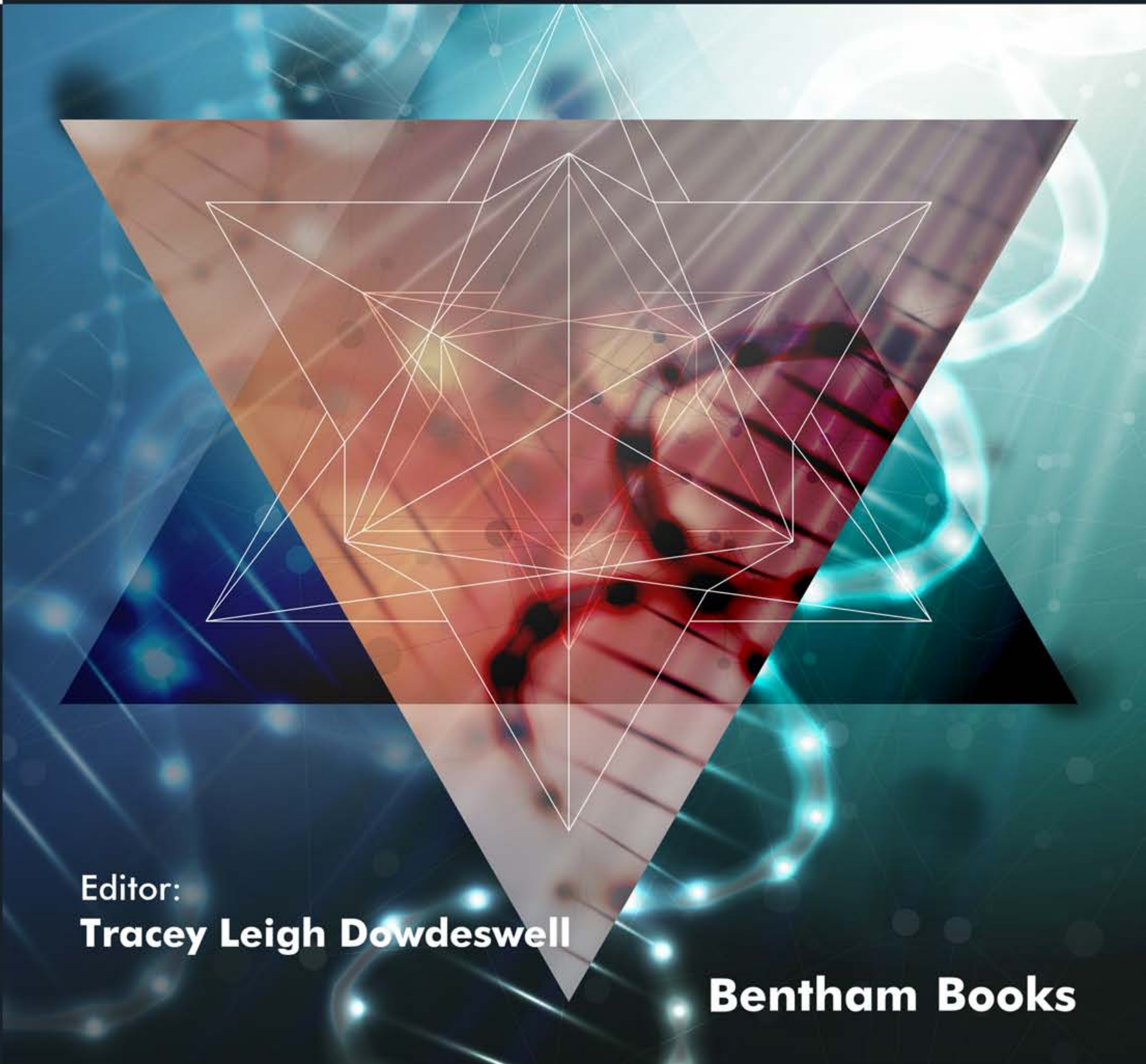
DATA GOVERNANCE FOR JUSTICE AND HUMAN RIGHTS

FORENSICS, FLOW AND FRONTIERS



Editor:

Tracey Leigh Dowdeswell

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Current and Future Developments in Law

(Volume 4)

Data Governance for Justice and Human Rights: Forensics, Flow, and Frontiers

Edited by

Tracey Leigh Dowdeswell
Department of Humanities & Social Sciences
Douglas College, 700 Royal Avenue
New Westminster, BC V3M 5Z5, Canada

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Data Governance for Justice and Human Rights: Forensics, Flow, and Frontiers

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FOREWORD

The use of data science in law has gone a long way with the recent industrial revolutions of digital technology and Artificial Intelligence. This book discusses these developments and sheds light on their positive and negative implications and potential. The book discusses these future directions in three parts: (1) using data to improve legal decision-making, (2) governing the legal use of data, and (3) the forensic use of genetic data.

The book begins with novel methods whereby data is being used in legal decision-making, examining some of the ways in which artificial intelligence (AI) and machine learning (ML) technologies are handling data to improve the legal system itself. The chapters in this section focus on the ways in which data can be used to improve legal decision-making – and thus to improve access to justice, to better justify decisions, to reduce biases in decision-making, and to evaluate evidence to reduce miscarriages of justice.

The second part of the book, governing the legal use of data, discusses how new strategies are required to deal with the technological advances in this field. The chapters in this section focus on eliminating the use of false legal information hallucinated by AI systems, identifying and addressing cognitive biases among forensic scientists, and regulating the forensic use of bioinformatics in the criminal justice system.

The third and last part of the book, the exploration of the forensic use of genetic data, explores some of the most significant advances that have been made in the field of bioinformatics and its application to human identification. The chapters in this section focus on these developments, examining how advances in SNP sequencing, combined with computational methods for kinship identification, are leading to the clearance of previously unsolvable cases.

Data science and law have always been intertwined. Nonetheless, outstanding recent advances in data science, and especially the technologies that underpin it, have made this interconnection a powerful and often contentious paradigm. This book is an innovative, comprehensive, and future-facing scholarly work that aims to explore the benefits, as well as the hazards, coupled within this novel nexus.

Nachshon Goltz
Edith Cowan University
270 Joondalup Dr Joondalup
WA 6027
Australia

PREFACE

I am very pleased to have been involved in this project and to help bring about this volume at a very exciting time in which data science, bioinformatics, and artificial intelligence are transforming our legal and criminal justice systems.

I would like to thank our editors at Bentham Science Books, including Humaira Hashmi and Areeba Tanveer, for all their efforts in bringing this volume to completion.

I would also like to thank the following contributors for their excellent and diverse contributions to this book: Bruce Budowle, James French, Jianye Ge, Sean Goltz, Ellen Greytak, Swathi Ashok Kumar, Ruth Morgan, Taryn Mulvihill, Sherry Nakhaeizadeh, Karen Richmond McGregor, Antti Sajantila, Zhao Yuxin, and John Zeleznikow.

I would also like to thank each of our outstanding contributors, as well as our peer reviewers, including CeCe Moore, for taking the time to review this work and for their helpful suggestions.

I am deeply grateful to all of you for the role you have played, not only in this work, but also for your contributions to the field as a whole. It is only with the contribution of such esteemed scholars that we were able to create this book.

