Genetic testing in cardiovascular diseases

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It is widely known that cardiovascular diseases (CVD) are one of the leading causes of death. In India, recently the disease burden has been shifting from communicable/infectious diseases to non-communicable diseases, and CVD constitutes a significant percentage. India ranks as one of the countries with a high death rate due to CVD. The prevalence rates among younger adults and women (in the age group of 40 years and above) is also increasing. Premature mortality in terms of years of life lost because of CVD in India increased by 59%, from 23.2 million (1990) to 37 million (2010). Hence, India is dealing with a CVD epidemic and to tackle it, three major concerns need to be addressed: accelerated build-up of cases, the early age of disease onset in the population, and the high case fatality rate.

Current prevention strategies revolve around lifestyle management like smoking, diet, and exercise among others. However, extensive research has shown that CVDs have a sizeable hereditary component. When lifestyle factors add on to hereditary ones, it increases an individual's risk for diseases. Hence there needs to be more emphasis on studying genetic predisposition as a risk factor to get a better understanding of the disease. Genetic predispositions to disease occur due to gene mutations which result in altered biological function of the gene/protein which can result in increasing an individual's risk for disease. These mutations are commonly known as polymorphisms. There are also polymorphisms that indirectly predispose individuals to CVD by rendering them genetically susceptible to other related diseases, such as diabetes. Conversely, positive genetic predispositions also have been discovered, wherein an individual's genetic tendency to have elevated levels of high density lipoprotein (HDL) decrease a person's chances of developing CVD.

Assessing CVD risk is integral to the prevention and treatment of the disease. Several markers, polymorphisms and genes have been identified with the onset for CVD. Disease management options for CVD are available, and can substantially mitigate the burden of CVD, if implemented effectively. The mapping of the human genome has contributed to a better understanding of the genetic causal factors associated with CVD. Genetic testing has been proved an effective tool in estimating predisposition of individuals to several complex diseases and are being widely used worldwide. When genetic testing is done in a family with some affected members, it not only provides prior information about the predisposition of physically unaffected members, but also relieves the genetically unaffected members of unnecessary lab tests and concern.

Genetic testing for CVDs and many other non-communicable diseases are available in India. One of the issues with the use of most of these tests was their design, which reflected the genetic makeup of Caucasian population, since the pioneering studies were done on this population. However, recently ongoing studies on the Indian populations have identified specific variants and genes associated with CVD risk in the Indian population. There is still a need of extensive studies on Indian cohorts/populations to conclusively identify novel genetic markers and validate some known markers causing or protecting against CVD. There is recent interest to take it forward to provide a composite diagnostic chip consisting of these genetic markers and develop a risk score for prediction of CVD. This will lead to the development of more precise and effective treatments and management of the CVD in the future.

The awareness and use of genetic testing is in its growing phase in India. Next generation sequencing has provided high throughput multiplex platforms to perform routine genetic testing in a cost effective manner. However, in order to reach a wider population, the costs need to be moderated further. The emerging epidemic of cardiovascular diseases is a big health challenge. Therefore, improving CVD risk prediction to reduce the mortality rate is an important public health goal.
Considering the huge impact genetic testing might have on public health management of a major health concern like CVD, government funds and policies might come in handy to bring down cost of genetic testing in India. Funding for large projects involving studies on the Indian populations to develop a CVD risk prediction models, creating awareness about early prevention of CVD and the role of genetic testing and providing cost relief to patients needing genetic testing, are some areas which need the attention of policy makers.

