Genomics in health and disease

Rabah M. Shawky[1] and Sahar M. Nour El Din[2]

[1]Pediatrics Department, Ain Shams University, [2]Medical Genetics Center, Ain Shams University

ABSTRACT

Genomics is the study of all person's genes including interactions of those genes with each other and person's environment. Many Factors contribute to human health and disease. Our environment and our biology are two factors that strongly influence our health. For along time, it was believed that disease resulted entirely from our environment or entirely from our biology. Now we are seeing that many human diseases are a result of a complex interaction between our biology and our environment and many other factors. The completion of the Human Genome Project signaled that the genome revolution was here to stay and symbolized its promise that knowing the DNA sequence of our genome and those of hundreds of other organisms would allow us to take on the greatest challenges of human health and alleviate human suffering. The Aim of this review is to discuss the influence of genomics on global health and genetics susceptibility to disease.

Key Words:

Genomics, genetics, gene regulation, mutation, genetic testing.

Corresponding Author:

Rabah M. Shawky
E-mail: shawkyrabah@yahoo.com

What is Genetics and Genomics?

Genetics is a term that refers to the study of genes and their role in inheritance. Gene influence traits such as hair and eye color as well as health and disease development. Genetics determines much (but not all) of a person's appearance and health status, but environmental differences also play a part. Examples of single gene disorders that would be considered as "Genetics" include cystic fibrosis and PKU (Phenyl ketonuria).[1]

Genomics is a relatively new term that describes the study of all of a person's genes including interactions of those genes with each other and person's environment. Genomics involves the scientific study of complex diseases such as heart disease, asthma, diabetes and cancer because they are caused more by a combination of genetic and environmental factors.[2] Understanding the relationships among these factors at the population level may provides new opportunities for prevention and intervention.[3]

Why is genetics and genomics important to our health?

Genetics and genomics both play a role in health and disease. Genetics helps in-

dividuals and families learn about how conditions, such as sickle cell anemia, are inherited in families; what screening and testing is available and in for some genetic conditions treatment.[4]

Genomics is helping to discover why some people get sick from certain infections, environmental factors and behaviors, while others do not. Genomics holds the key to these differences.[5] All human beings are 99.9% percent identical in their genetic makeup. Differences in the remaining 0.1% hold important clues about the causes of diseases. Having a better understanding of the interactions between genes and the environment is helping us find better ways to improve health and prevent disease.[6]

Why is genetics and genomics important to our family's health?

Understanding more about single gene disorders (Genetics) and complex diseases (Genomics) can lead to ealier diagnosis, interventions and targeted treatments. A person's health is influenced by his/her family history and shared environmental factors. Family history is an important personalized tool that captures many of the gene/environment interactions, for conditions that are genetic and genomic in origin.[7] The family history can serve as the cornerstone for learning about genetic and genomic conditions in family and individual disease prevention.[8]

Impact of gene regulation on health and disease

Looking at single inherited variations in the human genome, the researchers try to gain new insights into gene regulation and their impact on health and disease. They are especially interested in the development of regulation throughout millions of years of human evolution. To do this, they combined two methods in genome research-the research for single nucleotide polymorphisms (SNPS), which can influence the correct expression of genes, with population genetics. This strategy enables the researchers not only to look at gene regulation, but also to detect those variations in the genome which can cause human disease[9]. Gene regulation is a basic process which regulates human development and is important for health and disease. Genes are sections of the hereditary molecule DNA which contain the blue print for proteins, the building blocks and molecular machineries of life[10]. The DNA molecule is made up of 3,2 billion nucleotides. However, each human individual has a different sequence of nucleotides. One individual, for example, can have an Adenin (A) nucleotide, whereas, at the same position, another individual has cytosine (C) nucleotide[11]. Researchers estimate that, the human genome has eleven million of such single nucleotide variants polymorphisms (SNPs). Their aim is to detect these SNPs and find out their function in health and disease. The key to human variation is mutation[12]. A mutation is simply a change in the genetic information. Our genome consists of more than billion nucleotides that can acquire a mutation at any time throughout our lives. Every humanbeing carries several mutations in their genome. Most of these mutations are "Silent" and harmless and never have an effect on our health. Others may make us more susceptible to some diseases and whether or not we develop those diseases depends on many other factors, including the rest of our genes and

our environment. Still others can predispose us to disease that may develop later in life, which is also influenced by other factors. Finally, some mutations result directly in disease if they interrupt an essential function in our body.[13]