Tracey Leigh Dowdeswell
Department of Humanities & Social Sciences
Douglas College, 700 Royal Avenue
New Westminster, BC V3M 5Z5, Canada

List of Contributors

Antti Sajantila	Department of Forensic Medicine, University of Helsinki, Helsinki, Finland Forensic Medicine Unit, Finnish Institute for Health and Welfare, Helsinki, Finland
Bruce Budowle	Department of Forensic Medicine, University of Helsinki, Helsinki, Finland
David Gurney	Investigative Genetic Genealogy Center, Ramapo College of New Jersey, 505 Ramapo Valley Road, Mahwah, New Jersey 07430, USA
Ellen Greytak	Parabon NanoLabs, Inc. Suite 406, Reston, 11260 Roger Bacon Dr., Virginia 20190, USA
James French	Department of Security and Crime Science, Centre for the Forensic Sciences, University College London, Gower Street, London WC1E 6BT, United Kingdom
Jianye Ge	Department of Microbiology, Immunology & Genetics and Center for Human Identification, University of North Texas Health Science Center (UNNTHSC), 3500 Camp Bowie Blvd., Fort Worth, Texas 76107, USA
Karen McGregor Richmond	Kellogg College, University of Oxford, Oxford OX2 6PN, United Kingdom
Ruth M. Morgan	Department of Security and Crime Science, Centre for the Forensic Sciences, University College London, Gower Street, London WC1E 6BT, United Kingdom
Sean Goltz	Edith Cowan University, 270 Joondalup Drive, Joondalup, Perth WA 6027, Australia
Sherry Nakhaeizadeh	Department of Security and Crime Science, Centre for the Forensic Sciences, University College London, Gower Street, London WC1E 6BT, United Kingdom
Swathi Kumar	QIAGEN Sciences, 19300 Germantown Rd, Germantown, Maryland 20874, USA
Taryn Mulvihill	Cognitive Systems, University of British Columbia, BC, Canada
Tracey Leigh Dowdeswell	Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada
Zhao Yuxin	National University of Singapore, University Town, 2 College Avenue West, #01-03, (Stephen Riady Centre), Singapore 138607

CHAPTER 1

Introduction: Current and Future Developments in the use of Data Science in Law

Tracey Leigh Dowdeswell[1,*]

[1] *Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada*

Abstract: This chapter introduces the volume as a whole, outlining the current and future directions in the legal use of data, and exploring how data can be utilized in the legal system to promote justice and human rights, as well as enhance the legal decision-making process itself.

Part I examines some of the ways in which Artificial Intelligence (AI) and Machine Learning (ML) technologies are marshalling data to improve legal decision-making. The included chapters examine the ways in which data science has been used to improve legal decision-making, from its inception in the mid-twentieth century to Groningen's school's more recent work on using AI to model the fact-finding process itself. Data, when governed judiciously, can be used to improve legal decision-making and thus improve access to justice, better justify decisions, reduce biases in decision-making, and evaluate evidence to improve accuracy and reduce miscarriages of justice.

Part II examines new strategies to govern the use of data in the legal system: identifying and addressing cognitive biases among forensic scientists, regulating the forensic use of bioinformatics in the criminal justice system, and eliminating the use of false legal information hallucinated by AI systems.

Part III of this book describes some novel forensic applications of genetic data, particularly in the field of bioinformatics and its advances in human identification. Some of the most significant advances have been made in the field of bioinformatics and its application to human identification. The chapters in this section focus on these developments, examining how advances in SNP sequencing, combined with computational methods for kinship identification, are leading to the clearance of previously unsolvable cases.

Keywords: Artificial Intelligence, AI and law, Argumentation theory, Bioinformatics, Data science, Ethics of AI, Forensic genetics, Forensic genomics, Forensic science, Genetic genealogy, Human identification, Kinship, Legal ethics, Machine learning, Philosophy of evidence.

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[*] **Corresponding author Tracey Leigh Dowdeswell:** Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada; E-mail: dowdeswellt@douglascollege.ca

Tracey Leigh Dowdeswell (Ed.)

INTRODUCTION

This book examines current and future directions in the application of data in the legal system. The authors discuss various ways in which data can be utilized to promote justice and human rights, as well as to enhance the legal decision-making process itself.

Part I examines the ways in which data science has been utilized to enhance legal decision-making, from its inception in the mid-twentieth century to Groningen's more recent work on using AI to model the fact-finding process itself, as described by McGregor Richmond. Dowdeswell provided one such case study, applying argumentation theory to the evaluation of evidence in a notorious miscarriage of justice. Part II examines new strategies to govern the legal use of data: eliminating the use of false legal information hallucinated by AI systems, regulating the forensic use of bioinformatics in the criminal justice system, and identifying and addressing cognitive biases among forensic scientists. Part III describes several novel applications of the forensic use of data, particularly in the field of bioinformatics and its advances in human identification.

PART I: CURRENT AND FUTURE DIRECTIONS IN USING DATA IN LEGAL DECISION-MAKING

As discussed above, part I of this book examines some of the ways in which Artificial Intelligence (AI) and Machine Learning (ML) technologies are marshalling data to improve the legal system itself. Each of the chapters in this part examines the ways in which data can be used to improve legal decision-making, thereby enhancing access to justice, justifying decisions more effectively, reducing biases in decision-making, and evaluating evidence to enhance accuracy and prevent miscarriages of justice.

Karen McGregor Richmond, in her chapter, "*The Integration of Data Science and Evidential Analysis*," reviews the use of data science to understand the analysis of evidence and the processes of juridical proof [1]. Innovations in data science and ML have led to advancements in the modelling of legal reasoning and legal prediction science, where argumentation modelling has become a major research focus [1]. This, in turn, has enabled data science to play a leading role in understanding and modelling the fact-finding process itself [1].

This chapter reviews the work of the Groningen School, which has played an important role in shaping this field. A significant contribution of the Groningen School has been to adopt an integrated, hybrid perspective of the fact-finding process [1]. This has led to the proliferation of research that integrates various strengths of the Wigmorean, Narrative, and Bayesian accounts of fact-finding – the three main normative approaches used in the field [1]. Hybrid approaches provide a better account of the adversarial setting of arguments, both pro and con, the globally coherent perspective offered by scenarios, and the dynamic uncertainty of probabilities than do previous approaches, such as those relying on legal positivism [1]. This also enables explainability in ML systems that model the fact-finding process [1]. Explainability, in turn, is crucial for these systems to be accepted by courts and legal professionals [1].

Tracey Dowdeswell, in her chapter "*Argumentation Schemes*, *AI, and Criminal Law: Evaluating Evidence in a Miscarriage of Justice*," uses argumentation schemes to analyze evidence in a criminal case [2]. She reviews the application of artificially intelligent systems to the assessment of evidence in criminal cases and proposes a system for formalizing argumentation schemes for future computational applications [2]. She uses argumentation schemes to examine the evidence in a notorious miscarriage of justice – that of Robert Earl Hayes [2].

She draws upon the themes raised in McGregor Richmond's chapter. Her work combines abductive reasoning with narrative or scenario-based reasoning and integrates them within argumentation schemes [2]. She identifies six different scenarios, or narrative explanations, as having been put forward by the parties over the years to explain the evidence in Hayes' criminal prosecutions [2]. Each scenario is evaluated separately; in the final step, their relative plausibilities are compared using an abductive argumentation scheme [2].

The final result is a plausibility value that indicates how justified we are in believing that a given scenario is the most plausible explanation for the available evidence at the time of evaluation [2]. The plausibilities proposed here are qualitative, relative and are correlated with legal standards of proof [2]. This system assists in identifying and removing prejudices and cognitive biases that impede reliable evaluations of evidence [2]. It also assists in better formalizing and defining argumentation schemes to evaluate evidence in criminal cases, and to prepare the way for future computational applications [2].

Dowdeswell argues that miscarriages of justice are best prevented by using methods that are objective, accurate, reproducible, and sufficiently formalized that any agent, including a computational decision support system, would obtain a similar result [2]. Systematization and formalization are key to accurately evaluating the plausibility of evidence, and they form the basis for further application of ML technologies in this domain of legal decision-making [2].

CHAPTER 2

The Integration of Data Science and Evidential Analysis

Karen McGregor Richmond[1,*]

[1] *Kellogg College, University of Oxford, Oxford OX2 6PN, United Kingdom*

Abstract: This chapter reviews the use of data science and machine learning applications to facilitate the analysis of evidence and the processes of juridical proof. Innovations in data science and machine learning have led to advancements in forensic interpretation, risk prediction, and the efficient processing of large datasets of 'open source' material. However, the '*holy grail*' of data science development in the evidence domain is the modelling of legal arguments themselves. Data science holds the potential to play a leading role in understanding and modelling the fact-finding process. Argumentation schemes have thus become a major research focus and hold the potential - when aligned with parallel developments in legal reasoning – to fully automate the trial process. This chapter considers the standard applications of data science to legal evidence – forensics, prediction, and data processing - before reviewing the work of the Groningen School, which has played an important role in shaping this field. Crucially, it is intended that this introductory chapter offer an integrated perspective on evidential reasoning, demonstrating not only the unique affordances of particular inferential modes encountered in the surveyed literature but also highlighting the compound and contextually dependent nature of these features. This chapter, therefore, highlights the importance of attention to conditioning factors based upon the structure of, and inter-relations between, the overall form of the legal system, legal sub-field, mode of argumentation, and form of adjudication.

