Rev Med Hosp Gen Méx. 2017;80(2):71–72

REVISTA MÉDICA DEL
HOSPITAL GENERAL
DE MÉXICO
www.elsevier.es/hgmx

EDITORIAL

Something about genetics

Acerca de la genética

Genetics studies inheritance and everything about it. Human genetics has made substantial progress in recent years. The series of molecular tools discovered have made possible to know, in a precise way, the causes of a great variety of diseases, especially of hereditary origin. The beginning of genetics can be defined in 1865 when the laws of Mendel are published. In these laws, the elementary rules on the transmission of traits from generation to generation are established. In 1900, the laws of Mendel are taken back to the humans realizing the first studies in Drosophila. In 1909, Wilhelm Johannsen coined the term gene and in 1905 William Bateson of genetic.

In 1956, Tjio identifies the correct number of chromosomes in the human species, being 46 instead of 48 as initially proposed. In 1959 the French, Jérôme Lejeune, described the cause of the first chromosomal disease, Down syndrome.

By the 1940's, Rosalind Franklin describes the DNA forms A and B and recalling Franklin's data and Chargaff's bases, Watson and Crick describe in 1953 the double-helix structure of DNA.

But it is not until the 1980's that Kary Mullis develops the polymerase chain reaction (PCR), a technique that involves the amplification of specific DNA sequences; this tool revolutionizes the molecular methodology.

In 1986, it is described the first gene causing a human disease, chronic granulomatous disease, and in the early 90's, the Human Genome Project began, and the human DNA sequence is completed in 2003.

The development of molecular technology has allowed a breakthrough in medicine for diagnosis, prognosis and treatment of some diseases. One of the most important tools is the Polymerase Chain Reaction (PCR). Through PCR we can: detect DNA mutations for the diagnosis of hereditary diseases and carriers of these diseases, establish prenatal diagnosis from amniocentesis and preimplantation genetic diagnosis, genotype infectious agents such as HIV or hepatitis, or personalize therapies in patients with certain types of cancer, among other things.

Derived from PCR we have the MLPA (Multiplex ligation-dependent probe amplification) technique which has a diversity of applications as identification of mutations or single nucleotide polymorphisms, study of DNA methylation, relative mRNA quantification, characterization of duplications and deletions of genes, detection of gene predisposition in human cancer such as BRCA1, BRCA2, hMLH1 and hMSH2. The advantage of this technique is its relatively low cost.

Recently, next-generation sequencing (NGS: Next-Generation sequencing) is used for molecular diagnostics in the detection of pathogens, polymorphisms, diagnosis of cancer, etc. In general, through NGS, DNA can be sequenced in hours, exceeding traditional methods that take days (Sanger). NGS is used for the diagnosis of genetic, infectious diseases and cancer. One difficulty is the large amount of information generated that has to be analyzed through specialized software, in addition to the cost it represents.

DNA ''microarrays'' allow to automate the process of diagnosis and genotyping; it consists of a group of DNA probes attached to a solid base to which complementary fragments of DNA present in the sample to be analyzed are joined. On each platform, hundreds or thousands of DNA polymorphisms can be identified in one day.

The Genetic Service of the General Hospital of Mexico, Eduardo Liceaga, is at the forefront of most of these techniques offering molecular diagnosis to a large number of patients with different hereditary diseases as well as establishing the prenatal diagnosis within a few weeks of pregnancy, it represents a calm for the couple who are waiting for the normal birth of their child.

The molecular methodology advances and it is expected that in a few years we can correctly identify risk populations, especially for diseases that represent a major health problem such as diabetes, obesity, hypertension and cancer.

http://dx.doi.org/10.1016/j.hgmx.2017.03.002

S.A. Cuevas-Covarrubias[a,b,*]
[a] *Servicio de Genética, Hospital General de México, Mexico City, Mexico*
[b] *Facultad de Medicina, Universidad Nacional Autónoma de México, Mexico City, Mexico*

[*] Correspondence to: Dr. Balmis 148, Col. Doctores, C.P. 06726, Mexico City, Mexico.
Tel.: +52 5 2789 2000x1278/1279.
E-mail addresses: sergiocuevasunam@gmail.com, sercuevas@yahoo.com, sergioa@servidor.unam.mx