GeNeDit

Care for the Rare: Steps in the Right Direction

Editorial

February 28th was celebrated across the world as Rare Disease Day. Many centres in India held events to commemorate this occasion. This has once again thrown the spotlight on rare diseases and the patients and families affected with them. There are around 7000 to 8000 rare diseases and around 80% of them are due to genetic causes. Though individually rare, these disorders collectively pose a huge health burden. In India alone, due to the large size of our population, the number of patients affected with rare diseases is estimated to be at least around 70 million. Majority of these disorders are lifelong conditions and cause significant disability, and affected individuals need supportive care and multidisciplinary management throughout life. Disease-specific therapies and disease course-altering treatments are available for some of the conditions, especially the inborn errors of metabolism, inborn errors of immunity, hematological disorders, and some neuromuscular disorders such as spinal muscular atrophy and Duchenne muscular dystrophy. However, the cost for many of these therapies remains prohibitively high. Recent initiatives by the government such as creation of the National Registry for Rare and Other Inherited Disorders (NRROID) by the Indian Council of Medical Research (ICMR), formulation of the National Policy for Rare Diseases 2021 (NPRD-2021) by the Ministry of Health and Family Welfare (MoHFW), Government of India, and the Unique Methods of Treatment and Management of Inherited Disorders (UMMID) initiative by the Department of Biotechnology (DBT), are commendable steps towards improving the diagnosis and treatment of these patients. The year 2023 began on a very positive note, with the MoHFW releasing grants for treatment of many patients with rare diseases under the Rasthriya Arogya Nidhi (RAN) financial assistance scheme, and inclusion of three more government medical institutes as centres of excellence (CoEs) for rare diseases to cater to the large population across India, taking the number of CoEs from eight to eleven.

Like the disorders treated by them, medical geneticists in India have been a rare (!) group of specialists. Until recently, only few centres had trained medical geneticists and even fewer offered specialized training in the field. The number of available trained medical professionals adequately equipped to handle the huge burden of rare disorders in the country has been dismally low. The scenario is fortunately changing for the better. More medical institutes and tertiary care hospitals are now setting up medical genetics units and departments and more centres are now offering training courses in clinical genetics compared to about a decade ago. Though these efforts are in the right direction, there is still a long way to go to create a sufficiently big workforce of clinicians in the country to manage patients with rare disorders. It is a matter of great pride for the Indian medical genetics fraternity that Professor IC Verma, one of the founding fathers of the specialty in India and patron of the Society for Indian Academy of Medical Genetics (SIAMG), has been conferred the Padma Shri award for the year 2023. Not only is this very inspiring and encouraging for the budding medical geneticists in the country, it will also help to bring greater recognition for the field and create more awareness about rare diseases in India, something that Professor Verma has striven for throughout his entire professional career.

One of the best strategies to reduce the burden of inherited disorders and birth defects is to perform preconception and prenatal screening, offer appropriate prenatal genetic testing to identify these disorders associated with significant mortality and morbidity, and terminate affected pregnancies to prevent the birth of affected babies. However, at times, overzealous prenatal screening and testing may lead to antenatal detection of conditions such as sex chromosome aneuploidies, which in many cases do not cause significant disabilities. This could cause a lot of emotional trauma to couples and lead to wrongful termination of pregnancies. These issues have

been addressed in the GeneFocus and GeneVista articles in this issue. As for all other medical techniques, one has to ensure that while offering genetic screening and testing, the first and foremost principle of 'primum non nocere' ('first, do no harm') is followed.

As always, we hope our readers find the articles included in this issue relevant and useful in their clinical practice.

Dr Prajnya Ranganath
Associate Editor
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