GeNeDit

More and More for Rare

Editorial

Come February and there is a flurry of activity related to rare diseases to celebrate Rare Disease Day on 28th February. Last week I saw three patients of Gaucher disease type III on a single day, in the outpatient department. For a medical geneticist, rare diseases are not so rare, as 80% of rare diseases are genetic disorders. But still, getting 3 patients of a rare disease on one day is quite rare! Research and funding agencies are taking up rare and genetic diseases in a big way. There are many projects and multicentric task forces on rare disease groups like movement disorders, primary immunodeficiency disorders, undiagnosed disease network, etc. In addition, I would like to acknowledge targeted activities by government agencies. This includes the Virtual Centre for Molecular Medicine, the National Registry for Rare and Other Inherited Disorders (NRROID) by the Indian Council of Medical Research (ICMR), the Center for Rare Disease Diagnosis, Research and Training by India Alliance, and the Mission Program on Pediatric Rare Genetic Disorders as well as the UMMID (Unique Methods of Management and treatment of Inherited Disorders) initiative by the Department of Biotechnology, Government of India. Of these, UMMID and the Rare Disease Registry need special accolades for their ambitious nature and far-reaching positive implications. The Rare Disease Registry will provide much needed data about rare disorders, to assess the burden of these conditions in our population and for policy-making. UMMID is an ambitious program involving training of doctors in genetic diagnostics and establishing genetic diagnostic centres called NIDAN (National Inherited Disorders AdministratioN) Kendras at many hospitals and medical colleges. The UMMID initiative also has a component of prevention of genetic disorders in the form of newborn screening and thalassemia screening programs in district hospitals in aspirational districts. This is an example of an outreach program where the women and children of lower socio-economic strata in less developed districts will get modern preventive medicine services. This will also help in direct training of the medical and paramedical staff of district hospitals and prepare them for countrywide programs of this nature. Thus, UMMID has shown rays of hope for integration of medical genetics services in maternal and child care.

The rays of hope have really brightened the lives of families with spinal muscular atrophy (SMA) in 2020. The currently available three new modalities of treatment including gene therapy have been received by some patients of spinal muscular atrophy in India and clinicians are excited about it. On this Rare Disease Day, SMA families in association with the Indian Academy of Pediatrics, organized a program on an update on SMA and aptly titled it 'All India SMA (SMArt) Update and Ray of Hope in 2021'. Many well-wishers of rare disease families celebrated the Rare Disease Day with various activities; to mention a few: the 'Pledge 4 Rare: 3 decades of caring for Rare Diseases, from our home to yours' organized by Sanofi Genzyme on 27th Feb 2021 and the 'Race for 7' organized by the Organization for Rare Diseases, India (ORDI). The 5th National Conference of the Indian Society of Primary Immune Deficiency on 6th and 7th March was also an important and informative event for rare diseases. These celebrations brought a lot of positivity amongst families of affected individuals as well as among doctors. All the stakeholders have come together on various platforms and are jointly working for the goal of better treatments for patients with rare disorders in India. The trained medical geneticists, 'awakened clinicians', and pharmaceutical companies with humanitarian approach will work with government policy-makers to make the much-awaited rare disease policy a reality soon. Many new therapeutic developments and gene therapies for many disorders have shown success in the research setting and they will hopefully become available to our patients in the near future.

'Care for the Rare' – This is the message we would like to share!

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