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## **Forensic Genetics: From the laboratory to the courts**

### **Book Review**

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#### **DESCRIPTION**

Forensic Genetics is an exciting scientific discipline in constant evolution. Traditionally it has been considered as a subspecialty of Genetics and Legal Medicine. However, in recent years it has acquired an entity of its own; Its evolution is linked to the technological revolution of the last decades.

Nowadays, it has consolidated as a tool of undoubted usefulness in the daily work of the Courts of Justice, responding to the challenges that arise from the judicial scope. Since the introduction of the genetic fingerprint in 1985 by Alec Jeffreys, there has been a continuous evolution in the type of markers and in the technologies used. However, as Professor Ángel Carracedo already pointed out, "the priority challenges of forensic genetics are not essentially technological: the statistical evaluation of DNA testing in complex cases (particularly in mixtures or contact samples), communication the value of the test, quality control, the future of R & D, training, ethical standards, among others, are problems that we have to face urgently "[1].

But in addition, we must face other challenges that arise in a globalized society, where the media play a role of great importance. In this way, television series such as CSI, NCIS, Law and Order or Criminal Minds, which have so much impact and audience, arouse concerns in viewers and have managed to socialize this discipline of applied science, to the extent that the receiver of these series is normalizing concepts and knowledge (such as DNA, sequencing, CODIS database), previously restricted to specialists working in the field of Forensic Genetics.

If we add some media cases (such as that of OJ Simpson in the USA, Amanda Knox in Italy, Eva Blanco in Spain), the need for clear and fluid communication with different interlocutors is clear, not only with the judicial collectives (magistrates, prosecutors judges, or lawyers), but also with society in general, since the introduction of juries in the legal framework of some countries, will have their effect on the communication of expertise. In this framework, some initiatives are already being developed, such as "Sense about Science" [2], whose English-language guide for the general public is already being translated into other languages, including Spanish.

On the other hand, it must be borne in mind that, in a majority way, scientific texts, including those related to Forensic Genetics, are written in English. It is evident that for a greater diffusion of science, generated in any country, it is necessary to use that extended language. Regardless of the scientific articles published in impact journals (such as: Forensic Science International: Genetics. Journal of Forensic Sciences, Science & Justice, Journal of Forensic and Legal Medicine), there are major reference works in the field of Forensic Genetics, all they published in English [3]. However, more and more texts in Spanish are being demanded.

This book aims to contribute a small grain of sand to the knowledge of Forensic Genetics, and in particular, to the knowledge of Forensic Genetics in Spanish. There are few books in this area published in Spanish, despite having one of the most important working groups within the International Society of Forensic Genetics (ISFG), the Spanish and Portuguese Speaking Group (GHEP-ISFG), which includes more than 150 laboratories belonging to 23 Spanish and Portuguese speaking countries, with excellent professionals and remarkable scientific productivity. We insist that, without any kind of pretension, with this book we pursue, if not fill a gap, yes at least contribute to fill it.

We believe this text is quite complete; it covers all the major issues that correspond to the Forensic Genetics. We are very aware of its temporality, as we have indicated, due to the constant evolution of technologies within forensic genetics, and to the revolutions that are expected in the next 5-10 years. You just have to take a look at the main scientific journals in this area of knowledge. However, we also believe that the content of this book constitutes a robust conceptual basis (in Spanish), which may be useful for future (and necessary) updates and revisions.

# Presentation and justification of the structure and content of this book

This work aims to address the current situation of forensic genetics by breaking down the theme into three content blocks. The first block includes three chapters, which deal with general aspects of the Forensic Genetics and the laboratories that are dedicated to this field, making a review of the history and evolution of Forensic Genetics fundamentally through their work groups and scientific societies who row for the evolution and standardization of this discipline (chapter 1). Likewise, the need to establish a quality system in the forensic genetics laboratories that provide their services to the Justice Administration will be highlighted, and how it will be achieved through the implementation of ISO / IEC 17025: 2017 (chapter two). This block closes, analyzing one of the Achilles heels of the genetic test, the contamination, its transcendence and its repercussion in the course of the investigation. To this end, some cases of significant media coverage that try to offer a practical perspective to the reader are included (chapter 3).

A second block includes a set of 10 chapters that will deal with more technical and methodological aspects within the Forensic Genetics. This block is initiated by addressing a phase of great importance in genetic analysis, such as the collection of evidence and its referral to forensic laboratories, as well as the methodologies applied to the characterization of biological fluids and other indications (Chapter 4). The molecular bases of the DNA extraction process are reviewed in the different methods used for the recovery of DNA from the evidence (chapter 5), as well as an updated view of the methods of quantification of human DNA (chapter 6). In addition, the autosomal STRs markers of forensic interest and the possible difficulties and challenges raised by their analysis will be assessed (chapter 7). We will analyze one of the main techniques currently applied for the detection of DNA polymorphisms, capillary electrophoresis (Chapter 8), offering a practical view of its use, incidents and possible solutions to them. Chapters 9 and 10 review the socalled haplotypic markers, Y chromosome markers and mitochondrial DNA, of undoubted utility in forensic genetics. But other markers that are gradually being introduced in genetic forensic phenotypic investigations, markers (chapter 11), and that will help in complex cases will also be analyzed. Chapter 12 includes a complete review that deals with the analysis of evidences or remains of non-human origin (animals, plants. microorganisms). And to close this block, a complete revision of the new mass sequencing techniques (MPS) that are beginning to be validated and applied to the field of forensic genetics will be made, both for classical applications and in other new research channels (Chapter 13).

Finally, the third block of this book will deal with aspects related to the interpretation of the results obtained in the genetic test. The first chapter of this block will address the genetic study of kinship relations, with the analysis of both simple and complex paternity (chapter 14). The next one will focus on the topic of the socalled "critical DNA profiles", such as mixtures or those in which the DNA is in low quantity and / or degraded (chapter 15). Other specific cases will also be assessed, such as genetic identification in large catastrophes or missing persons (chapter 16). In chapter 17, the importance and transcendence of DNA databases of criminal interest and their use for the resolution of judicial cases with a worldwide vision will be reviewed. The assessment (statistics) of the genetic test and its presentation to the courts of justice will be developed in chapter 18, addressing those aspects that are most relevant and critical of this phase of the analysis. Chapter 19 develops the format of expert reports on genetic analysis, exposing characteristics the and requirements thereof, in accordance with the requirements of ISO / IEC 17025: 2017, focusing on the expression of results and conclusions, offering tools to the reader, through the presentation of examples and communication strategies (chapter 19). This last block will close with an interesting chapter on the legal and bioethical aspects of the study of DNA for forensic identification purposes (chapter 20).

Forensic genetics: From the laboratory to the Courts, is a work in which multiple authors have participated. The professionals who have contributed to the elaboration of the different chapters of this book, are experts of a wide experience. professional of proven solvency and international reputation, which guarantees the quality and rigor of this work. Forensic Genetics: From the laboratory to the Courts is designed to support and consult the different groups that, from different perspectives, may be interested in this discipline of Forensic Sciences: students, professionals in the scientific field and in the legal field, as well as members of security forces and bodies.

The editors of this book trust that the future reader of this book, both experienced and those who are new to the subject, will be grateful for the treatment of all the topics that compose it and will be very useful. To carry out the approach of all these contents, it has been tried to give a vision as practical as possible, with a didactic objective. Thus, in all those topics that lend themselves to this, concrete and real cases have been exposed. All this to find the best way to illustrate the topics discussed and make them as understandable as possible. This book attempts to respond to the need for quality reference texts in Spanish within the field of forensic genetics, as well as to serve as an extension of this area of knowledge.

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