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Care for the Rare

Editorial

Twenty-ninth February 2020 was celebrated all over the world, by patients' families and clinicians, especially medical geneticists. This year being a leap year, this rare disease day is really rare. Other than being a really rare day, the Rare Disease Day of 2020 really is a milestone in the history of rare diseases in the world as well as India. The diagnosis of rare genetic disorders has become easy, thanks to the availability of next generation sequencing-based clinical exome and whole exome sequencing. Gradual introduction of whole genome sequencing in clinical diagnostics is improving the diagnostic yield by way of identification of rare disease-causing variations like deep intronic variations, structural rearrangements disrupting genes and novel genes for rare phenotypes. The GenExpress in this issue highlights the use of whole genome sequencing in clinical practice and the utility of complementary techniques of RNA-seq and metabolomics. Promising treatments for certain genetic disorders have been developed with the help of genetic engineering technology in the past few decades. The success stories of treatments for lysosomal storage disorders, spinal muscular atrophy, ectodermal dysplasia and epidermolysis bullosa are a few such examples. Many treatments are rapidly getting approved for patient care and many such as drug therapies for plexiform neurofibroma, Proteus syndrome etc. have shown miraculous results.

For rare diseases in India, this decade marks a new beginning. State-of-the-art diagnostics for genetic disorders is now available at an affordable cost. Trained medical geneticists and clinicians of the twenty-first century are ready to take up the challenge of medicine in the molecular era. As next generation sequencing is the first diagnostic test for monogenic genetic disorders, not only clinicians but the country as a whole need to develop guidelines for issues related to next generation testing. This issue of Genetic Clinics discusses the issues of secondary findings/ incidental findings which need urgent attention in the Indian scenario.

The second reason to rejoice in India is the Government's first step towards rare genetic disorders. After an ambitious but turbulent initiation of a policy for rare diseases, the Government retreated back making all of us unhappy and gloom descended on the concerned stakeholders. But now, the policymakers have floated a draft on the Government's plan for rare diseases. Though it does not cover the access to treatment in a major way, the will to help is obvious. The Government plans to support prenatal diagnosis in a big way, which is a cost-effective strategy. We hope to see implementation of this in this year and rapid modification to pave the way for coverage of treatment in the coming years.

With the positive developments for rare diseases, no clinician can afford to miss a genetic diagnosis. The clinical presentations of many rare genetic diseases overlap with those of non-genetic, infectious, autoimmune and other environmental diseases. Charting a pedigree and clinical suspicion in appropriate clinical situations are the *'MANTRAS'* for the accurate diagnosis of genetic disorders. We do our job of education and awareness about genetic disorders by way of short term courses, quiz in the Genetic Clinics, etc. I would also like to remind you about the 'Cases for Opinion' assistance on the website of SIAMG [http://iamg.in/New_Cases_For_Opinion_2018/New_Cases.html] where experts can suggest the diagnosis for your challenging cases.

'Care for the Rare' is our motto. Challenge and hope for a better future in this new decade!

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