

1

Introduction

1.1 Weight-of-evidence theory

The introduction of DNA evidence around 1990 was a breakthrough for criminal justice, but it had something of a ‘baptism of fire’ in substantial controversy in the media and courts over the validity of the technology and the appropriate interpretation of the evidence. DNA profiling technology has advanced since then, and understanding by lawyers and forensic scientists of the appropriate methods for evaluating standard DNA profile evidence has also improved. However, the potential for crucial mistakes and misunderstandings remains. Although DNA evidence is typically very powerful, the circumstances under which it might not lead to satisfactory identification are not widely appreciated. Moreover, new problems have arisen with low-template DNA (LTDNA) profiles, which can be subject to stochastic events such as drop-in and drop-out.

The report of Caddy et al. [2008] was commissioned by the UK Government in response to the controversy over the 2007 acquittal of a defendant charged with the 1998 Omagh bombing in Northern Ireland. It found the underlying science to be ‘sound’ and LTDNA profiling to be ‘fit for purpose’, while admitting that there was lack of agreement ‘on how LTDNA profiles are to be interpreted’. We find those phrases to be mutually incompatible. Fortunately, much progress has been made since 2008, but the international controversy surrounding the legal process arising from the murder of Meredith Kercher in Perugia, Italy, in 2009, in which LTDNA evidence played a central role, highlights the challenges that can arise. We aim in this book to present the fundamental concepts required for interpretation of DNA profiles, including LTDNA. We will initially focus on the general issues concerning the measurement of evidential weight, develop the weight-of-evidence theory based on

Weight-of-Evidence for Forensic DNA Profiles, Second Edition.

David J. Balding and Christopher D. Steele.

© 2015 John Wiley & Sons, Ltd. Published 2015 by John Wiley & Sons, Ltd.

Companion Website: www.wiley.com/go/balding/weight_of_evidence

2 WEIGHT-OF-EVIDENCE FOR FORENSIC DNA PROFILES

likelihoods and discuss some alternative probability-based approaches. We will then apply the theory to forensic DNA profiling.

The primary goal of this book is to help equip a forensic scientist charged with presenting DNA evidence in court with guiding principles and technical knowledge for

- the preparation of statements that are fair, clear and helpful to courts, and
- responding to questioning by judges and lawyers.

The prototype application is identification of an unknown individual whose DNA profile was recovered from a crime scene, but we will also discuss profiles with multiple contributors, as well as paternity and other relatedness testing, and consider profiles that are subject to drop-out and other consequences of LTDNA and/or degraded DNA. We assume the setting of the United States, the United Kingdom and Commonwealth legal systems in which decisions on guilt or innocence in criminal cases are made by lay juries, but the general principles should apply to any legal system.

We will introduce and develop a weight-of-evidence theory based on two key tenets:

1. The central question in a criminal trial is whether or not the defendant is guilty.
2. Evidence is of value inasmuch as it alters the probability that the defendant is guilty.

Although these tenets may seem self-evident, it is surprising how often they are violated. Focussing on the right questions clarifies much of the confusion that has surrounded DNA evidence in the past.

It follows from our tenets that evidential weight can be measured by likelihoods and combined to assess the totality of the evidence using the appropriate version of Bayes' theorem. We will discuss how to use this theory in evaluating evidence and give principles for, and examples of, calculating likelihoods, including taking into account relevant population genetic factors.

No theory ever describes the real world perfectly, and forensic DNA profiling is a complex topic. The weight-of-evidence theory developed here cannot be applied in a naive, formulaic way to the practical situation faced by forensic scientists in court. Nevertheless, a firm grounding in the principles of the theory provides:

- the means to detect and thus avoid serious errors;
- a basis for assessing approximations and simplifications that might be used in court;
- a framework for deciding how to proceed when the case has unusual features;
- grounds for deciding what information a clear-thinking juror needs in order to understand the strength of DNA profile evidence.

Fortunately, we will see that the mathematical aspects of the theory are not too hard. Of course assessing some of the relevant probabilities – such as the probability that

a sample handling error has occurred – can be difficult in practice, reflecting the real-world complexity of the problem. Further complications can arise, for example, in the case of mixed DNA samples (Section 6.5). However, the same simple rules and principles can give useful guidance in even the most complex settings.

