10. Pharmacogenetics and pharmacogenomics

François Noël, November 2017

Thirteen years after the creation of the national pharmacogenetics network in Brazil (www.refargen.org.br), it seemed timely to reassess the use of the terms *Pharmacogenetics* and *Pharmacogenomics*.

PHARMACOGENETICS, a term coined by Friedrich Vogel in 1959, can be considered the "*science that examines the genetic basis of individual variability observed in therapeutic responses to pharmacological treatments*" (Shi et al., 2001). This discipline was definitively established with the publication of the first book on the subject by Werner Kalow in 1962 (*Pharmacogenetics – Heredity and the Response to Drugs*). The evolution of this field has enabled its application in clinical practice as an important component of individualized medicine. Indeed, 13 years have already passed since the FDA approved the first pharmacogenetic test (the AmpliChip CYP450 test, Roche®, for the detection of CYP2D6 and CYP2C19 alleles).

More recently, the term **PHARMACOGENOMICS** was introduced, partly due to the trend of adding the "-omics" suffix to different areas of research, as well as the enthusiasm generated by the Human Genome Project, completed in 2003. This term is considered broader than "pharmacogenetics" by some researchers, as it encompasses the study of all genes in the genome that may determine drug response. Similarly, Meyer (2004) considered that the convergence of advances in pharmacogenetics and the rapid development of human genomics led to the evolution of pharmacogenetics into pharmacogenomics, a more comprehensive discipline that "*is also devoted to the use of the human genome in the discovery of new therapeutic targets*". According to the NIH, "*Pharmacogenomics is the study of how genes affect a person's response to drugs. This field combines pharmacology (the science of drugs) and genomics (the study of genes and their functions) to develop effective, safe medications that can be prescribed based on a person's genetic makeup*".

It should be noted that there is controversy regarding whether or not a distinction should be made between pharmacogenetics and pharmacogenomics, which some consider arbitrary, and that the two terms are currently often used interchangeably. It is worth mentioning two initiatives that highlight the importance of this topic in Pharmacology:

- The Clinical Pharmacogenetics Implementation Consortium (CPIC) is a consortium that publishes guidelines for the use of pharmacogenetic data in drug prescribing (Relling & Klein, 2011).

- The FDA regularly updates a table listing all drugs for which pharmacogenomic biomarker information is included in the package insert; as of November 1, 2017, there were already more than 197 such drugs .

Finally, it is worth noting that the Brazilian Pharmacogenetics Network (REFARGEN), established in 2003, is composed of researchers distributed across the five regions of the country and has been coordinated since its inception by a member of the SBFTE (Guilherme Suarez-Kurtz). REFARGEN aims to create a pharmacogenomic database for the Brazilian population, promote scientific interaction among network members, and encourage research on drugs tailored to the genetics of the Brazilian population, considered one of the most highly admixed in the world (Suarez-Kurtz, 2005).

References

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