Keywords: Artificial intelligence, Argumentation theory, Law, Machine learning, Philosophy of evidence.

INTRODUCTION

The rapid development of algorithmic, machine learning, and AI-driven processes and systems has led to the simultaneous proliferation of data science innovations across multiple system boundaries, and their subsequent encroachment into hitherto sacrosanct domains. Indeed, the deployment of data-science products

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[*] **Corresponding author Karen McGregor Richmond:** Kellogg College, University of Oxford, Oxford OX2 6PN, United Kingdom; E-mail: karen.richmond@kellogg.ox.ac.uk

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within sensitive disciplines, such as medicine, defense, and law, is generating significant practical efficiencies [1], systematic distortions [2], and social concerns [3].

Within the legal field alone, proprietary algorithms have been developed to analyze complex DNA mixtures [4], judges have utilized data science models to predict recidivism during sentencing [5], human rights NGOs have developed AI models to process vast 'open source' datasets [6] whilst lawyers have mobilized the potentials of, and discovered the limitations of, generative AI in the courtroom [7].

Thus, whilst the past three decades have witnessed the steady incorporation of data science products into fields, such as criminal investigation, legal reasoning, and adjudication, contrapuntal tensions have also necessitated a deeper inquiry into the legal and regulatory interventions necessary to govern the scientific production and utilization of data science technologies [8].

Such are the concerns and controversies surrounding the potential for data-science innovations in high-stakes decision-making (tempered by a healthy skepticism regarding the most fervid claims and ebullient predictions) that proactive steps have been introduced to moderate data science innovation at the global, regional, and domestic levels [9, 10].

Nonetheless, within the legal field, innovations continue apace, and novel techno-legal products continue to be developed, the majority of which are borne on a current of innovative research whose genesis and antecedents crystallize around a trifecta of topics, including reasoning with legal precedents [11], the representation of legal knowledge [12], and the modelling of normative concepts [13]. Before focusing on the sub-field of legal evidence and proof, it is first necessary to trace the origins of automated argument modelling in efforts to model legal reasoning more generally.

ARTIFICIAL NEURAL NETWORKS AND THE SIMULATION OF LEGAL KNOWLEDGE

As the potentials of digital automation became apparent, attention within the legal field turned to the systematic simulation of legal knowledge. Studies concerned with the representation of legal knowledge tended to focus on the modelling of specific legal sub-fields, most notably civil disputes involving property issues, trade secrets, and taxation [14]. These endeavors stimulated a greater awareness of the heterogeneous nature of legal disputes and the particularities that characterize each legal sub-domain and were facilitated in large part by the consolidation of knowledge management and data sharing potentialities generated by the development of the Semantic Web [15].

This in turn led to innovations, such as "linked data" principles and the adoption of standardized knowledge representation formalisms, such as RDF and OWL [16]. Systematization allowed for a dramatic increase in the quantity of information available for processing and led to further efficiencies in the organization of data, including document corpora and citation analysis, as well as a broad-based development in sub-symbolic methods and their subsequent utilization in Artificial Neural Nets (ANNs) [16].

However, the principal development in the field was the rapid evolution in the modelling of reasoning using legal cases and precedents. The development of this research stream was lent added vigor by another adventitious technological development: the advancement of Machine Learning (ML) technologies, which allowed for increasingly efficient and accurate prediction of relevant legal cases [17, 18]. In addition, this engendered the automation of further legal tasks, rendered tractable by the ability of ML systems, which could directly process natural language as opposed to abstracted feature vectors.

However, for the purposes of the instant survey, it is worth noting that these early stages also saw the steady, if less dramatic, development of research into the modelling of legal arguments. Beginning in the final decade of the last century, researchers began to utilize text dialogues in an attempt to encapsulate legal procedures, principally through the depiction of legal arguments. Moreover, parallel to the development of legal ontologies engendered by the consolidation of the semantic web, data-linking also allowed for argument mining and the transition from dialogues to argument schemes, thus establishing argument-modelling – and questions of legal evidence – as a central focus of law and data science research [19].

However, it should be noted that the purpose of this prolegomena is not solely to present data science innovations for a primarily legal audience, but to raise awareness amongst non-legal professionals of the systemic and procedural factors which necessarily condition the fact-finding process. Thus, before turning to matters of evidence analysis in the following section, it is worth highlighting that the categorizations and ramifications elaborated below are founded not only on the reviewed literature but on an extensive published survey of explainable data science and legal evidence research, as well as practical interdisciplinary experience in the development of an automated case-based reasoning system for automated asylum adjudication [19].

CHAPTER 3

Argumentation Schemes, AI, and Criminal Law: Evaluating Evidence in a Miscarriage of Justice

Tracey Leigh Dowdeswell[1,*]

[1] *Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada*

Abstract: This chapter discusses the application of artificially intelligent systems to the assessment of evidence in criminal cases and proposes a system for formalizing argumentation schemes for machine learning. It draws on recent work on argumentation schemes and adapts them to address common issues encountered in criminal cases, such as the credibility of witness testimony, the probative value of similar facts, and the evaluation of expert evidence. This is illustrated by systematically setting out the arguments presented over the course of several decades in the trials of Robert Earl Hayes and identifying critical questions to evaluate the evidence. Hayes' case is a well-known miscarriage of justice that has been extensively investigated and litigated. It therefore serves as a good model for evaluating the arguments put forward in the case over the course of several decades, as the facts of the case are well-known and reliable. The proposed system combines argumentation schemes with scenario-based reasoning. The arguments are separated into different scenarios put forward by the parties. Each scenario is evaluated separately; in the final step, their relative plausibilities are compared using an abductive argumentation scheme to evaluate the most plausible scenario. The final result is a qualitative plausibility value that signifies how justified we are in believing that a given scenario is the best explanation for the available evidence, at the time of evaluation. The plausibilities proposed here are qualitative, relative and are correlated with legal standards of proof. This system assists in identifying and removing prejudices and cognitive biases that impede reliable evaluations of evidence. It also assists in better formalizing and defining argumentation schemes to evaluate evidence in criminal cases, so as to prepare the way for future computational applications.

Keywords: Argumentation theory, Argumentation schemes, Artificial Intelligence, Forensic DNA, Law, Machine learning, Philosophy of evidence, Trial advocacy.

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[*] **Corresponding author Tracey Leigh Dowdeswell:** Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada; E-mail: dowdeswellt@douglascollege.ca

INTRODUCTION: THE TRIALS OF ROBERT EARL HAYES

The exoneration of Robert Earl Hayes for the 1990 murder of Pamela Albertson is well-known. Like many exonerations, it was told as a story about a man who was wrongly accused of a rape and murder he did not commit, but who eventually prevailed over a biased and unjust criminal justice system. However, stories can be deceiving, and the guilt and innocence of Hayes has been disputed for the better part of four decades – through a sprawling series of investigations and re-investigations across several jurisdictions. Given all the evidence put forward and examined over the years, it should be possible to determine whether or not Hayes' conviction was justified.

At the time of the murder, Hayes and Albertson worked together at a racetrack in Pompano Beach, Florida. Albertson's body was found on the morning of February 20th, 1990, in her room in the women's dormitory [1]. She was in a state of partial undress, and she had been sexually assaulted [1]. She died by manual strangulation; she had numerous bruises on her head and face; tissue was found under her fingernails, probably from scratching her attacker as he was choking her [1]. At his first trial, the jury convicted Hayes of capital murder, and he was sentenced to death [2].

The key pieces of evidence at Hayes's first trial were hearsay from a co-worker, testimony that he had attacked another female employee, and forensic DNA – still a relatively new science at the time. One of Albertson's co-workers testified that she had seen Hayes speaking to Albertson on the evening of her murder, and Albertson was telling Hayes that he could not come inside her room [1]. Albertson had previously told a co-worker she was afraid of Hayes, and he kept pressuring her for sex [1]. Neither of these witnesses had actual knowledge of Hayes' involvement in the murder itself [1].

Another female groom testified that Hayes had strangled her and tried to sexually assault her as well [1]. This happened after they had gone out together for an evening of drinks; she reported it to the police right away, but the charges were eventually dropped [1]. The defence argued at trial that this type of similar-fact evidence was too prejudicial to be admitted [1]. There is a danger that jurors might convict not because of the evidence, but because they think the accused has a bad character [1].

