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Clinical genetics and India

Sir,

India, with a population of 1.21 billion, having diverse origins is the second most populous country in the world. Clinical genetics/medical genetics, the "latest" branch is still in its infancy stage as compared to other developed countries. For the last 10 years, there has been an increase in awareness, but it still remains a subject of less interest for majority of the professionals working in healthcare.

The health services in India are mainly divided into government-funded public health system and private sector. The government sector lacks in terms of infrastructure, facilities, and "super-tertiary" branches such as clinical genetics, except few specialized centers in India. Every government district hospital does not have a clinical geneticist, and thus, the clinicians trained are not exposed and are unaware about the services. The private sector too has few clinical geneticists trained from these specialized centers but inadequate for the mammoth population of India.

The diagnosis of these rare diseases depends on the state of art testing which is expensive and not available in public sector. Only 5-10% of the Indian patients are insured, and thus, most of the patients need to spend money from their own pockets for the testing.

All categories of childhood genetic disorders can be seen in Indian population from thalassemia[1] to lysosomal storage disorders.[2] There lies a great need to study the epidemiology of these genetic disorders in Indian population. Most of these genetic disorders are "rare disorders" which are difficult to diagnose.

The referral centers for these genetic disorders need to be allotted where diagnosis, treatment, and prenatal diagnosis can be done under one roof. One such initiation is done by the Indian Council for Medical Research for lysosomal storage disorders in the form of National Task Force Multicenter Collaborative Study. Inborn errors of metabolism are an additional aspect of clinical genetics, and newborn screening is made compulsory in few states. Further, awareness in the field of oncology is underway where additionally cancer physicians and cancer surgeons are utilising the knowledge of genetics for predictive, preventive and therapeutic management of breast, colon and lung cancers.

Initiation of public policy/rare disease policy will definitely help patients with genetic disorders in the diagnosis, management, and also counseling for subsequent pregnancies. Few of the genetic disorders such as lysosomal storage disorders have an expensive treatment in the form of enzyme replacement therapy which is extremely difficult for Indian patients.[3] There has been rapid growth in scientific and technological advances, which necessitates additional professional and public awareness regarding genetic disorders through continuing medical education, scientific conferences, and other such programs.

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Reply to the Letter to the Editor

Reply to Clinical genetics and India

The availability today of the means to know the entire sequence of every human gene, before we even know the meaning of this information, has led to increased interest in genetic counseling. Breast cancer molecular diagnosis has brought up the possibility of a definitive or at least predictive diagnosis of other cancers. Recent advancements have allowed for high through output gene expression profiling using new technologies like next generation sequencing and genome-wide association studies.

Once the DNA has been analyzed, many ethical issues come up that affect not only the patient but also the children and other family members. Checks and balances are needed, and testing should remain the prerogative of the medical practitioner, and be carried out by laboratories that have secure methods of storing personal information.

To be able to diagnose whether, we will actually get a disease is only part of the story. Hence, many new questions can potentially be asked, regarding severity, drug responsiveness, and prognosis, which one hopes that genomics will provide a roadmap to answer.

The field of clinical genetics has changed completely from a more classical approach of family history based thoughtful analysis and prediction to a knowledge-driven result-oriented approach. Being able to treat previously "untreatable" conditions is becoming a reality today.

There is so much work to be done in the next decade to collect data from a large number of individuals for research so we have a greater understanding of gene function and can better predict disease occurrence. This will require teamwork between MDs, basic scientists, statisticians, geneticists, computational, and bioinformaticians. This, along with continuing education provided to physicians by geneticists through a public health program to allow them to keep updated with this fast-moving field. It is an exciting time ahead, but the benefits we reap will depend on our continued thirst for knowledge, correct use of information, and collaboration among experts to move with greater efficiency.

Genomics is in its infancy in India, and we have "miles to go before we sleep…"

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