Chapter 16
Genetic Nomenclature and Quantitative Techniques for Modern Genome Epidemiologists, Clinicians, Educators and other Behavioral Scientists

ABSTRACT

This chapter explored the relevance of understanding genetic nomenclature in the age of genomic science. There are several aberrant gene chromosomes which serve as the underlying causes of several organic and neurological diseases. Modern genome epidemiologists use specific gold standard for determining the efficacy of a given test which consists of sensitivity and specificity of a test, the positive and negative predictive indices. The risk factors associated with Alzheimer's disease were explained. The gene chromosomes associated with various genetic diseases were compiled as well as the timeline for various scientific accomplishments in the annals of biological research initiatives

PART I: GENETIC NOMENCLATURE IN GENOMIC SCIENCE

After the International Human Genome Sequencing Consortium successfully sequenced the human genome, more than ever before, biology which was previously the mere pursuit of expedition, observation and basic experimentation became a significant scientific discipline. The first startling observation was the extent to which molecular revolution spurred by discovery of DNA in 1953 changed our assumption about biology. In fact, biological science became more quantitative, authentic in assisting scientists to discover the mysteries which underlie human evolution and the trajectory and the incipient onset of diseases. Biological scienctists, were now trained to sequence the possible gene chromosomes which serve as the etiological agent of diseases. In fact, gene-based innovative pharmacogenomic intervention

DOI: 10.4018/978-1-4666-8559-8.ch016

now guides more precise diagnosis, coupled with targeted and personalized patient treatment. Hitherto physicians were perceived as the third leading cause of patients' death, when even correctly prescribed medications easily led to the death of patients. Then, the mode of trial and error treatment-modality created imperfect specificity and the clinical complications of patients' ill-health. Realistically, genomic medicine can restore patients' confidence in the health care system. To become an effective genomic medicine practitioner, medical school curriculum must include not only Mendelian's disorders, dsymorphology, chromosomal disorders, inherited metabolic diseases, and the multifactorial basis of complex diseases such as cardiovascular diseases, but also additional sciences such as epidemiology, genomics, pharmacogenetics, bioinformatics, ethics and phenomenology(Ebomoyi, 2010).

Between 1960-2000, biologists were merely guessing the precise number of gene chromosomes in humans. We have guessed between 20,000- 25000 gene chromosomes. Today a more precise estimate is 40,000 and even now one-half of these genes have specific functions which have not been discovered. Besides, the reported genes which serve as the underlying causes of diseases and death are characterized in extortive genetic nomenclature. Physicians may hesitate to communicate using this genomic language with the lay patients, therefore the patients may be referred to the compassionate and ingenious nurse who has the mystique of getting into the soul of her patients very congenially. As spinoff benefits of the HGSP, many of the common diseases described with their genetic nomenclature, and the offending genes are kept under public domain at the United States' Agency for Energy at Oak Ridge, Tennessee.

Location of a Gene by the Geneticist

Bear in mind, your primary care physician may not be a geneticist. Your cardiologist may not be a geneticist, neither is your urologist who may not have received the requisite training in genetics. Scientifically, a geneticist uses maps to characterize the location of a specific gene on a chromosome. In one type of map, the cytogenetic location is used to describe a specific gene location. Most geneticists agree that cytogenetic location is based on a distinctive pattern of bands which are created when chromosomes are stained with certain chemicals. The other of map adopts the molecular location which involves a precise description of gene's position on a chromosome. This latter molecular approach is based on the sequence of DNA building blocks which make up the chromosome. A geneticist may use a standardize technique of describing gene's cytogenetic location. In genetic science, the location identifies the position of a particular band on a stained chromosome such as 17q12 or 17q12-q21 if less is known about the exact location.

A very lucid example is The CFTR gene, which provides instructions for making a protein called the cystic fibrosis trans-membrane conductance regulator (CFTR) gene (Figure 1). This protein functions as a channel across the membrane of cells that release mucus, sweat, saliva, tears, and digestive enzymes whenever the patient is seriously ill. The CFTR gene is illustrated as being positioned on the long arm of chromosome 7 at the exact position 7q31.2

CFTR Gene Location

After the accomplishment of HGSP by March 25, 2003, by the International Human Genome Sequencing Consortium (IHGSC) determined the sequence of base pairs for each human chromosome. This list of the gene chromosomes and the diseases involved as shown in Table 1. This sequence data provide researchers more specific address than the cytogenetic location used previously for many genes. A