At trial, the most probative evidence was a DNA profile from semen recovered from Albertson's vagina and anus at her autopsy, and semen recovered from a tank top found beside her when she died [1]. The defence argued that the analyst's conclusions were unreliable because she had used an RFLP method called "band-shifting" which could have produced a false positive result [1]. RFLP typing for forensic analysis – the original method of "DNA fingerprinting" pioneered by Alec Jeffreys – was largely replaced with PCR-based fluorescent typing of Single Tandem Repeats (STRs) by the end of the 1990s [3]. By that time, the relatively novel laboratory techniques used in Hayes's case were already obsolete.

The Supreme Court of Florida heard Hayes's appeal in 1995 – the first appellate case in Florida to examine the reliability of DNA evidence [1]. The court looked at the band-shifting method used by the lab analyst in this case and determined that key experts, including those at the National Academy of Sciences, found it to be unreliable. The DNA evidence was inadmissible because it may have incorrectly identified the accused as being the source of the semen, *i.e.*, the band-shifting may have led to a false positive result [1]. The court ordered a new trial – but without the DNA, hearsay, or similar fact evidence that had led to his original conviction [1].

Without this evidence, the jury at Hayes's second trial acquitted him. Hayes's defence counsel, the Florida Supreme Court, and the jury all accepted the argument that Hayes was factually innocent, and that another worker at the racetrack – a Caucasian man with a history of sex offences – was the true culprit [4]. In 2002, Hayes's wrongful conviction was featured in an award-winning play, *The Exonerated*, by Jessica Blank and Eric Jensen, which tells the story of Hayes and five other Americans who were exonerated after being on death row [5].

Hayes's freedom was short-lived. In 2004, he was convicted and sentenced in upstate New York for the very similar murder of Leslie Dickenson at the Vernon Downs Raceway in 1987 [2]. Her body was found inside her apartment after reports of a fire inside her room [6]. She had been hung from her shelf, her throat was cut, and there was blood on her wrists [6]. Reports surfaced that she suffered from depression as a teenager, and police initially ruled her death a suicide [6]. It was only after they were contacted by police in Pompano Beach, who were investigating Albertson's murder, that they reopened the case: they exhumed Dickenson's remains, and a new autopsy determined that she had also died by manual strangulation [6]. In 2004, Hayes pled guilty to manslaughter, arson, and burglary [2]. He will be eligible for parole in 2025 [2].

The Innocence Project in New York sought to exonerate Hayes for Dickenson's murder as well. Lawyers from the project contacted the Broward County Conviction Review Unit in late 2020 to seek further DNA testing of evidence collected at Albertson's murder [2]. They were seeking evidence on the alternative suspect who was thought to be the source of the hairs found clutched in Albertson's hand. This was the defence theory that had won Hayes his acquittal in 1997 [2].

CHAPTER 4

AI Hallucinations as an Existential Risk to Human Society: Mitigating the Risks

Sean Goltz[1,*] and **Zhao Yuxin**[2]

[1] *Edith Cowan University, 270 Joondalup Drive, Joondalup, Perth WA 6027, Australia*

[2] *National University of Singapore, University Town, 2 College Avenue West, #01-03, (Stephen Riady Centre), Singapore 138607*

Abstract: AI hallucinations pose an existential risk to existing laws and social order. The authenticity of documents in general and legal documents (*e.g.*, statutes, regulations, bills, caselaw) in particular, is seriously jeopardized if Large Language Models (LLMs) produce – and we accept – false legal precedents. With the rise of the "AI empire", humans must secure our societies' legal foundations. This chapter will explore the risks of AI hallucinations and will provide a solution based on a document authenticity verification system. This is crucial to our survival as a society and should be built into all AI processors that deal with legal documents. Governments should enact legislation that requires AI systems to include methods of verification. This is fundamental to the rule of law in the age of AI.

Keywords: Artificial Intelligence, AI, AI hallucinations, Large language models, Law, Regulation of technology.

INTRODUCTION

AI hallucinations pose an existential risk to existing laws and social order. The authenticity of documents in general and legal documents (*e.g.*, statutes, regulations, bills, case law) in particular, is seriously jeopardized if Large Language Models (LLMs) produce – and we accept – false legal precedents. With the rise of the "AI empire," humans must secure the legal foundations of societies [1].

This chapter will explore the risks of AI hallucinations, taking the reader beyond our current understanding, and will provide an innovative solution based on a document authenticity verification system, drawing upon existing academic

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[*] **Corresponding author Sean Goltz:** Edith Cowan University, 270 Joondalup Drive, Joondalup, Perth WA 6027, Australia; E-mail: n.goltz@ecu.edu.au

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research. This will greatly minimize the risks associated with the use of AI in legal contexts. This is crucial to our survival as a society and should be built into all AI processors that deal with legal documents. The goal is to offer an approach that can be used to balance the potential of AI with the ethical and professional implications of AI hallucinations.

A HISTORY OF AI HALLUCINATIONS

The most famous case so far regarding AI hallucinations in the legal sphere is a brief submitted by the lawyer for the plaintiff in the case of Avianca Airlines [2]. This brief cited fabricated cases that were generated by the LLM ChatGPT. Among others, the chatbot provided substantial information about a fake case, *Varghese v. China Southern Airlines Co. Ltd.,* 925 F.3d 1339 (11th Cir. 2019) (*Varghese*) [3], including a detailed factual background, legal reasoning, and the courts' opinions [2].

Such exhaustive content camouflages this case as if it were true. Even when reconfirming the veracity of this case to ChatGPT, the chatbot gave a very definite answer that *Varghese* is a real case [2]. Further, ChatGPT provided some authoritative legal resources for this case [2].

According to the statement of the lawyer who wrote the brief, he was not aware of any possibility that ChatGPT can or will fabricate case law. He thought that these cases, which could not be verified by other websites, were unpublished, appealed, or just difficult to access, but ChatGPT could find them somewhere [2]. The court held that it was unreasonable for the lawyer to fail to take any other steps to verify the case law [2]. Under these circumstances, signing and filing an affirmation constitutes bad faith [2]. Accordingly, the lawyer was sanctioned to send letters to people who could be negatively impacted and to pay a fine of $5,000 [2].

Regardless of the due diligence obligations of lawyers, it is evident that AI chatbots generate fictitious judicial cases, thus misleading legal professionals who rely on them [4]. This case serves as a caution that AI-generated content can be fictional and, thus, especially in the legal domain, may threaten future court decisions. This could have a devastating effect on human society. One judge of the US District Court for the Northern District of Texas warned that AI is prone to "hallucinations and bias" and generates bogus quotes [5]. He required attorneys to certify that their documents were not drafted by generative AI, not even partially [5].

However, AI hallucinations continue to infringe on the legal profession despite these warnings and precedents. There are still instances of lawyers using AI in their legal research and encountering AI hallucinations. The U.S. District Court for the Middle District of Florida suspended a lawyer from practicing in that court for a year after he filed pleadings based on fake cases [6]. The lawyer used AI without checking the authenticity of the cases that have been generated [6]. The Court's grievance committee noted that AI cannot replace lawyers' responsibilities to conduct reasonable diligence and provide reliable legal authority [7].

In another case out of Massachusetts, a lawyer submitted a legal brief with three previous rulings that appear to be authentic. Even though these three cases included authentic elements, including case numbers, letters, and punctuation, they were detected as fake. The lawyer was sanctioned to pay $2,000 for this misbehavior [8].

Another case in Vancouver resulted in lawyer Chong Ke being reprimanded by the BC Supreme Court for submitting two fake cases on behalf of her client for a hearing in family court [9]. She also claimed that she did not know the risks of using LLMs or that they could hallucinate cases. Ke stated that she used AI to help prepare her case because she was away on a business trip at the time she was preparing the case and was busy [9]. This case is notable because the hallucinated cases were submitted as part of a child custody hearing in which Ke's client was applying to take his two children out of Canada to China for a holiday – a request that is rarely granted by the courts due to the risk of child abduction [9]. The submission of false cases, therefore, had the potential to place these children at risk. The Court found that Ke's actions were not deliberate, but they fell below the standard required of a competent lawyer, and she was ordered to pay the costs of the opposing party [9].

