

The era of genetic testing; from imagination to practicability in Pakistan

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Genetic testing offers important insights about an individual's genetic composition, it has emerged as a potent technology that has the potential to completely transform healthcare. This emerging field has potential applications in disease risk assessment, targeted therapies, and personalized medicine. But the quick development of genetic testing also brings up important moral, legal, and societal issues that need to be carefully thought through.¹

The ability of genetic testing to determine a person's susceptibility to specific diseases is among its most important advantages. Healthcare practitioners can learn a great deal about a patient's risk of getting certain ailments by examining their DNA. This information enables them to take preventive steps to reduce the patient's risk. Genetic testing can also help doctors make better treatment options by allowing them to customize medications based on a patient's genetic profile. This can maximize positive outcomes and reduce negative ones.²

The possibility of genetic discrimination is a major concern. Genetic information may be misused by employers, insurers, and other organizations to make discriminatory and privacy-violating judgments about hiring, insurance coverage, or other opportunities. To stop these kinds of discriminatory actions and shield people from genetic discrimination, laws must be passed.

The field of genetic testing is among those that is gradually becoming more significant and well-known in Pakistan's healthcare system. The need for genetic testing to detect, treat, and prevent genetic illnesses and hereditary conditions has surged despite the limited availability of genetic testing services due to technological developments, financial aspects and increased knowledge.

In Pakistan, the field of inherited genetic disorders is one of the main areas where genetic testing is having a big influence. In Pakistan, consanguineous marriages are comparatively

widespread, which may raise the population's incidence of genetic illnesses. Therefore, genetic testing is essential for identifying people and families who may be affected by hereditary diseases. This information enables early intervention, counseling, and family planning.³

Moreover, the use of genetic testing is growing in the diagnosis and treatment of a variety of illnesses, such as neurological disorders, cancer, and cardiovascular diseases. Healthcare practitioners in Pakistan can obtain important insights about disease risk, prognosis, and treatment response by examining an individual's genetic makeup. This allows for more individualized and focused healthcare interventions.

Notwithstanding the possible advantages of genetic testing, there are a number of difficulties in the Pakistani setting. Certain localities may have restricted access to genetic testing services, especially rural ones, and many people may find the cost to be a barrier. Furthermore, the general public, legislators, and healthcare professionals need to be better informed about the benefits of genetic testing.

In Pakistan, ethical issues are particularly significant when it comes to genetic testing. A priority should be given to securing informed consent, safeguarding privacy, and using genetic data responsibly. To protect people's rights and guarantee that moral principles are respected, regulatory frameworks and norms governing genetic testing procedures, data storage, and secrecy must be developed.

The initiatives have been undertaken recently to create genetic testing facilities and increase capacity for genetic counseling and testing services. In order to advance the infrastructure for genetic testing and guarantee its responsible and equitable implementation throughout the nation, cooperation between researchers, legislators, healthcare professionals, and international organizations is vital.⁴

It is imperative to tackle the obstacles and prospects linked to the assimilation of genetic testing into the healthcare system, given its ongoing evolution and growing significance in Pakistan. Pakistan has the opportunity to enhance disease control, promote customized therapy, and enhance the general health of its people by creating a conducive atmosphere for genetic testing. All the geneticists and health care provider community must come forward on one page along with strong proposal to sit with the government bodies and guide them the importance of genetic testing particularly those recommend by the FDA for certain diseases and drug therapies. This will reduces the financial burden in the long run to start personalized medications to the patients rather than hit and trial method.

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