Available online at http://www.journalijdr.com

IJDR

International Journal of DEVELOPMENT RESEARCH

ISSN: 2230-9926

*International Journal of Development Research*
*Vol. 06, Issue, 06, pp. 8051-8054, June, 2016*

## *Full Length Review Article*

# THE NEW ERA OF HUMAN GENOMICS AND EMERGING BIOETHICAL ISSUES

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**ARTICLE INFO**

***Article History:***

Received 24th March, 2016
Received in revised form
16th April, 2016
Accepted 21st May, 2016
Published online 30th June, 2016

***Key Words:***

Bioethics,
Human genome,
DNA, Genetic privacy,
Genes, Chromosomes,
ELSI

**ABSTRACT**

The new era of human genetic research has begun with the completion of Human Genome Project in 2003. Consequently, a standard functional human genome map has been produced depicting location, sequence, functions and mode of regulation of approximately one lakh genes of entire human genome. Because of advances in information and communication technology and genomic technologies, DNA sequencing, testing and manipulation are now faster, cheaper, and more accurate as compared to 15 years ago. The research outcomes are increasingly being used to enhance resource capabilities of medical, pharmaceuticals and health care systems. Scientists predict of genomic revolution in coming decade that will have transformational impact on our society through beginning of new disciplines in biomedical sciences such as predictive medicine, prenatal genetic screening, prenatal selection for genetic enhancement, and gene therapies for incurable diseases. Nonetheless, bioethical challenges encountered in applications of genetic research information with respect to genetic privacy, genetic discrimination, genetic spying, genetic manipulation and patenting of genetic information need to be resolved through stringent government regulations to prohibit the misuse of genetic databases of genome of the research subjects (individuals, communities, or populations).

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## INTRODUCTION

Approximately sixty billion cells constitute human body, each of which contains a nucleus harboring twenty three-pairs of thread like structures known as chromosomes, made up of DNA (deoxyribonucleic acid) and proteins. DNA is the genetic material that makes up genes (Watson and Crick, 1953). Estimated one lakh genes or three billion base pairs constitute human genome. The new era of human genetic research begins with the inception of Human Genome Project in 1990 that successfully mapped and sequenced the entire human genome in thirteen years period. However, outcomes of Human Genome Project have raised many bioethical questions regarding the use of human genomic data in human affairs such as employment, health insurance, and court of law. Moreover, the issues of genetic privacy, genetic discrimination and free will versus genetic determinism also got differentiated (ELSI, 1992). Unfortunately, there is no stringent regulation directly addressing above said issues exists so far. The present paper deals with bioethical dilemmas and challenges encountered in generation, control and dissemination of human genetic information. Undoubtedly, there is an urgent need for more government regulation to prohibit the misuse of genetic data (Mc Ewen *et al*., 2012) as well as a need for companies to abide by bioethical standards to ensure the just applications of this information.

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### Basic concept of human genetics and genomic research

Scientists believe that the human genome is essentially the same in all people and that the genetic differences make up about one tenth of a percent of our DNA (0.1%). Each human somatic cell contains twenty three pairs of chromosomes of which a set of 23- chromosomes is contributed by each the mother and father. Thousands of genes are linearly arranged on a chromosome. Biochemically, a gene is deoxyribonucleic acid (DNA), which is characterized by a unique sequence of bases (adenine, cytosine, guanine and thymine). The sequence of bases is arranged in triplets in various orders represent the genetic code. DNA is regarded as the blue print or central dogma of life since it can replicate to make same kind of molecules and transcribe to produce RNA (ribonucleic acid) which may either instruct the cell to make a specific protein or act as regulator in gene expression. Proteins are very important bio-molecules for functioning of our body, some serve as the structural components of cells, tissues, some carry out

chemical reactions (enzymes) while some act as messengers (hormones) and some regulate gene expression as transcription factors. Human genome project (HGP, 1990) was a global effort to determine the location of estimated one lakh genes and to sequence the entire human DNA. The project was started in 1990 and completed in 2003. The goal of this project includes developing an information system for collecting, storing, retrieving, analyzing, interpreting and distributing the large amounts of data generated by the research (Human Genome Consortium, 2001). Consequently, a standard human genome map has been produced depicting location, functions and mode of regulation of genes. The standard human genome map can be used to identify deleterious disease causing genes in individual`s genomic maps. Because of advances in information and communication technology and genomic technologies, it is now much easier to compare and contrast DNA variations among individuals and to identify populations that are susceptible to certain kinds of diseases such as breast cancer and heart disease.

