Medical Genetics in India: Coming of Age

Editorial

The Society for Indian Academy of Medical Genetics (SIAMG) was formed in 2012 and is now more than a decade old. Last month the eighth conference of SIAMG (IAMG 2023) was held successfully in New Delhi. The scientific program was excellent and reflected the current areas of research. The posters and oral presentations reflected the plethora of work being done in India. The number of participants has become sizable and was representative of the work being done in various areas. The increased number of participants means more patients are being catered to. The burden of rare diseases in India is huge due to the large population.

In addition to the work on diagnostics, there were many talks on the therapeutic aspects. Success stories of gene therapies are showing promise for many more disorders. The presentation of research work in sickle cell disease and Duchenne muscular dystrophy being done in India gave confidence that the 'Make in India' policy will make the latest therapies available to Indian patients at an affordable cost. The funding agencies in India are also focusing on the space of therapeutics of rare diseases. Products made by Indian pharmaceutical companies for Wilson disease, tyrosinemia, hereditary angioneurotic edema, etc. have become available in India. The central government's rare disease policy is being implemented in a practical way. The panel discussion on the policy gave some facts and discussed the way ahead. The beginning has been done but the road ahead appears bumpy. Research proposals on therapies for spinal muscular atrophy and Gaucher disease are being presented in the task force of funding agencies and show the talent and capabilities of Indian scientists. IAMG 2023 provided a forum for clinician-scientists interactions which is an important mini-cosmos for the generation and nurture of seedlings of new ideas.

The revolution of next-generation sequencing (NGS)-based diagnostics has metamorphosed the field of genetic diagnostics, and its role is very important whether it is ordered at the first visit or after basic evaluation. The ways to by-pass the limitations of NGS-based diagnostics are discussed in the GenExpress of this issue. Hope we soon have one test for all types of genetic variations, and a big database of disease-causing variations in India. The IndVar database inaugurated at the conference needs to be strengthened. We wish it will be a resource not only for variant interpretation in diagnostics but also a platform to plan and develop collaborations for functional studies amongst Indian medical geneticists.

With improved diagnostics and new therapies and a big hope of therapies for many more diseases, I hope that in the near future, prenatal diagnosis will lead to treatment rather than termination of pregnancy. With the hope that 2024 will usher in many new therapies in the world and in India, I wish you all a very happy new year.

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