There exist other theories of weight of evidence based on, for example, belief functions or fuzzy sets. The theory based on probability presented here is the most widely accepted, and its philosophical underpinnings are compelling [Bernardo and Smith, 2009, Good, 1991]. So whatever is actually said in court in connection with DNA evidence, it should not conflict with this theory.

There has been debate about the appropriateness in court of using numbers to measure weight of evidence. We only touch on this argument here (Sections 6.3.4 and 11.4.5). It is currently almost universal practice to accompany DNA evidence by numbers in an attempt to measure its weight (but see Section 11.4.6), and so we focus here on issues such as which numbers are most appropriate in court, and how they should be presented.

1.2 About the book

Chapters 2, 3 and 11 are not scientifically technical and, for the most part, are not specific to DNA evidence. We therefore hope that lawyers dealing with scientific evidence, and forensic scientists not principally concerned with DNA evidence, will also find at least these chapters to be useful. Courtroom lawyers ignorant of the weight-of-evidence theory described in Chapters 2 and 3 should be as rare as theatre critics ignorant of Shakespeare, yet in reality, we suspect that few are able to command its elegance, power and practical utility.

We first set out the weight-of-evidence theory informally, via a simplified model problem (Chapter 2) and then more formally using likelihoods (Chapter 3). In Chapter 4, we briefly survey DNA-based typing technologies, starting with an introduction to autosomal¹ short tandem repeat (STR) typing, emphasizing possibilities for typing error, then moving on to other DNA typing systems, digressing briefly to discuss fingerprint evidence and finishing with some newer evidence types: methylation, RNA and phenotype and ancestry prediction from DNA. Next, we survey some population genetics theory relevant to DNA profile evidence (Chapter 5). These two chapters prepare us for calculating likelihoods for DNA evidence, which is covered in Chapters 6 (identification) and 7 (relatedness). In Chapters 8 and 9, we extend identification inferences to LTDNA profiles and give a brief introduction to our freely available software for LTDNA profile evaluation, `likeLTD`. Chapter 10 discusses some alternative probability-based approaches to assessing evidential strength: none of these methods is recommended but each has its merits. In Chapter 11, we discuss some basic fallacies in the evaluation of DNA profile evidence and briefly review the opinions of some UK and US legal and scientific authorities.

¹ The nuclear chromosomes excluding X and Y.

1.3 DNA profiling technology

For the most part, we will assume that the DNA evidence is summarised for reporting purposes as the lengths of STR alleles at multiple autosomal loci (typically 10–25), reported as the number of tandem repeats of a DNA motif, usually 4 base pairs (bp) in length. The final result at four of the loci might be reported as

STR locus:	D18	D21	THO1	D8
Genotype:	14,16	28,31	9.3,9.3	10,13

in which each pair of numbers at a locus indicates the numbers of repeats on the individual's two alleles. Although whole repeats are the norm, partial repeats sometimes occur (Section 4.1.1); the profile represented here is homozygous for a THO1 allele that includes a partial repeat: the 9.3 indicates 9 full copies of the 4 bp DNA motif plus a 3 bp partial repeat.

Autosomal STRs now form the standard DNA typing technology in many countries. Both the 13-locus Combined DNA Index System (CODIS) and 15-locus European standard set (ESS) were derived by expanding earlier, smaller sets of STRs, and both have been superseded by larger sets. Most of Europe currently uses a 16-STR system developed from the ESS, which includes the amelogenin sex-identifying locus. The GlobalFiler[®] set from Life Technologies-Applied Biosystems offers 22 STRs, amelogenin and a Y-indel marker [Life Technologies, 2014], while the PowerPlex[®] Fusion System from Promega offers 23 STRs and amelogenin [Promega, 2014]. The different systems have many loci in common, but alleles at a locus can have different flanking regions (Section 4.1) in different systems. For more details of current STRs in use and developments that have allowed the expansion of STR sets, see Phillips et al. [2014].

The process of typing STR profiles is introduced in Section 4.1 but is not covered in great depth in this book. For further details, see Butler [2010]. Rudin and Inman [2001] give a general introduction to both technical and interpretation issues. Although we emphasise STR profiles, the principles emphasised in the following apply equally to any DNA profiling system. Interpreting profiles from the haploid parts of the human genome (the Y and mitochondrial chromosomes (mitochondrial DNA, mtDNA)) raises special difficulties. These systems are introduced in Sections 4.2 and 4.3, and interpretation issues specific to them are discussed briefly in Section 6.4. In Section 4.5, we briefly discuss profiles based on single-nucleotide polymorphism (SNP) markers.

