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â??Itâ??s imperative to manage indigenous production of drugs to reduce cost of therapies for rare diseasesâ?•

31 May 2024 | Interviews | By Shivani Thakar

Dr Meenakshi Bhat, Faculty, Centre for Human Genetics, Bengaluru who has been involved in initiatives to raise awareness about rare diseases for nearly two decades, entered the field when there was hardly any awareness about a handful of rare genetic diseases like Downâ??s Syndrome. Now, with the government's involvement and a steadily increasing effort from aware citizen volunteers are slowly but surely advancing the establishment and development of the infrastructure required for tackling rare diseases and helping patients. While multiple challenges are faced on various dimensions â?? be it lack of awareness about rare diseases, relatively slow-paced translational research to develop new drugs and the exorbitant price of existing therapies that mostly need to be brought in from other developed nations, a handful of organisations such as Organisation for Rare Diseases in India (ORDI), are contributing their fair share to alleviating patient distress. In an interaction with BioSpectrum Dr Meenakshi Bhat shared her views on ongoing translational research in the area of rare diseases in the country and plans for the next one or two years in tackling rare diseases, among other issues.



What has it been like to raise awareness about rare diseases among patients and medical professionals?

It is important to emphasise that quite a few times, doctors fail to recognise that the symptoms of a patient are due to an underlying rare genetic disease. So, we need specialised doctors who have the time to take detailed patient histories, dissect the spectrum of problems the patient has, and then order the right kind of diagnostic tests the patients may require. This is especially important when the diagnosis is required to take place in a key time frame so that it does not become too late to help these patients. This is the case mostly in children.

Very few doctors in the current medical infrastructure are trained to correctly diagnose rare diseases, and even fewer to do pregnancy-related diagnoses. We have less than 200 doctors in each category of rare diseases for a population of 140 crores. Stressing the importance of training doctors to deal with rare diseases, we have been conducting training programmes for a one-year fellowship course for people with a Doctor of Medicine (MD) in Paediatrics to consult, diagnose, and treat patients with rare diseases. This has been running through the Rajiv Gandhi University for Health Sciences (Indira Gandhi Institute of Child Health) since 2013. As of today, the fellowship has successfully imparted the necessary skills to fifteen doctors and they are now spread all over the country.

Compared to the West, where people get diagnosed with rare genetic diseases when the baby is in utero, India is still a few steps behind wherein ‘we are lucky if we can diagnose a child with a rare disease in the first seven years of life when the first symptoms start to arise.’ This gap is due to the current lack of awareness about the existence and understanding of rare diseases in several communities and social classes, and with events like ‘Racefor7’ and organisations like ORDI, this is exactly what we aim for – to raise that initial awareness at a very early stage so that families can be prepared and an infrastructure of medical help for the new-born children can be set-up to fruitfully deal with alleviating the disease manifestations.

What are your views on the fundamental and translational research ongoing in the country?

At the level of fundamental research on rare diseases, it seems to be ample in the country. The level at which medicine and science talk to each other is when real development happens. In that area, the Department of Biotechnology (DBT) and Indian Council of Medical Research (ICMR) have been supportive in terms of research funding, as well as in aiding the set-up of registries for recording epidemiological data of patients with rare diseases which is an important aspect of being able to tackle these.

The next question is being able to afford the medicines. One of the most challenging parts of dealing with the issue of rare diseases begins after a diagnostic test is done because either suitable treatments or medications do not exist, or they are often not affordable. We need to increase the awareness about diagnosis and also the type of treatments we need to develop. In 2021, the Government of India announced the National Policy for Rare Diseases, wherein any individual, child or adult, who has a rare disease that has a definitive treatment will be provided with a health cover of rupees Rs 50 lakh per year by the Government of India.

What infrastructure needs to be in place to create an ecosystem of opportunities for all kinds of stakeholders involved in tackling rare diseases?

I think it will probably always be a “Hub-and-spoke Model”. There will always be a central body in a city or state that has the ease of communication. But without a patient advocacy group like ORDI, one can never make everything successful by operating in isolation. Spoke-wise, enabling an infrastructure of online consultation by doctors for those patients who cannot travel to the healthcare facility.

What, according to you, would be a good plan for the next few years for tackling rare diseases?

I think we should start focussing more on core, translational research to boost ‘Make-in-India’ efforts. Start small, reap the low-hanging fruits by aiming to manufacture and commercialise the existing drugs, and aim to develop new drugs in the long term.

To achieve the aim of lowering cost of therapies for patients with rare diseases, it is imperative to manage indigenous production of the required drugs.

How many patients have been able to receive treatment or help for alleviating the pain of rare diseases?

Independent of ORDI, our Centre for Human Genetics, combined with the state’s children’s hospital, we have seen and consulted around 30,000 families (not just children) affected by rare diseases.

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