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Genetics: an essential tool for the pediatrician of the 21st century

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The impact of genetics on medicine, which had not been clearly understood when Watson and Crick described the DNA structure in 1953, became increasingly more evident throughout the following 50 years due to several landmarks that have been established in this field: mainly the discovery of the genetic code, the creation of techniques that allow for the manipulation of genetic material and the decoding of the human genome sequence, which was completed in 2003.

A significant amount of information has been progressively made available, and the great majority of medical journals have been filled with articles approaching the genetic aspects of diseases and/or using genetic tools to understand, diagnose or treat these diseases, with direct impact on the clinical physician's activity.

Understanding this new information became a necessity not only for geneticists but also for the general pediatrician, since it is time for this "genetic revolution" to have an impact on the diagnosis process being implemented, the therapeutic measures being taken and the counseling being provided to the patient's family.

Another aspect that is closely related to the impact of the development of genetics on the medical practice, and mainly on the pediatrician's practice, is the relevance of the information obtained from a patient for the whole family, which raises new ethical issues that need to be discussed.

This supplement is not intended to approach the whole set of over 5,000 genetic diseases currently known, but its objective is to provide pediatricians with some important examples of the consequences of the new genetic technologies for the clinical practice, presenting a summarized overview of the recent changes that are affecting the manner in which the diseases are faced due to genetic advances.

We selected topics that are constantly present in the pediatrician's clinical practice and authors with broad experience in these topics. The texts have been written not only to provide pediatricians with updated information on genetic advances but also to provide guidelines on what is already possible to use in the medical daily routine.

Enjoy!

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