AI providers are also paying the price of AI hallucinations. In a recent scandal, Google's Gemini AI image generation service produced inaccurate, racist, and biased depictions of some historical figures [10]. A user had asked Gemini to generate images of German Nazi soldiers in 1943 [10]. However, Gemini returned several images of people of color in German uniforms [10]. Meanwhile, this service has refused to depict images of certain races [10]. Besides being criticized by the public, the negative effects of AI hallucinations reached Wall Street [11]. Google's parent company, Alphabet, lost about $90 billion in market value due to the controversy over Gemini's hallucinations [11]. Analysts pointed out that if Google is viewed as an unreliable source of AI, this could be highly detrimental to its business [11].

AI has made great advances in recent years, and this has dramatically impacted various fields. AI constitutes a comprehensive instrument, facilitating a paradigm

CHAPTER 5

Recognizing and Mitigating Cognitive Bias in Forensic Science and Human Identification

Sherry Nakhaeizadeh[1,*], **Ruth M. Morgan**[1] and **James French**[1]

[1] *Department of Security and Crime Science, Centre for the Forensic Sciences, University College London, Gower Street, London WC1E 6BT, United Kingdom*

Abstract: The research concerning the effect of cognitive bias in forensic science decision-making has been well documented. Numerous studies across legal and forensic investigations have shown a number of distinct ways that expectations, motivations, and beliefs can influence memory, as well as the selection and evaluation of evidence. In human identifications, studies have shown how contextual information can affect the interpretation of skeletal remains when establishing a biological profile. Although the concept of cognitive bias is now well established, there is arguably still a lack of empirical research looking at possible mitigation strategies across different fields of forensic science, including the human identification field. This chapter discusses some of the empirical research conducted on cognitive bias and forensic decision making, with a focus on human identification. Mitigation strategies and proposed solutions are explored and discussed, along with future avenues.

Keywords: Cognitive bias, Forensic science, Forensic anthropology, Human identification.

INTRODUCTION

For the last decade, forensic science methods and outputs have been subjected to scrutiny. Various government reports have highlighted the need for an improvement in transparency, quality control, and efficacy of forensic techniques [1, 2]. Concerns have also been raised globally regarding reproducibility, validity, as well as the risk of cognitive biases in human judgements [3]. As a result, there has been an increase in publications in relation to the scientific basis of forensic

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[*] **Corresponding author Sherry Nakhaeizadeh:** Department of Security and Crime Science, Centre for the Forensic Sciences, University College London, Gower Street, London WC1E 6BT, United Kingdom;
E-mail: sherry.nakhaeizadeh@ucl.ac.uk

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science and the application of forensic techniques in courts of law [4, 5]. What has been recognized is the complexity and nuances of decision-making involved in forensic science, given the critical role of humans in the interpretation of forensic science evidence.

Many of the identification methods used in forensic science are inherently subjective, relying mostly on visual interpretations of evidence, with the experience of the observer playing a key role in the accuracy of classification decisions and positive identifications [6]. The criticisms directed toward forensic science have therefore led some scholars to shift their research to focus on the forensic scientist and their evaluative interpretation approaches [7, 8], leading to the re-evaluation of the perceptual and decision-making processes involved in evidence interpretations [9]. This shift in research focus also encompasses the field of human identification [10, 11], including forensic anthropology [12 - 14].

Overall, the findings of the empirical studies conducted to date have shown a range of results with respect to the effects of cognitive bias and human factors in evidence interpretations [15]. Some studies have revealed the impact of contextual information on decisions and evaluations of relevant materials in a crime reconstruction scenario [16]. In other studies, scholars argue that bias arising from exposure to contextual information may not necessarily lead to biased interpretations and that contextual information can, on the contrary, assist forensic examiners in reaching more accurate conclusions [17, 18]. Many of the more recent published papers have proposed solutions to minimise the impact of cognitive bias on evidence interpretations [19, 20], focusing mainly on the concept of "task-relevant" information [21]. Although it is now well established that decision-making can be susceptible to cognitive bias in forensic science [22, 23], there is arguably still a lack of empirical research looking at possible mitigation strategies of the negative impacts across different fields of forensic science, including the human identification fields.

This chapter highlights the research being conducted in forensic science and cognitive bias, with a primary focus on human identification and forensic anthropology, respectively. It provides a brief overview of the psychological factors and mental processes that arguably impact evaluative judgements, followed by research on cognitive bias in forensic science and criminal investigations. Proposed solutions and mitigation strategies are discussed in addition to future avenues of research and innovation.

HUMAN DECISION MAKING

Psychologists have long established that humans do not process information objectively, but rather this process is characterised by our individual experiences and other contextual factors [24]. Some scholars have argued that our judgments and decision-making are influenced by a variety of cognitive processes allowing the brain to create "mental shortcuts" and strategies when organising and processing information, resulting in an ability to make fast, frugal, and accurate decisions [25, 26]. This process, although beneficial, eventually results in our choices and actions not functioning without being initiated by an initial stimulus [27]. Consequentially, information processing can be impacted by these limitations [28, 29]. A growing body of research in experimental psychology suggests that judgments and decisions are often affected by seemingly irrelevant factors, which can result in a failure to include important information in the decision-making process, and demonstrates that decision-making is not a rational, straightforward process [30]. This phenomenon of systematically deviating from rationality in decision making or judgments is referred to as cognitive bias [31].

Cognitive bias has, over the years, become an umbrella term for a number of distinct ways that expectations, motivations, and beliefs influence memory, selection, and evaluation of evidence [7]. The growing body of literature has distinguished between different external and internal factors that can impact decision-making, which are strongly related to time pressure [32], overconfidence [33], memory retrieval and storage [34], perceptions and expectations [35], and our motivations and pre-existing beliefs [36]. Research has also reported that human decision-makers often seek and interpret information in line with pre-existing beliefs [37]. This phenomenon is also known as confirmation bias, which is the tendency to selectively gather and process information to support a hypothesis [38]. More recently, research on cognitive bias has also focused on its impact in relation to expert domains [39].

Over time, experts become equipped with skills that are developed by repeated exposure to the task that they need to perform [40]. Experts can therefore come to "intuitive" conclusions by creating schemas through learning and experience without needing to be fully aware of the reason behind their thought processes or judgments [7]. This process also allows experts to be able to filter out noise and perform tasks relatively effortlessly, resulting in experts being efficient in their performances [41, 42]. For example, forensic experts rely to some extent on their knowledge and experience (in a manner consistent with other fields) when recognising and interpreting, for example, patterns and features when making comparisons. Equally, the limitations inherent with expertise, such as a lack of flexibility, may lead experts to miss important information [43] as well as being overconfident in their decision outcomes [44]. In forensic science and criminal investigations, the lack of transparency in decision-making and experts being influenced by cognitive factors has been a growing concern for the past decade [45].

CHAPTER 6

Regulating Investigative Genetic Genealogy

David Gurney[1,*]

[1] *Investigative Genetic Genealogy Center, Ramapo College of New Jersey, 505 Ramapo Valley Road, Mahwah, New Jersey 07430, USA*

Abstract: Investigative Genetic Genealogy (IGG) has emerged as a premier investigative tool for identifying human remains and perpetrators of violent crimes where other methods have failed. As the number of cases resolved with the help of IGG has increased, there have been concomitant moves to regulate the field by applying existing regulations where possible and creating new ones to address issues specific to IGG. Regulation has come from all levels—from the Department of Justice, state legislators, judges, district attorneys, non-profit organizations, forensic laboratories, and IGG practitioners themselves—and consists of direct and indirect regulation, and binding and non-binding rules. This chapter provides an overview of all forms of IGG regulation to date and traces a few possible directions for the future of IGG regulation.

Keywords: Forensic, Human remains, Investigative genetic genealogy, Regulation, Violent crime.

INTRODUCTION

Investigative Genetic Genealogy (IGG) is an investigative technique that uses genetic genealogy and publicly available information to arrive at hypotheses about the identity of an individual who may have committed a serious violent crime or whose remains are unidentified [1]. IGG first came to the public's attention in 2018 with the arrest of Joseph James DeAngelo for the Golden State Killer crimes and the identification of Michelle Marcia King as "Buckskin Girl" [2]. However, the idea of IGG had been rumbling around certain corners of the genetic genealogy world since at least 2010 [3]. Early forms of IGG were tried in 2011 and 2014 without success, and a "Living Doe" was identified through a form of IGG in 2015 [3 - 5]. This was the pioneering phase of IGG, when several brilliant individuals realized that the methods they had been using to help adoptees find their birth parents could be used to bring justice in cases of violent crime and unidentified human remains [6].

