

EDITORIAL

Genetics, Epidemiology and Ethics.

Shaikha Salim Al Arrayed, MBChB, DHCG, PhD*

The human genome project is a \$3 billion global project. The goal is to map and sequence 100,000 genes that make up each individual. This project is already transforming health care regardless of specialty.

Genetic factors contribute to nearly every common disease. New genes linked to specific conditions are discovered and reported every day. We are bombarded by this genetic information through the media, the Internet, support groups and advertising agencies.

Worldwide, according to world health organization, about 5% of children are born with hereditary disorders, and 40% of adults are genetically predisposed to common diseases during their lifetime¹.

In developed countries congenital and genetic disorders account for 25% of deaths under the age of one. An estimate of 250 million people, or 4.5% of the world population, carry a potentially pathological haemoglobinopathy gene. Prevalence varies from under 0.1 births per thousand in some parts of the world to more than 20 births per thousand in parts of Africa and Asia².

More than 9,000 single gene conditions have been identified. Some are more common in certain ethnic groups or geographic areas due to certain demographic and cultural factors, such as maternal age group or the prevalence of cousin marriages.

By implementing the new genetic approaches, it is expected to reduce the mortality and disability caused by genetic diseases. About 50% of congenital abnormalities, 10% of inherited diseases and 2% of chromosomal disorders can be treated or prevented³.

Among the new techniques described will be Gene therapy- the introduction of a gene sequence into a cell with the aim of modifying the cell behavior, either to correct a genetic mutation, or to destroy a cell, or to modify susceptibility to diseases⁴.

Before the end of the century genetic screening and counseling will become major components of both public health and individual medical care. This includes prenatal diagnosis, newborn screening, carrier screening, forensic screening and susceptibility screening.

Soon, we will be able to predict a patient's risk for a disease by using genetic information encoded on a small and

inexpensive chip. This new information is forcing new fundamental changes in our view about health and disease, and in our practice of medicine.

Now we are asking, what is the disease? and how can we treat it? Soon, we will be asking why the disease occurs in an individual at a specific time? How health can be restored in that individual? Answering these questions can help to maintain health and prevent diseases. This can produce a change in medical thinking.

The patient will be viewed not as a case, but as an individual who because of specific genetic predisposition, environmental and development causes has developed a disease. As it is known now, that all genetic diseases have a genetic base that involve many genes interacting together, and interacting with the environment.

Controversies arise over issues such the ethics of genetic testing, gene therapy and cloning. All physicians should be aware of these ethical problems, and the damage that can be caused by misusing or misunderstanding the genetic information^{2,3}.

Human dignity and well being are at the center of these ethical, legal and social issues. It is recommended now that genetic services should be available to all patients. Adequate information should be ensured before testing. Appropriate, non-directive counseling should be offered. Equality of access should be provided, respecting the self-determination of those tested.

Genetic data must not be used to stigmatize or discriminate. It should be used for the advantage of the patients and family. Information should be kept confidential and should not be given to a third person such as insurers or employers, without the consent of the person tested¹.

At this stage, all physicians need to understand genes and genes interactions in order to manage and counsel patients with common disorders such as cancer, cardiovascular disease, hypertension, asthma, diabetes, and rheumatoid arthritis etc.,

For this reason, integration of the new genetic knowledge into routine practice is needed. Unfortunately, few are prepared for this revolution, and current educational efforts are inadequate in this respect. This is why some universities in USA are developing new genetic curriculum. This is to

* Consultant clinical geneticist
Genetic Unit
Salmaniya Medical Complex
State of Bahrain

combine teaching of genetics, epidemiology and ethics in one curriculum.

These new ways of thinking about illness need to be incorporated into all medical education for future physicians. Continuing educational programs are essential for all health care personnel. We need to prepare the future physicians and health care personnel to meet both the present and the future challenges of new genetics⁵.

REFERENCES

1. World Health Organization. Ethics in genetic counseling, Community control of genetic and congenital disorders, Emero technical publication series 24. Regional office for the east Mediterranean: WHO, 1997:108.

2. World Health Organization. Role of education in the control of genetic disorders. Community control of genetic and congenital disorders, Emero technical publication series 24. Regional office for the east Mediterranean: WHO, 1997:160-172.
3. The human Genome project, National reference center for bioethics literature, The Joseph and Rose Kennedy institute of ethics 1998;1-22.
HYPERLINK <http://www.georgetown.edu/research/ncbl/>
4. Human gene therapy, National reference center for bioethics literature, The Joseph and Rose Kennedy institute of ethics 1998;1-22.
HYPERLINK <http://www.georgetown.edu/research/ncbl/>
5. Report from the American society of human genetics information and education committee. Medical school core curriculum in genetics. Am J Hum Genet 1995;56:535-7.