DNA sequencing, testing and manipulation is now faster, cheaper, and more accurate as compared to 15 years ago when human genome project was completed at the cost of 500 million US dollar (Greely, 2015). This kind of information could help the pharmaceutical and medical science facilities to increase resource capabilities. Genetic information obtained through DNA fingerprints is also being used in criminal prosecutions as an ultimate evidence of murder, rape and paternity litigations, just to name a few (Greely *et al*., 2006). However, bioethical challenges encountered in applications of genomic database including predictive medicine or predicting the future, privacy and confidentiality of genetic information, genetic manipulation, Intellectual Property Rights and patenting of genetic information need to be amicably resolved. Government regulations to prohibit the misuse of genomic database by sponsors, researchers, service providers, and companies need to be enacted and implemented in accordance with the bioethical standards.

**Emergence of bioethics as a social institution**

Bioethics is a new coinage in social philosophy that deals with ethical, legal, social and moral questions arising from the applications of biological knowledge to human affairs (Narayan, 2006). Reich (1972) first time officially used the term which now became a global enterprise or movement based on psycho-social, economic, legal and religious issues in the background of ethics (Reich and Thomas, 1994). It aims at providing meaningful resolution to the problems of humanity`s ethical and biological present and future. However, bioethics is not simply application of philosophical notions to scientific problems, it is a dialogic form of deliberation considering social interest and the cultural or religious norms about what is proper, what is good and what is just (Pellegrino, 1990). Current scientific research particularly in genome science and biotechnology such as human genome project, human embryonic stem cell research, and gene therapy along with their ethical, legal and social implications (ELSI) constitute an expanding area of bioethical reflection. Bioethics emphasizes on dialogical nature of decision making based on deliberation. All stakeholders and beneficiaries such as sponsors, researchers, administrators, service providers, research subjects and public at large should partake of the ideas, risks, benefits and outcome of research. In the past 5-10 years, the field of bioethics has grown rapidly and several laws have been enacted globally in relation to applications of the new human genetic research and testing, viz., predicting the future or predictive medicine, prenatal genetic testing, privacy of genetic information, genetic manipulation, genetic discrimination, cloning, eugenics, IPR and patenting of genetic information under its purview.

**Predictive medicine or predicting the future**

Human genomic research has ushered in an era of new human genetics known as predictive medicine. The postnatal human genome map can accurately predict about individual`s future with respect to traits, behavior and risk of diseases (IGVC, 2005). It will have positive impact on our society; a disease that may happen in future could be checked through simply change in the life style and behavior (Mukhopadhyay, 2011). Yet, the majority of the population is forgoing the benefits of new predictions, diagnoses and therapies because they do not have confidence in the privacy of their genes (Privacy and Progress in whole genome sequencing, 2012; Mc Guire, 2013). One of the reasons for this unwillingness and decrease in research subjects includes the fact that researchers refuse to be held responsible for the selling of database systems as well as stolen genetic information from database system breaches. Moreover, genetic test results may pose psychological problems for those tested or affects the patient`s relationships with family members. Prenatal DNA screening of parents, embryos and fetuses can accurately predict of fatal diseases or chromosomal abnormalities in the new born thereby the prospective parents could use that information to avoid marriages or having children (Ethical aspects of human testing, 2000). However, testing of fetuses for genetic conditions has been controversial. Some have opposed it because it often leads to abortion or disabilities in new born which they condemn as murder and unethical respectively. Prenatal selection for genetic enhancement refers to pre-implantation genetic diagnosis for selecting children based on genetic traits (Genetic medicine related societies, 2003). According to Parens, 1998, the idea of using genetic testing to select enhanced genetic traits for offspring has raised many concerns. Some fear that allowing parents to choose the traits of their children would deny those children their right to an open future not chosen by their parents. Others worry that the rich will be able to buy genetic advantages for their offspring, denied to those who conceive in the traditional manner. Still others worry that prenatal selection will lead to homogenization of the human gene pool, as all parents opt for children with similar traits that could lead to a self-perpetuating caste system. Sex selection is a special case of genetic enhancement. In India, though illegal, prenatal sex selection by abortion is common with female fetuses being aborted much more frequently than males. It may have grave social consequences of imbalances in the sex ratios. Moreover, such practice reinforces the subordinate position of women in the society.

