The Promise of Genomics
and Its Ethical Implications

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Introduction

Genomics has to be differentiated from genetics which is the investigation of the roles and functions of single genes. Genes are the DNA sequences that code for a polypeptide that shall ultimately function as structural or regulatory proteins or enzymes through the processes of transcription and translation. In genetics, the focus of study is that of diseases or conditions that are brought about by the expression of single genes, the monogenic disorders which are usually rare.

On the other hand, a genome is the totality of an individual organism's genes. For example, the human genome is made up of 20,000 plus genes . Thus, genomics is the study of all the genes of a cell, or tissue at the level of the DNA (genotype), mRNA (transcriptome), or protein (proteome). It includes the study of interactions between loci and alleles within the genome. In genomics, if a particular gene is studied – it is to find out how it interacts with other genes that impact on a phenotype or characteristic or illness in question.

Genomics has many applications. Human genomics, for example, has wide applications in health, forensics, and population studies especially in

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the identification of ethnic groups. Plant genomics has been shown to be very useful in agriculture, especially in the propagation of selected crops for specific characteristics. Similarly, genomics has been used in animal breeding studies and in the understanding of biodiversity and species interaction.

This presentation shall focus on human genomics and aims to explain some ethical concerns on how the promise of genomics is being touted to the public, to suggest how to surface or identify ethical issues in genomic research or its application and, finally, to recommend some measures that can be undertaken in order to ensure that ethical principles are not violated in the process of "fulfilling the promise" of genomics.

The Promise of Genomics

The 13-year Human Genome Project that was completed in 2003 and coordinated by the U.S. Department of Energy and the National Institutes of Health has not only generated much information on the organization and regulation of genetic information but also spawned technologies for further research and for prediction and diagnosis of diseases. The great support for the Human Genome Project was obtained because of the expectation that mapping the human genome will allow the prevention of common polygenic and multifactorial diseases like neurological diseases (Parkinson's), mental illness (schizophrenia, depression), cardiovascular diseases (coronary heart disease) and cancer through the identification of susceptibility genes or risk genes. The project generated much enthusiasm, because of the possibility of personalized health care through pharmacogenomics and nutrigenomics such that drugs and foods are designed and formulated to address the specific needs of individuals. Genomics was seen as being useful in forensics for the prevention and solution of crimes and that it can help bring about global peace and understanding by clarifying the nature of ethnicity and relatedness of peoples and nations.

Surfacing Ethical Issues

There are, however, concerns that genomic research exacerbates the usual issues on possible violations of the research principles of respect for persons, beneficence, maleficence and justice, because of the uniqueness of

genetic information and the magnitude of resources required in the activities. It is, therefore, important that researchers, ethical review committees and policy makers be guided on how to surface ethical issues arising from genomic research.

I suggest that the following questions be asked whenever a particular research is being considered:

Would the genomic research or application

- challenge our values on human dignity and integrity?
- have the potential to harm individuals or populations or put them at risk? And, would the possible benefits outweigh these harm or risks?
- widen the gap between the rich and the poor, between the developing and the developed countries?
- shift government priorities at the expense of public health?
- discriminate against groups of people?

Is there a conflict of interest ?

Who will use the data? How?

Will the samples be banked? How will privacy be respected and confidentiality of data be protected?

Some Ethical Issues

Most genomic research will not benefit the participant as many genetic diseases do not yet have definitive treatment. Most risks are not yet known but one recognized risk is that genetic research may uncover information about one's lineage that maybe unwanted by concerned individuals, e.g., paternity issues. Also, genomic research has the potential for stigmatization of community groups especially when the research aims to generalize susceptibilities to mental illness, social behaviors, etc. In developing countries, the issue of justice needs to be carefully resolved because of the need to prioritize the research agenda in the light of a low resource environment.

The informed consent process in genomic research and practice is problematic because information on genomics is not that accessible to the ordinary citizens. For example, the Filipino language and various dialects are inadequate for a practical understanding of genomic studies and their applications. The consent obtained cannot, therefore, be effectively "informed".

Testing for susceptibility genes or risks is conceptually difficult. The interpretation of test results is probabilistic and cannot be highly predictive of future health. There is a lack of clear understanding and guidelines on how to use the information from many of these tests. Moreover, satisfactory treatment interventions that can address the diagnosed genetic disorders are still wanting. Genetic counselors are very few and most deficient in developing countries. How can the benefits be appreciated in situations when there is lack of genetic counseling services? One possible harm is to cause inappropriate anxiety. Further, because of the familial implications of gentic information, should not informed consent include the rest of the family members?

Testing for susceptibility genes/risk genes has also brought to fore the fear of eugenics, and the possible justification for abortions and the curtailment of the reproductive freedom of women.

One worrisome practice is that of direct marketing of genetic tests to the public. The promotional strategies can take advantage of the uninformed public. The public is unprotected when no one is checking on the possible existence of a conflict of interest like when the service provider has proprietary claims on the test.

There is tension between personalized medicine and public health care because of the need for the appropriate allocation of resources and the application of the principle of equity in health. The enjoyment of the benefits of medical genomics will be expensive and has the potential to worsen the gap in health care between the rich and the poor, and at the international level, between the developed and the developing country. On the other hand, Prof. Peter Sy (in a personal communication) pointed out that, in fact, health care should really be personal to be effective and responsive. Each individual is best treated in a manner that takes into consideration his/her special circumstances, e.g., family relationships, food habits, coping mechanisms, and social environment, including economic situation and religious beliefs! Besides, he added, there

is not much motivation in designing and manufacturing drugs that are highly individualized, because market size is an important factor in considering profitability. It would seem, therefore, that what is needed is a clarification of how to operationalize personalized medicine as a promise of genomics.

Biobanking is the organized collection of biological samples and data associated with them. The biological samples include left-over samples from patient care or previous study, DNA sequences, tissue and blood samples and patient data. Presumed consent or a blanket consent (where patient permission is given for any future use of his/her biological sample/data) are frowned upon based on the principle of respect for persons. There are also no clear policies on ownership and transfer of samples. Biobanking needs a closer examination in terms of protection of the privacy of individuals and confidentiality of health information.

Recommendations:

In order to better prepare people in taking advantage of the "promise of genomics" and to ensure that there is equity in the applications of genomic medicine, governments must consider the following strategies:

1. Improvement of education in genetics and ethics.
2. Development of the national and local languages in the sciences for a practical understanding of the nuances of genetics and genomics.
3. Promotion of ethical conversations between scientists, policy makers and the public on the public health implications of genomics, including issues in resource allocation and intellectual property rights.
4. Establishment of ethical oversight structures for research.
5. Location of genomics in the National Research Agenda.■

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