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[*] **Corresponding author David Gurney:** Investigative Genetic Genealogy Center, Ramapo College of New Jersey, 505 Ramapo Valley Road, Mahwah, New Jersey 07430, USA; E-mail: dgurney@ramapo.edu

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That early spirit has now seen the resolution of over 1,000 cases [7], with little to no direct regulation. It has also led to ethical and legal concerns about the use of IGG. In that vein, several state laws have been enacted since 2019, and more are likely to follow. Many IGG practitioners and professionals from adjacent fields have also recognized the need for regulation and have established organizations to address this need [1, 8]. It can be fairly said that IGG has moved from the pioneering era to an era of professionalization.

This chapter provides an overview of the regulation—both direct and indirect—of IGG to date, and it traces a few possible future directions for additional regulation.

THE REGULATORY LANDSCAPE FOR IGG

The current regulatory landscape for IGG consists of a spattering of state laws, agency guidelines, genetic genealogy database terms of service, constitutional requirements, and self-regulation by IGG practitioners. Some of these regulations present mandatory requirements for IGG practitioners, while others serve as guidelines and best practices. At this early stage in the development of IGG regulations, it is worthwhile to consider the overall regulatory goals and to observe how each regulatory model attempts to address one or more of those goals. This analysis will also provide the benefit of identifying gaps in the existing regulatory landscape.

Regulatory Motivations and Goals

In general, two goals can be identified within the existing regulatory landscape for IGG. First is the protection of the rights of criminal suspects and defendants. Second is the protection of the rights of genetic and genealogical relatives of criminal suspects and defendants.

Protecting the rights of criminal suspects and defendants is a perennial concern of United States law and policy. Indeed, it is foundational to the founding documents of the country, with the overall thrust of the Bill of Rights providing protection for ordinary citizens from the far greater power of the government [9]. Thus, it is no surprise to see this principle extended to the new investigative method of IGG. However, it remains an open question whether there are rights or interests implicated by the use of IGG that require greater – or even on-par – protections than are provided in the case of traditional DNA investigations and the use of publicly-available information in investigations that do not involve IGG. In any case, protections for the rights of criminal suspects and defendants are found in regulations that:

a. Require warrants and judicial oversight for all or part of an IGG case;
b. Mandate disclosure of all or part of the records, notes, and other materials collected and compiled in an IGG case;
c. Disallow ascertainment of physical and psychological traits or propensities from genetic information;
d. Create rules for retention and removal of genetic profiles and public records compiled in an IGG case (and penalties for failure to comply);
e. Provide a right to IGG for criminal defendants and criminal petitioners in postconviction; and
f. Potentially lead to the exclusion of all DNA evidence in a case when the regulation is not followed.

The reasoning behind these regulations is presented in the specific analyses below.

Somewhat (though not entirely) unique to IGG is the implication of the rights of genetic and genealogical relatives of criminal suspects and defendants, who are themselves entirely innocent. In contrast, there are other contexts where a criminal suspect or defendant's relatives may be swept up in the investigation – whether as witnesses or simply unwilling subjects of law enforcement and media attention. The number and extent of relatives for whom varying degrees of information are obtained and made use of in an IGG case are unprecedented [10, 11][2], including not only close relatives like parents and siblings, but distant relatives, extending to fourth, fifth, sixth, and even greater cousins [12].

Some of these relatives have uploaded their genetic profiles to sites, such as FamilyTreeDNA and GEDmatch, and have explicitly chosen to allow their information to be shared with law enforcement if they share DNA in common with that obtained from a crime-scene sample, but most have not. Most are relatives who are discovered through public records and other publicly available information, such as public social media posts [13]. Thus, there is at least *prima facie* reason to consider that the rights of these relatives deserve attention in any IGG regulation, and indeed, many regulations do take them into account. The protection of the rights of genetic and genealogical relatives of criminal suspects and defendants is found in regulations that:

a. Require IGG practitioners only to view the direct genetic relationship (in the form of amount of shared DNA, measured in centimorgans, "cM") of relatives who have uploaded their genetic profiles to FamilyTreeDNA and GEDmatch [14][3] and "opted in" to IGG searching;
b. Exempt IGG reports and other materials from FOIA-like [15][4] laws;
c. Allow for *in-camera* or sealed presentation of IGG reports and other materials

CHAPTER 7

Laboratory Methods and Bioinformatics for Investigative Genetic Genealogy

Ellen Greytak[1,*]

[1] *Parabon NanoLabs, Inc. Suite 406, Reston, 11260 Roger Bacon Dr., Virginia 20190, USA*

Abstract: Investigative Genetic Genealogy (IGG) is a powerful technique that combines DNA analysis with traditional genealogical research to narrow down the possibilities for the identity of an unknown individual. Since the arrest of the Golden State Killer in May 2018, IGG has been publicly acknowledged in 464 Unidentified Human Remains (UHR) and 318 perpetrator cases as of December 2023, and has been used in many more unacknowledged cases. Parabon alone has contributed to over 320 identifications, more than 1/3 of which have not been acknowledged publicly. A critical component of successful IGG is the quality of the genotypes generated from the forensic sample. This chapter will provide a brief overview of the IGG process, describe how data quality impacts IGG cases, and discuss various laboratory and bioinformatics techniques for generating high-quality genome-wide Single-Nucleotide Polymorphism (SNP) genotype data from challenging forensic DNA samples.

Keywords: Bioinformatics, DNA, Forensic genetic genealogy, Forensic genetics, Human identification, Investigative genetic genealogy, Single-nucleotide polymorphism.

INTRODUCTION

Investigative Genetic Genealogy (IGG) is a powerful technique that combines DNA analysis with traditional genealogical research to narrow down the possibilities for the identity of an unknown individual. Since the arrest of the Golden State Killer in May 2018, IGG has been publicly acknowledged in 464 Unidentified Human Remains (UHR) and 318 perpetrator cases as of December 2023 [1, 2]. It has been used in many more unacknowledged cases. Parabon alone has contributed to over 320 identifications, more than 1/3 of which have not been acknowledged publicly.

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[*] **Corresponding author Ellen Greytak:** Parabon NanoLabs, Inc. Suite 406, Reston, 11260 Roger Bacon Dr., Virginia 20190, USA; E-mail: ellen@parabon.com

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The IGG process has been described in detail in various studies [3 - 5], although a brief overview is included herein. A critical component of successful IGG is the quality of the genotypes generated from the forensic sample. This chapter discusses various laboratory and bioinformatics techniques for generating high-quality genome-wide Single-Nucleotide Polymorphism (SNP) genotype data from forensic DNA samples. Throughout this chapter, the unknown individual whose identity is being narrowed down using IGG will be referred to as the "Subject".

FORENSIC DNA ANALYSIS

Traditional forensic DNA analysis uses ~20 Short Tandem Repeats (STRs) to create an individual profile [6]. STRs are a type of genetic variant that differ in length according to the number of repeats of a nucleotide motif [6]. Determining which alleles a person has at each locus is called genotyping. Genotypes can be homozygous (two of the same allele) or heterozygous (two different alleles).

STR profiles provide an excellent mechanism for comparing an unidentified DNA sample to a known individual or to a database. However, beyond identity, they can only be used to detect close relationships because of the small number of loci. Single-Nucleotide Polymorphisms (SNPs) are changes in the DNA sequence at a particular site in the genome. Tens of millions of SNPs have been found in the human genome, and hundreds of thousands to millions of SNPs can be genotyped in a single assay.

Given the density of genome-wide SNP profiles, it is possible to use them to detect distant relationships, either on a one-to-one basis or by searching a database that contains other SNP profiles. Such a database is referred to as a Genetic Genealogy (GG) database. Generating genome-wide SNP genotype profiles from forensic samples can be challenging due to the low quantity and quality of the DNA, which can result in reduced numbers and accuracy of the SNP genotypes in the profile. In turn, this can affect the accuracy of the kinship inferences.