Genomics and disease:

Many factors contribute to human health and disease. Our environment and biology are two factors that strongly influence our health. For a long time, it was believed that disease resulted entirely from our environment such as tuberculosis or entirely from our biology, such as an inherited disease like cystic fibrosis. Now, we are seeing that many human diseases are a result of complex interaction between our biology and environment and many other factors.[14]

So Genomics is a powerful tool for understanding the totality of the factors that contribute to health and disease, i.e. genetic and environmental interactions.[15]

Translating genomics into usable information:

Three major efforts are underway to translate genomics into usable information for improving population health:[16]

1. Integrating genomics into public health Investigations (PHIS). (Therefore we can define public health genomics as the inclusion and applicaton of genomic information and tools in public health planning, policy and programs).

2. Analyzing genetic variation among specimens in the National health and Nutrition Examination Survey (NHANES) lllDNA Bank.

3. Expanding the Human Genome Epidemiology Network (HUGE NetTM).

Thus genomics should be considered in every fact of public health: infectious disease, chronic disease, occupational health, environmental health, in addition to maternal and child health.[17]

Genetics, genomics and patient management:

The influence of genomics on global health certainly extends well beyond our ability to better diagnose, prevent and treat human disease. The 2002 World Health Organization report on Genomics and Global Health, while affirming the importance of genomics, also warned of a "Genomic divide"-poor countries may be unable to share the benefits of genomic research-thus further exacerbating the inequities in global health.[18] New genetic and genomic research discoveries are ushering in a new era of health care-personalized medicine. Personalized medicine has the potential to transform health care by associating genotypes with phenotypes, through improved diagnostic methods, early detection of predisposing genetic variation (Disease susceptibility), more effective prevention and treatment of disease and avoidance of drug side effect.[19]

Genetic testing:

The term "Genetic testing" cover an array of techniques including analysis of human DNA, RNA or protein. Genetic tests are used as a health care tool to

detect gene variants associated with a specific disease or condition, as well as for non-clinical uses such as paternity testing and forensics. In the clinical setting, genetic tests can be performed to confirm a suspected diagnosis, to predict the possibility of future illness, to detect the presence of a carrier state in unaffected individuals and to predict response to therapy. They are also performed to screen fetuses; newborns or embryos used in-vitro fertilization for genetic defects.[20]

Ethical, Legal and social implications: (ELSI)

Today's genomics research and applications rest on more than a decade of valuable investigation into their ethical, legal and social implications. As the application of genomics to health increase along with its social impact, it becomes more important to expand on this work.[21]

Ethical, Legal and Social Issues include:

1. ***Fairness in the use of genetic information*** by insurers, employers, courts, schools, adoption agencies and the military, among others.

2. ***Privacy and confidentiality*** of genetic information.

3. ***Psychological impact and Stigmatization*** due to individual's genetic differences.

4. ***Reproductive issues including*** adequate informed consent for complex and potentially controversial procedures, use of genetic information in reproductive decision making and reproductive rights.

5. ***Clinical issues*** including the education of doctors and other health service providers, patients and the general public in genetic capabilities, scientific limitations and social risks; and implementation of standards and quality-control measures in testing procedures.[22]

6. ***Uncertainties*** associated with gene tests for susceptibilities and complex conditions linked to multiple genes and gene–environment interactions.

7. ***Conceptual and philosophical implications*** regarding human responsibility and concepts of health and disease.

8. ***Health and environmental issues*** concerning genetically modified and microbes.

9. ***Commercialization of Products*** including property rights and accessibility of data and materials.[23]

Finally we can say that genomics will make a world of difference.

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