EDITORIAL

GENOMIC MEDICINE

Until the twilight of the 20th century, genetics was a branch of medicine applied to diseases of rare occurrence. The advent of the human genome sequence and the possibility of studying it at affordable costs for patients and healthcare institutions, has permitted its application in high-priority diseases like cancer, cardiovascular disease, diabetes, and Alzheimer's, among others.

There is great potential in predictive and preventive medicine, through studying polymorphic genetic variants associated to risks for different diseases. Currently, clinical laboratories offer studies of over 30,000 variants associated with susceptibilities, to which individuals can access without much difficulty because a medical prescription is not required. These exams permit conducting a specific plan of preventive medicine. For example, upon the possibility of finding a deleterious mutation in the BRCA1 and BRCA2 genes, the patient can prevent the breast cancer by mastectomy or chemoprophylaxis and in the presence of polymorphisms associated to cardiovascular risk preventive action may be undertaken through changes in life style (diet, exercise, etc.).

Legal aspects are also present in this new conception of medicine. For example, currently there is legislation for medications to indicate on their labels the different responses such medication can offer regarding the genetic variants of the patients, given that similar doses may provoke adverse reactions in an individual, while for another such dosage may be insufficient. This scenario would allow verifying the polymorphisms of drug response prior to administering medications like anticoagulants, hyperlipidemia treatments, or chemotherapy, among others.

We must specially mention recessive diseases, produced by the presence of two alleles of a mutated gene, which are inherited from the mother, as well as the father. By studying the mutations, we may learn if a couple is at risk of bearing children with the disease; thus, conducts may be undertaken like selection of embryos or opting for adoption.

Although genetic studies of molecular profiles are promissory, there are questions on how these are requested and how the results are communicated to the patients, their clinical use, application of the pre-symptomatic diagnosis – especially in children, the bad intended use of this information by medical insurance companies considering pre-existing conditions, and finally, the great amount of information and its application in populations where prior studies of association have not been conducted may be cause for concern.

In developed nations, these types of exams are requested without medical prescription in pharmacies that mail the results directly to the interested party. It is worrisome what can happen to patients upon receiving this information; for example, if the report indicates susceptibility to Alzheimer's or cancer, without medical support to identify the presentation risks and the possibility of transmitting the disease to their offspring. This suggests the validity, more than ever, of genetic counseling when delivering analysis results.

On the other hand, insurance companies must consider molecular genetic profiles as a preventive tool that permits improving service quality while lowering costs; for instance, in cancer cases where prophylaxis measures are notably less costly than treatment.

Another aspect to bear in mind is that association studies of genetic polymorphism and disease need thousands of cases and controls, making them very costly for implementation in Colombia. For this reason, the exams include the ancestral report for the purpose of interpreting the results according to ethnic origin. For now, this is the most adequate conduct considering that it will take a long time before validation studies can be conducted.

As with all great progress in medicine, genomics opens a path that will bring enormous benefit inasmuch as the criteria for good medical practices are considered.

Ignacio Briceño Balcázar, MD, PhD
Professor, Universidad de la Sabana
Full Professor, Pontificia Universidad Javeriana

