GeNeDit

# Genetic Literacy: Is the Population Ready?

## Editorial

Health is a topic close to everyone's heart! Health columns in newspapers and magazines and health-related programs on television are very popular. Over the decades, they have played a major role in creating awareness about common health problems and their preventive measures. The importance of a healthy, balanced diet and exercise has been perceived by all and implemented by many in their lifestyle. With the exception of self-claimed 'knowledgeable' laypersons who try to treat common ailments with home remedies, conventionally, the diagnosis and treatment of medical disorders has chiefly been the domain of physicians including some practitioners of various forms of traditional medicine. Now, however, over-the-counter drugs and the internet are at everyone's service and many laypersons search for their diagnosis on the internet before coming to the physician. This may or may not be useful; but we are worried for those who use the internet as a proxy doctor and play with their lives or the lives of their loved ones. Now, they have 'direct-to-consumer' testing at their service. This type of testing is offered for even high-end tests like exome sequencing or whole genome sequencing.

An article in this issue discusses various issues related to whole exome or whole genome sequencing offered to consumers directly. The tests may be offered for identification of risk for multifactorial disorders, cancers and even for paternity testing or ancestry testing, though the latter do not have much market in India at present. But genetic testing to predict the future of one's health is likely to be tempting and has a great appeal for the masses. The takers may increase as the cost is reducing rapidly. Many of the next generation sequencing (NGS) based tests, especially for multifactorial disorders like hypertension, heart disease and Alzheimer disease, are not clinically validated. The psychosocial effects of pre-symptomatic testing are also not interrogated in Indian population. The tests are also offered for carrier screening for recessive monogenic disorders or cancer susceptibility genes. The data about the disease-causing pathogenic variations in Indian population is also different from other populations in the world. Hence, testing using the powerful next generation sequencing technique, needs pretest and post-test counseling and knowledgeable physicians to order and interpret the results.

As these tests are becoming the first-tier testing for phenotypes suggestive of monogenic disorders or genomic disorders and will soon be used for pharmacogenomic testing, all physicians need to be equipped with knowledge of their appropriate applications. So also, laypersons need to be educated about the applications, limitations and other issues like secondary findings. This is because many individuals or their family members may be required to undergo such type of genomic tests. For successful use of such tests, the individual and the family need to be involved in decision-making for ordering the test and return of results. The results may have long term / lifetime implications for the individual and the family members. The time is not far where exome / genome sequencing will become the test of choice for carrier screening for reproductive decisions and newborn screening. At this juncture, when laboratories are advancing rapidly in quantum and quality of such high throughput testing, we need to educate the population about the power and limitations of these tests, whether for patient care, population-based screening or research, especially for exploring genetic etiologies of multifactorial disorders.

In this issue, the Genexpress has articles about the utility of exome sequencing in identifying treatable disorders. This is the future we are hoping for. With advances in the development of therapeutic modalities from drugs acting on pathways and receptors to gene therapy, monogenic disorders may become easily treatable in the not-so-distant future. The notion that most of the genetic disorders are untreatable may soon be forgotten and identification of monogenic disorders will become more important for the patient rather than the family. As exome and genome sequencing

are rapidly becoming common tests offered to a large chunk of population, we need to educate the population about genomic technology and prepare them to be equal partners in decision-making. As this testing has many psychosocial issues, engagement of Indian society is needed to make policies about pretest / posttest counseling, return of secondary findings, etc. Geneticists and physicians need to initiate dialogue with social groups and get them actively involved in understanding the perspective of lay persons so that the powerful technique of next generation testing is used for the benefit of the society without causing harm or undue anxiety. We need a population with more genetic literacy rather than direct-to-consumer testing.

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