1.4 What you need to know already

Chapters 2, 3, and 11 have essentially no technical prerequisites. To follow Chapter 5, you should know already what an STR profile is, have a rudimentary genetics vocabulary (locus, allele, etc.) and know the basic ideas of Mendelian

inheritance. In statistics, you should be familiar at least with the theory of the error in a sample estimate of a population proportion (binomial distribution). The reader with experience of calculating with probabilities will be at an advantage in Chapters 6 and 7, but few technical tools are required from probability theory. In Sections 5.4.1 and 10.3, familiarity with statistical hypothesis testing are assumed, but these sections are labelled with a †, which means that they can be skipped without adverse impact on your understanding of the remainder of the book. The most important tool for computing probabilities is the sampling formula (5.6), which is expressed in a remarkably simple recursive form that can be used repeatedly to build up complex formulas. We give examples of its use, which requires only an ability to add and multiply, and with practice, anyone should be able to use it without difficulty.

We do not provide a general introduction to statistics (for an introduction in forensic settings, see Aitken and Taroni [2004]) and give only a brief introduction to population genetics (Section 5.1). We believe that many complications and much confusion have arisen unnecessarily in connection with DNA evidence because of a failure to grasp the basic principles of assessing evidential weight. If one focusses on the questions directly relevant to the forensic use of DNA profiles, the number of ideas and techniques needed from statistics and population genetics is small.

While the central ideas are not very difficult, inevitably, there are special cases with their unique complexities. In addition, new ideas always take some time to absorb. Given some effort, this book should equip you with the basic principles for tackling any problem of interpreting forensic DNA evidence. The details of complex scenarios will never be straightforward, and no book can replace the need for thought, care and judgement on the part of the forensic scientist. The goal of this book is to complement these with some technical information and bring them to bear on the appropriate questions with guiding principles for assessing weight of evidence.

1.5 Other resources

Part of the reason for writing the book is to synthesise and extend our previous contributions to the forensic science and related literature in a coherent manner. In particular, Chapter 3 is a development of Balding [2000], Section 7.1 extends the paternity section of Balding and Nichols [1995] and Section 10.1 is based on Balding [1999]. Perhaps the most important feature of the book is the introduction of the population genetics sampling formula (Section 5.3), and its systematic application to various identification and relatedness problems. This draws in part on Balding and Donnelly [1995a], Balding and Nichols [1995] and Balding [2003], but some of the development is new here. Chapter 8 draws on the development in Steele and Balding [2014a].

There are several other books that deal with the statistical interpretation of DNA and other evidence. Aitken and Taroni [2004] gave a thorough introduction to the statistical interpretation of scientific evidence in general, including DNA evidence among other evidence types. Robertson and Vignaux [1995] also dealt with a range of

6 WEIGHT-OF-EVIDENCE FOR FORENSIC DNA PROFILES

evidence types and emphasised interpretation issues from a lawyer's perspective, giving less attention to technical scientific aspects; for example, they did not discuss population genetics. Evett and Weir [1998] is perhaps closest to this book, but the treatment of population genetics issues by these authors is different from ours, as is their approach to introducing the relevant statistical issues. Butler [2014] gave an introduction to the interpretation issues raised by STR profile evidence, while Buckleton et al. [2005] offered a more extensive treatment (second edition expected in 2015).

As far as we can see, there is no major philosophical difference between us and these authors: we all embrace the use of likelihoods and Bayes' theorem to evaluate evidence. We emphasise different aspects according to our individual perspectives, experience and target audiences. Our book develops the weight-of-evidence theory in general and from an introductory level, and its approach to population genetics issues is unique, while remaining concisely focussed on DNA profile evidence, without extensive related material.

Charles Brenner's 'Forensic Mathematics' website dna-view.com provides much information and discussion; see also the encyclopaedia article [Brenner, 2006]. Weir [2007] offered a more extensive, one-chapter summary of many issues. The International Society for Forensic Genetics <http://www.isfg.org/> is the principal professional organisation for forensic genetics, and it sponsors the leading journal *Forensic Science International: Genetics*. Biedermann et al. [2014] presented a recent collection of articles on issues related to the interpretation of DNA profile evidence. The International Conference on Forensic Inference and Statistics is held every 3 years and in 2014 was in Leiden, Netherlands; the next meeting is expected to be in 2017 in North America.