Journal of Global Public Health (JGPH)
Volume 2 Number 1, January–June 2020

Social Implications of Genetic Testing: The Indian Perspective

Ipsa Mohapatra[1], Purushottam Giri[2]

Author's Affiliation: [1]Associate Professor, Department of Community Medicine, Kalinga Institute of Medical Sciences, KIIT University, Bhubaneswar, Odisha, [2]Professor and Head, Department of Community Medicine, IIMSR Medical College, Badnapur, Jalna, Maharashtra, India

Corresponding Author: Purushottam Giri, Professor and Head, Dept. of Community Medicine, IIMSR Medical College, Badnapur, Jalna, Maharashtra, India

E-mail: drpgiri14@gmail.com

How to cite this article:

Mohapatra I, Giri P. Social Implications of Genetic Testing: The Indian Perspective. Journal of Global Public Health. 2020;2(1):33–35.

Dear Sir,

India is the sixth largest country of the world in terms of size, with a population 1.25 billion has the distinction of being the second most populous country of the world; with around 19 million people added every year, the country adds more than any other country annually to the world population growth.[1-2]

There are four main ethno-racial groups in India viz. the Caucasoids, Australoids, Mongoloids, and Negritos. In addition to the complexity and the magnitude of population, the presence of disparity in economical, cultural and infrastructural resources at the level of population subgroups as well as individuals further complicates the access and utilisation of genetic testing services. Almost all known reported genetic disorders have been reported by medical geneticists and clinicians from India. However, this data having been reported from limited institutions and individuals is not representative of the nationwide prevalence and still figures of most disorders is not known.[3]

Genetic tests available have raised serious issues for medicine, public health, and social policy regarding the circumstances under which the test should be used, how the test is to be implemented, and of what use will be the results. Uses of genetic testing can be medical (eg. identify serious disorders in the fetus or infant; conditions or predispositions that signal serious, imminent diseases etc.) and non-medical (eg. DNA testing for law enforcement, DNA fingerprint to identify criminal offenders, identify bodies of deceased soldiers etc).[4]

Issues in genetic testing are numerous - autonomy, confidentiality, privacy and equity. Autonomy, with respect to genetic testing is to provide adequate information, based upon which a person can make a decision whether or not to undergo testing. A proper informed consent, with information on risks, benefits, efficacy and alternatives to the procedure should be informed. Multiplex testing is performing multiple genetic tests on a single sample of genetic material often using techniques of automation. Informed consent should be gained in advance of such multiplex testing. Hence genetic counselling is a major part in assuring autonomy of a participant. Pre- and post-test non-directive counselling should be given by persons who are qualified and experienced in communicating the meaning of genetic information.

Confidentiality encourages the free flow of information between patient and doctor, so that the patient's disease may be adequately treated and is generally protected in the doctor-patient relationship. However, genetic testing may not always occur within a doctor-patient relationship; a screening may occur in the employment setting, a sample itself may be stored (as in DNA banking) for future use. Genetic information being unlike other medical information reveals not only potential disease or other risks to the patient, but also information about potential risks to the person's children and blood relatives. The fact that geneticists may wish to protect third parties from harm by breaching confidentiality and disclosing risks to relatives is still not spoken on. While general principles of counselling require the presence of both

spouses, necessary care and caution must be taken so as not to break families. Truthful counselling with extreme caution and patience is essential to explain the situation in a proper perspective in order to minimize psychosocial harm.[5]

There are several difficulties in applying the public health model for prevention of diseases to genetics. In contrast to infectious disease where there is visible immediate detrimental effect, the transmission of genetic diseases may not pose an immediate threat and devastation to a community, but the transmission of genetic disorders to offspring creates a potential risk for a future generation. Moreover the concept of "prevention" of most genetic diseases does not prevent the disease but rather the birth of an individual with the disease; individuals with religious or other personal moral objections to abortion disapprove of it. Most genetic defects cannot be corrected. Mandatory genetic testing might have devastating effects on the individuals who are tested, by being the basis for discrimination. Genetic disease may thus be percieved by people as an integral part of them. Persons who learn, against their will, that they carry a defective gene may view themselves as defective having a negative impact on self-image; unasked-for revelation that occurs through mandatory genetic testing can be a constant source of worry and tension for the rest of his or her life and can have widespread reverberations in the family, including others who may be at risk or related as partners.

There is a growing concern for ethical issues in human genetics in recent years, with a very narrow gap between routine genetic testing and research raising several ethical, legal and social issues. Regulation of genetic diagnostic services, in India, is presently governed by the ICMR's ethics guidelines for biomedical research. Prenatal diagnostic services are regulated by the Preconception and Prenatal Diagnostics Techniques (Prohibition of Sex Section) Act, 1994. There is often an overlap between genetic research and services for the physician as well as the patient and therefore, adequate safeguards against therapeutic misconception are needed. Genetic manipulations may have known or unknown consequences for the future and therefore, greater caution against potential dangers is necessary. Emerging genetic/genomic technologies cause emergence of newer ethical concerns and issues. Therefore, there is a need for professionals to keep abreast of such advancements and understand their implications. The Ethics Committee reviewing genetic research should have necessary expertise to understand the ethical implications and provide safeguards for research participants. Genetic testing and research often require dealing with persons who are unable to protect their rights and safety and may be vulnerable, such as children, individuals with mental illness, cognitively impaired individuals, people with rare diseases and others.[5]

Common genetic disorders like Hemophilia, Achondroplasia, Huntington disease, Lysosomal storage disorders, and many others have been reported in large numbers from all parts of the country. Information from small scale newborn screening projects, case reports and personal experiences of clinicians reveals that all inborn errors of metabolism are seen in India. The progress over last 2–3 decades have brought state of art molecular diagnostics to India and along with excellent clinical skills in dysmorphology and metabolic disorders; specialty of medical genetics has been well-established in India bringing the greatest benefit to the patients with genetic disorders and their families. However, though a beginning has been made, the research in the area of genetic disorders still lags behind in the areas of understanding pathogenesis and new drug developments. Research in gene therapy and stem cell therapy is almost nonexistent. Research and development in these areas and recombinant products will go a long way in making the new therapies available to Indian patients at affordable costs.

Despite ongoing promising expansions in the field and availability of clinical and diagnostic facilities; the number of geneticists and laboratories are too scarce to cater to the huge population of India. Improved health budget allocations, sensitization of the health authorities and policy makers toward the burden of genetic diseases, inclusion of genetics in medical curriculum and fostering awareness amongst medical practitioners from other fields are important steps required for this purpose. Medical Council of India (MCI) needs to take up the education of modern genetics at all levels of medical curricula; so that the coming generation of medical practitioners is prepared for the era of molecular medicine.

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