**Genetic manipulation**

Genomic information is currently being used to identify genes of interest which can then be inserted or deleted in the genome

to enhance genetic capability. Somatic cell gene therapy is uncontroversial since it aims to correct defective genes in a cell, tissue or organs. However, the success rate is not very promising until 2014. Germ line gene therapy involves introduction of normal human genes into the eggs, sperms, fertilized egg or an early embryo of the offspring. This could be done in order to avoid a genetic disease or in order to enhance a character. However, there has been an informal moratorium in the scientific community on trying such experiments in humans on bioethical grounds (Nelson, 2000). Chimeras result from genetic mixing of genomes of two unrelated species. With respect to human beings, introduction of nonhuman genes into humans could be seen as lessening the humanity of the recipient, which has not been attempted. However, human genes have been successfully introduced and gene products harvested in bacterial or yeast cells. Human insulin, erythropoietin and human growth hormones are being produced through biotechnological means. The idea of introducing human genes into more closely related organisms, such as chimpanzees could provoke more serious concern about blurring the definition of humanity. Creation of artificial gene or genomes, not found in any existing species that would create new or modified proteins. One branch of the biotechnology industry specializes in such efforts, named directed evolution. However, bioethical implications of safety of making new genes, new proteins, new genomes, and finally new organisms have provoked wide debate and discussion.

**Privacy and confidentiality of genetic information**

The huge genetic database can only be stored through electronic means. Which makes it susceptible to misuse by third parties (Genetic Spying)? Cyber risk is always associated with such kind of electronically stored information. DNA information can easily be acquired from objects of contact such as comb, handkerchief, hairs, semen, blood spots, saliva and facial tissues. Since acquisition of DNA information is easier than other medical information, genetic privacy and confidentiality needs to be addressed as a universal right because everyone should have the right to protect and determine who has access to their DNA information. One should have an exclusive genetic privacy right over DNA information since it is the most revealing about our genetic makeup and character than any other kind of technology (Rothstein, 1997). Insurance companies could collect people`s DNA data to decide whom to insure and what to charge them. This would result in a certain proportion of the population that would become uninsurable and discriminated against based on their genetic background something that they have little control over. Genetic screening in the employment is on the rise and could lead to discrimination against people who might be screened with potential risk of diseases (Murray, 1991). However, rejection of some talented professionals screened positive with the risk of certain diseases may adversely affect our economy.

**Intellectual Property Rights and patenting of genetic information**

The issues concerning patentability of genes, genomes and genomic technologies are unresolved till now. Some people have fundamental objections to patents on genes, on human genes or on genetically modified organisms (GMOs), one set of objections, focusing on genes, advocate that they are discoveries, not inventions, therefore should not be patented. While others point out that since genes are creation of nature, evolutionary process or God so, the question of IPR does not arise. However, patents on genes, human genes and GMOs are allowed in Europe, United States and Japan. U.S. Supreme Court, in Diamond vs. Chakrabarty case allowed a patent on oil eating superbug, a genetically altered bacterium *Pseudomonas syringae* (Diamond v. Chakrabarty, 1980). In 2013, U.S. Supreme Court ruled in Association for Molecular Pathology v. Myriad Genetics (Association for Molecular Pathology v. Myriad Genetics, 2013) that naturally occurring genomic sequences could not be patented as compositions of matter because they were products of nature. However, the court ruled that legitimately modified genomic sequences, such as complementary DNA could be patented. As a rule patents expire after twenty years from the date of application filed, hence all patents on human genes will expire by 2021.

**Conclusion**

The new genetics refers to a comprehensive understanding of the biochemical basis of life, the sequence of DNA, and its physiological significance. The Human Genome Project was completed in 2003 at the cost of 500 million US dollar. The project developed an information system for collecting, storing, retrieving, analyzing, interpreting and distributing DNA sequence database. Consequently, a standard human genome map depicting location, functions, and mode of regulation of genes has been produced. The applications of human genomic information include *inter alia*, identification of deleterious disease causing genes; DNA fingerprints for prosecution of criminals; pre and post natal genetic screening for identification of specific traits; prenatal selection of enhanced genetic traits in offspring; genetic manipulation; somatic cell and germ line gene therapies; and establishing forensic, personal, family and ethnic identity (National Research Council, 1996).

Undoubtedly, genomic research outcomes would have profound impact on our society. It would certainly bring new dimensions to our medical and healthcare systems. However, some of the issues related to applications such as genetic privacy, genetic discrimination, genetic spying, genetic manipulation, and patenting of genetic information are controversial. Bioethical implications of such issues need to be amicably resolved and stringent government regulations to prohibit the misuse of genetic data need to be enacted. Nonetheless, medical, pharmaceutical and health care companies need to abide by bioethical standards to ensure the just applications of genomic information. Moreover, in the light of ethical imperatives, it seems well justified to recommend as suggested by Watson, (1990) that there should be 5-7% fund allocation for bioethical studies from each sanctioned research project particularly dealing with the new human genetic or genomic information.

**Acknowledgements**

The author is thankful to the head of the institution, and Dr. A. Kumar, member of the Indian Genome Variation Consortium for valuable discussion on the topic. Timely help rendered by Kirti Narayan and Yash Narayan in preparation of manuscript is highly appreciated.

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