GeNeDit

Ray of Hope for Patients with Rare Diseases

Editorial

Many genetic diseases continue to lack curative treatment. Being rare, the numbers of patients with these disorders are small and this limits the interest of researchers involved in developing treatments for rare disorders. Thus, rare diseases continue to get neglected all over the world. Many countries have shown commitment to these disorders by way of grouping these 'Orphan' diseases and putting in separate efforts for drug development and for making the existing drugs available to the patients. India was and still is battling against nutritional diseases and infectious diseases which contribute greatly to morbidity and mortality, especially in infants and children. Any efforts for diagnosis, prevention and management of genetic diseases were resisted at the first step as they were not considered a priority as against other major contributors to mortality and morbidity. But now as the situation in India is changing as reflected by decreasing infant mortality rates, the contribution of rare genetic diseases and birth defects has become important. This is being realised by the medical community as well as government policy makers. There is a welcome change in the attitude towards genetic diseases and this comes at the right time when the country is in the transition phase of going from the status of a developing to a developed nation.

Thalassemia and haemophilia are the commonest of the genetic disorders and government funding for the comprehensive management of these diseases is already in place in many states in India and is gradually taking the shape of well organised programs. The recent "Initiative on Hemophilia Care III - A sensitization programme and the way forward" organized on the World Hemophilia Day, 17th April 2017, gave insights into the programs running in various states and the commitment of doctors and the Haemophilia Federation. The presence of policy makers and funding agencies of government at the meeting was very soothing and the complete support to establish state-of-the-art haemophilia management in India was obvious. Similar improvement in the situation for thalassemia and hemoglobinopathies is visible over the last few years. As these health care programs are getting established, one looks forward to similar support for other rare disorders. An initiative for other rare disorders has begun. The first important step in this direction was to launch a national rare disease registry which has now been initiated with the support of the Indian Council of Medical Research (ICMR). This was a part of the National Initiative for Rare Diseases (NIRD) organized by the Department of Paediatrics, All India Institute of Medical Sciences, New Delhi and ICMR on 26th and 27th April 2017. This meeting was attended by all stakeholders: patient support groups, clinical geneticists, pharmaceutical companies, and government representatives and the deliberations will guide the government policy for rare diseases. In addition to making the novel drug treatments available to Indian patients, issues related to research in drug development in India were also discussed. Indigenous drug development may be the ideal long-term solution as the drugs available currently are exorbitantly costly and funding treatment for patients in India may be a mammoth task, because due to the large population, the absolute numbers of patients of rare diseases are also not small.

The third important meeting in April 17 in the area of rare diseases was the ICMR-Inserm-DBT Symposium on Ethical and Scientific issues of Gene Editing using Crispr-Cas9 Technology on 27th – 28th April 2017. CRISPR–Cas9 technology has revived the hope of gene therapy for all monogenic disorders. As the world is trying to make the best use of the technology to provide simple answers to the gigantic problems of untreatable disorders, Indian scientists also need to take up the challenge. At this stage this meeting is a very welcome step. This small group meeting brought scientists and clinicians in India together and provided an opportunity to interact with scientists and bioethicists from France and other countries in Europe as the

international collaborator for the meeting was the French National Institute for Health and Medical Research.

The hope for curative therapies has again brightened the horizons for patients with genetic disorders as is reflected in some of the articles in the GenExpress of this issue. We hope that Indian scientists contribute to the development of gene therapy and novel drugs based on the understanding of pathophysiology of the diseases and provide affordable treatments. As new drugs develop, the Indian government has shown commitment to provide patients and families ready access to treatments. April 2017 had been a happening month for geneticists and patients with genetic disorders.

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