INVESTIGATIVE GENETIC GENEALOGY DATABASES

Investigative Genetic Genealogy (IGG) relies on the availability of GG databases that allow Law Enforcement (LE) usage, collectively referred to as "IGG databases." Currently, two such databases are available: GEDmatch and FamilyTreeDNA (FTDNA), both of which charge a fee for usage by LE, provide users with the specific option to opt-in or opt-out of LE matching, and prevent LE kits from appearing in non-LE users' match lists. In both cases, a DNA profile in the database is referred to as a "kit."

The GEDmatch database consists of individuals who have had their DNA tested by a Direct-To-Consumer (DTC) genomics provider, downloaded their raw data from that provider, uploaded it to GEDmatch, and consented to having their data used for matching. LE usage of GEDmatch is limited to “DNA obtained to identify remains of a deceased individual” or “DNA obtained and authorized by law enforcement to identify a perpetrator of a violent crime against another individual, where 'violent crime' is defined as murder, nonnegligent manslaughter, aggravated rape, robbery, or aggravated assault” [7]. Unidentified human remains (UHR) cases can use the entire public GEDmatch database, while perpetrator cases can only use the LE-opted-in portion.

The FTDNA database consists of voluntary uploads, as well as users who have had their DNA tested by FTDNA, who have also consented to having their data used for matching [8]. FTDNA currently limits LE usage of its database to “identify the remains of a deceased individual, regardless of whether or not the deceased individual is a suspected homicide victim”, as well as victims of violent crimes [9]. For FTDNA, a “violent crime” is identified as “any homicide (including murder or non-negligent manslaughter), any sex crime (including rape), or any of the following serious crimes or criminal offenses: (1) armed robbery, (2) aggravated assault, or (3) any other violent crimes as defined by the FBI Uniform Crime Reporting (UCR) Program” [9]. This includes attempted offences, and when there is an ongoing threat to public safety [9]. Both types of samples are compared only against the LE-opted-in portion of the database [8].

IGG databases consist of data produced by DTC genomics companies (*e.g.*, FTDNA, 23andMe, AncestryDNA, MyHeritage, collectively referred to as “DTCs”), and similar data must be produced from a forensic sample for it to be compatible with the database. DTCs use a variety of Illumina microarray chips to genotype ~500,000 - 900,000 SNPs across the genome, for a total of ~2.2 million SNPs across all the various chip versions. The two main chips that are currently used by DTCs, the OmniExpress and the Global Screening Array (GSA), both contain ~700,000 SNPs but only overlap by ~140,000 SNPs. As IGG databases allow uploads from multiple DTCs, they must accommodate data with a wide range of different SNP compositions. As long as new SNP data is consistent with microarray genotype data produced by one or more DTCs, it can be uploaded to IGG databases (Fig. **1**).

FINDING RELATIVES IN IGG DATABASES

When SNP data is uploaded to a GG database, it is compared to each profile in the database to determine whether they show signs of being genetically related. Genetic relatedness is determined by searching for Identical-By-Descent (IBD)

CHAPTER 8

Identification of Unknown Persons: Kinship and Forensic Investigative Genetic Genealogy

Bruce Budowle[1,*], **Jianye Ge**[2], **Swathi Kumar**[3] and **Antti Sajantila**[1,4]

[1] *Department of Forensic Medicine, University of Helsinki, Helsinki, Finland*

[2] *Department of Microbiology, Immunology & Genetics and Center for Human Identification, University of North Texas Health Science Center (UNNTHSC), 3500 Camp Bowie Blvd., Fort Worth, Texas 76107, USA*

[3] *QIAGEN Sciences, 19300 Germantown Rd, Germantown, Maryland 20874, USA*

[4] *Forensic Medicine Unit, Finnish Institute for Health and Welfare, Helsinki, Finland*

Abstract: Identification of unknown persons, be they missing or unidentified, is a significant challenge, an important humanitarian responsibility, and a fundamental part of the healing process for families and communities. The only resolution for the families (and friends) who have lost a relative is credible confirmation of the death of their family member (and friend) and that the remains of their relative (and friend) will be treated with dignity, in accordance with their culture, religious beliefs, and the laws of their respective countries. Forensic evidence can play a significant role in the humanitarian and investigative processes. Of the various forensic tools to assist in the identification process, DNA is particularly versatile because it can be found in all tissues of the human body and thus does not rely on a specific target tissue, as do, for example, fingerprints and dental records. Moreover, in the absence of antemortem samples or records, DNA identification can be carried out by kinship analyses as long as family reference samples are available. The concepts and foundations of relationship testing are well-established and described herein. Current DNA typing capabilities have been a mainstay in unknown person identifications and in disaster victim identifications. However, DNA technologies and databases have advanced substantially in the past few years to enhance the scope and reach of kinship analyses. Expanded technology capabilities (high throughput sequencing and concomitant bioinformatics) and additional types of DNA databases (genetic genealogy) enable higher accuracies in kinship testing, association with more distant relatives, and greater typing success due to analysis of lower quantity and quality samples. Combining kinship data with access to public records and enhanced bioinformatic and computational power enables forensic investigative genetic genealogy to provide better success of humanitarian services for families and communities, as well as identification of the source(s) of evidence found at crime scenes.

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[*] **Corresponding author Bruce Budowle:** Department of Forensic Medicine, University of Helsinki, Helsinki, Finland; E-mail: b.budowle@att.net

Tracey Leigh Dowdeswell (Ed.)

Keywords: Databases, Genetic genealogy, Human identity testing, Human remains, Identity by descent, Kinship, Missing persons, SNPs, Unknown persons, Victim identification.

INTRODUCTION

People who go missing, either forced, intentionally, by losing their identity, or dying under various circumstances, is a worldwide problem that continues to plague the world in the 21st century. The continued perilous world circumstances, such as war, forced migrations, human trafficking, mass disasters, murder, climate change, poverty, and organized crime, to name a few, have contributed substantially to poor human conditions and loss of life [1, 2].

A missing person can be defined as a person who is not physically present, and the whereabouts and fate of that person are unknown. In contrast, an unidentified person can be defined as a person who is physically present, but the identity of that person is unknown [3]. Missing persons and unidentified persons/human remains may be described interchangeably or generally as unknown persons herein, as essentially the same principles and practices can be used to determine identity. Some of those who are missing have died and need to be identified. Indeed, unidentified persons can be considered part of a “mass disaster over time” without the attention that mass disasters attract (note that it has also been termed “The Nation’s Silent Mass Disaster”) [4].

The only resolution for the families (and friends) in such circumstances is credible confirmation of the death of their family member (and friend) and that the remains of their relatives will be treated with dignity, in accordance with their culture, religious beliefs, and the laws of their respective countries [5]. The identification of these human remains is an important humanitarian responsibility and is a fundamental part of the healing process for families and communities. Furthermore, recovery and examination of human remains can be part of a medicolegal investigation to determine the cause and/or manner of death or if there are substantial public health-related concerns [6 - 8]. In addition, identification of remains can be the first step in a criminal investigation to determine the perpetrator if, for example, homicide was the manner of death. Forensic evidence can play a significant role in these important humanitarian and investigative processes. Forensic laboratories, however, do not make official determinations of identity; the responsibility of official identification of human remains is a legal determination assigned to a jurisdictional authority that should rely on all available forensic data and contextual evidence [9]. Forensic science, and in particular DNA, does contribute substantially to the identification process, and, in some cases, serves as the sole evidence of the identity of an unknown person.

In recent years, there have been notable developments in the forensic DNA discipline that can and have made identifications of unknown persons possible where standard methods have not been successful. This chapter describes the process of missing persons identification, the genetic underpinnings of kinship analyses, limitations of current technology, recent advances, such as high-throughput DNA sequencing technologies, and innovative databases that support identification processes and enhance capabilities to identify unknown persons *via* DNA analyses.

While the concepts and foundations of relationship testing are far from novel [10], it is the expanded technology capabilities (high-throughput sequencing and concomitant bioinformatics) and additional types of DNA databases (genetic genealogy) that will enable higher accuracies in kinship testing, association of more distant relatives, and greater typing success due to analysis of lower quantity and quality samples. Combining kinship data with access to public records and enhanced bioinformatic and computational power have enabled forensic investigative genetic genealogy (FIGG) to provide better success of humanitarian services [11, 12] for families and communities as well as identification of the source(s) of evidence found at crime scenes [13, 14].

