Current challenges in statistical DNA evidence evaluation

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Alla mia famiglia

The front cover of this book was drawn by my father, Paolo Cereda.
In the back cover Lord Ganesha, the god of wisdom and learning, the patron of arts and sciences, as well as the remover of obstacles. One of his two tusks was broken to write down a very important textbook, the Mahabarata. According to some myths, he was generated by his mother, Parvathi, alone, using turmeric paste. For this reason, he represents women's independence.

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Preface

DNA profiling has become one of the most widely used techniques for human identification in forensic science since its introduction in 1984 by Alec Jeffreys. Despite the common belief that DNA evidence is a "damning evidence" which leaves no space for uncertainty, it actually needs strong statistical models in order to be used as a support for particular conjectures. The process which allows forensic experts to evaluate the statistical meaning of DNA evidence is one of the most interesting domains of forensic science of the last decades. This thesis started with the aim of building a statistical interpretative framework for a new genotyping methodology, the DIP-STR marker system, conceived to deal with the problem of extremely unbalanced mixtures.

While working on this project, we were confronted with the so-called 'rare type match problem', a very interesting open problem of forensic DNA statistics. The term refers to the situation in which there is a correspondence between the DNA profile of a suspect and that of a recovered stain, but this profile was never observed in a previously collected reference database. The evaluation of such a correspondence is very challenging. This problem is very common when using Y-STR markers or new genotyping techniques, such as DIP-STR markers, since the coverage of the available databases is limited. Therefore, we started investigating several statistical methods to deal with the rare type match problem. This led to the in-depth study of other delicate methodological issues, such as uncertainty assessment, data reduction, hybrid solutions.

As a closing loop to this Phd project, one of the discussed methods is proposed as a solution to the DIP-STR rare type match problem.

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