the Dystrophy Annihilation Research Trust (DART), the Indian Institute of Technology (IIT), Jodhpur has established a research centre for Duchenne Muscular Dystrophy (DMD). It is characterised by advancing muscle weakness and degeneration as a result of changes to a protein called dystrophin.

Exploring treatment methods

Pfizer is exploring options for some early-stage treatments for rare diseases in a bid to focus on high-impact medicines. According to the company, there are plans to focus on internally developing rare disease treatments using technologies such as gene editing, while exploring external opportunities for early-stage gene therapy programmes.

The Subject Expert Committee functional under Central Drug Standard Control Organisation (CDSCO) has given a waiver to Cipla to conduct Phase IV study of Tocilizumab Injection for giant cell arteritis, a form of rare disease.

Takeda Biopharmaceuticals has launched two medicines namely Adynovate, an extended half product for the treatment of haemophilia patients and Cinryze to treat hereditary angioedema (HAE) in India.

Ruchi Sogarwal, Head, Public Affairs and Patient Advocacy, Takeda Biopharmaceuticals, says, “It will be instrumental to build innovative access initiatives and strategic partnerships across the rare disease landscape to bring customised solutions for patients with targeted education, awareness, diagnosis, and treatment for effective disease management. Takeda is committed to introducing medicines in focus areas that have a huge unmet need. With this aim in mind, we launched two of our innovative medicines.”

Researchers at the Indian Institute of Technology (IIT), Madras, Tel Aviv University and Columbia University are working on developing a drug to treat a rare genetic brain disease known as GNB1 Encephalopathy, which affects individuals at the foetal stage. The research is supported by an Indo-Israel Binational grant offered by Israel Science Foundation (ISF) and India’s University Grants Commission (UGC).

In December 2022, the Centre was directed by the Delhi High Court (HC) to release Rs 5.35 crore to enable clinical trials for medicines that can treat rare diseases such as Duchenne Muscular Dystrophy (DMD). The HC observed that the “magnitude of the problem suffered by children with rare diseases leaves no doubt in the mind of the court that the creation of treatment

therapeutics for children with rare diseases would be considered as a nationally important project.” It also observed that the efforts for crowdfunding for treatment of DMD and other rare diseases had not yielded much results as the issue does not appear to have gained enough importance in the society in general.

The Biotechnology Industry Research Assistance Council (BIRAC) with Hanugen Therapeutics was to conduct clinical trials, however, things were delayed due to lack of funds. The HC also directed BIRAC to release the above-mentioned fund to Hanugen Therapeutics to begin the trial.

The way forward

Affordable medicines are the need of the hour. Epidemiological data needs to be in place with proper policy measures. Awareness among the ASHA workers who can go to the interiors to update on the exact number of rare disease patients across the country will help to create a database which will eventually help to come up with certain policies needed. Pharma companies need to re-look at strategies to do more clinical research for rare diseases.

On a good note, the Government of Tamil Nadu offered to treat a girl diagnosed with Systemic Lupus Erythematosus free of cost.

Sanjiv Das

sanjiv.das@mmactiv.com