FORENSIC PROCESSES OF IDENTIFICATION WITH EMPHASIS ON DNA

To exploit the power of forensics for identification, information needs to be collected all the way from the scene through to the authority that makes the official determination of the identity of an unknown individual. If at all possible, metadata regarding an unknown person should be gathered to include general personal information, physical appearance, medical and dental history, distinguishing features, clothing worn, and any personal items when last seen or when found, and any circumstances that may be related to the disappearance [3, 9, 15]. Such information can assist authorities when rendering an official identity declaration. Protocols for the proper and effective recovery of remains and associated items, including but not limited to documentation of recovery, chain of custody, security, transport, and storage, should be implemented and performed according to best practices. These best practices should account for exigent circumstances that may limit the collection of remains as well as metadata.

Once evidence and remains have been collected, multiple forensic techniques are used to examine and assist in identifying an unknown person [6, 14]. These techniques may include dactyloscopy, anthropology, odontology, tattoos, medico-

CHAPTER 9

Beyond Codis: Clearing Cases Using Genetic Data from Rootless Hair

Taryn Mulvihill[1] and **Tracey Leigh Dowdeswell**[2,*]

[1] *Cognitive Systems, University of British Columbia, BC, Canada*

[2] *Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada*

Abstract: This chapter reviews developments in forensic DNA that enable case clearance using genetic data from rootless hair. Advances in Massively Parallel Sequencing (MPS) have enabled the production of DNA profiles of Single-Nucleotide Polymorphisms (SNPs). The resulting SNP profiles can then be used to identify persons of interest – usually unidentified decedents or suspects in criminal cases – using Forensic Investigative Genetic Genealogy (FIGG). FIGG uses genetic genealogy databases of SNP profiles to generate relatedness estimates to identify persons of interest. Once candidates are identified through FIGG, the identification is normally confirmed through the direct comparison of Short Tandem Repeat (STR) forensic DNA profiles – either through direct comparison for crime scene samples, or through kinship testing for human remains. However, SNP profiles can be generated in cases in which the DNA evidence exists in too low quantities or is too degraded to generate a traditional (STR) forensic DNA profile. This allows for cases to be cleared, even where an STR DNA profile is not available to be uploaded and searched against CODIS databases. Novel statistical methods are available for direct comparison between SNP profiles, which enables the probative value of the evidence to be assessed by triers of fact in criminal cases. In this chapter, we focus on the clearance of cases using rootless hair, as these samples are commonly found in criminal and human remains investigations but have resisted conventional DNA analyses. Forensic DNA analysis is therefore poised to move beyond CODIS, and this poses novel challenges for courts and legal professionals.

Keywords: CODIS, Forensic DNA, Forensic investigative genetic genealogy, Forensic genomics.

INTRODUCTION

Over the past few decades, advances in forensic genetics have transformed the way we use DNA to solve investigations and prosecute criminal offences. At the

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[*] **Corresponding author Tracey Leigh Dowdeswell:** Department of Humanities & Social Sciences, Douglas College, 700 Royal Avenue, New Westminster, BC V3M 5Z5, Canada; E-mail: dowdeswellt@douglascollege.ca

Tracey Leigh Dowdeswell (Ed.)

core of this lies the Combined DNA Index System (CODIS), a database designed to store and search DNA profiles [1]. As of February 2024, the CODIS-NDIS database in the US had generated almost 700,000 'hits' in over 680,000 separate investigations [1].

As powerful as CODIS is, there are still cases that cannot be solved using CODIS and traditional forensic DNA methods. One of the limitations of the CODIS system is that it is difficult to generate forensic STR profiles from small amounts of DNA or DNA that is highly degraded [2]. Rootless hair, for example, resists conventional genetic analyses yet is commonly found at crime scenes and with unidentified human remains [2].

Recent advances in laboratory methods, bioinformatics, and computational applications are beginning to solve cases that are beyond the scope of CODIS. This chapter will focus on current developments in using rootless hairs to obtain SNP profiles to identify persons of interest – including suspects in criminal cases as well as unidentified decedents – which can be used in cases where there is insufficient DNA to develop a traditional STR DNA profile. New statistical methods enable the comparison of SNP profiles so that the probative value of the evidence may be evaluated by finders of fact.

In this chapter, we first review CODIS and STR-DNA typing and some of their inherent limitations. Next, we discuss some of the leading alternatives to clearing cases using DNA, including familial searching in CODIS databases, Massively Parallel Sequencing (MPS), and Forensic Investigative Genetic Genealogy (FIGG). We then review IBDGem, a method of comparing SNP DNA profiles to assess their relatedness, which can generate Log-Likelihood Ratios (LLRs) for samples where STR profiling is not possible. Finally, we review some recent cases in which criminal suspects and human remains have been identified using only SNP profiles generated from rootless human hair. These novel techniques have the potential to clear many currently unsolvable cases and provide reliable evidence for use in legal proceedings.

This promise will also come with significant challenges. Criminal justice professionals – police, coroners, medical examiners, judges, and lawyers – will need to be trained to understand and apply these new methods. With previous advances in forensic DNA, powerful technologies that enable us to collect and identify trace evidence located at crime scenes pose new challenges for privacy, for regulation, and for understanding just how, when, and under what circumstances trace evidence has been left at crime scenes.

CODIS

The most common method for DNA-based identification involves sequencing autosomal DNA from biological samples collected from crime scenes or from unidentified decedents [3]. Certain hypervariable regions of the genome – known as Short Tandem Repeats (STR) – are highly individualizing and easily amplified through Polymerase Chain Reaction (PCR), followed by capillary electrophoresis [4]. Even a small panel of STR loci is sufficient for genome mapping and determining genetic relatedness, and this has enabled STR DNA typing to become one of the most reliable methods of identification [4 - 6].

Less commonly, the mitochondrial genome may be sequenced to identify a maternal lineage. Mitochondrial DNA (mtDNA) – in contrast to autosomal DNA – is not individualizing. Since mtDNA mutates very slowly, it can identify a group of individuals who share the same haplotype through a common line of matrilineal descent, often going back thousands of years [7]. An mtDNA profile may be used by investigators to exclude suspects, narrow down investigative leads, and provide additional evidence regarding identity [7].

STR typing also forms the basis of the CODIS database system. The Federal Bureau of Investigation (FBI) began working on CODIS in 1990; it is still the largest working database of STR profiles, and it has been adopted in countries around the world [8]. In the United States, the DNA Identification Act of 1994 authorizes forensic DNA laboratories to store DNA profiles from crime scenes, offenders, and missing persons [9]. CODIS operates on a 3-tier hierarchy, with the highest tier being the National DNA Index System (NDIS) [8]. The State DNA Index System (SDIS) and the Local DNA Index System (LDIS) contain profiles from their respective geographical areas [8].

To make an identification using a CODIS database, an unknown sample is first sequenced to produce an STR DNA profile and is then searched against a CODIS index of DNA samples from previously convicted offenders, crime scenes, or missing persons [8]. When a profile hit is not obtained on the SDIS or LDIS level, laboratories may submit the profile to the NDIS. For submission to the NDIS, samples must meet certain qualify assurance standards to ensure stringency in the index as required by the Quality Assurance Standards for Forensic DNA Testing and DNA Databasing Laboratories [10]. As of January 1st, 2017, the CODIS core loci consist of 20 loci as well as amelogenin [8]. The NDIS contains several indices to separate samples: Forensic (biological samples), Offender (arrestees and convicted offenders), Missing Person, Relative of Missing Person, and Unidentified Human Remains [8].

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SUBJECT INDEX

Tracey Leigh Dowdeswell (Ed.)

Tracey Leigh Dowdeswell

Tracey is a lawyer and a legal scholar who studies the forensic applications of bioinformatics and data science in the criminal justice system, with an emphasis on forensic genetics. She has a degree in biological sciences from the University of Alberta, a master's degree in liberal arts from Simon Fraser University, degrees in civil and common law from McGill University, and an LLM and a PhD from Osgoode Hall Law School of York University. She teaches criminal law, victimization, and the law of evidence. She has authored several books and articles on forensic evidence in the legal system. She is the co-author, with contributor Sean Goltz, of The Imaginationless Generation: Lessons from Ancient Culture on Regulating New Media (Brill, 2019), and Real-World AI Ethics for Data Scientists: Practical Case Studies (Chapman & Hall/CRC, 2023) for the Routledge Data Science Book Series.