
Forensic Genetics and Kinship Analysis: A Survey

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Abstract

Forensic genetics, a multidisciplinary field, applies genetic analysis to legal investigations, focusing on kinship analysis and DNA profiling using genetic markers such as SNPs and STRs. This survey examines the evolution of forensic genetics, highlighting its applications and the integration of advanced methodologies like omics technologies and probabilistic models. The field's expansion into nonhuman DNA analysis and the use of genetic genealogy have revolutionized the resolution of cold cases and missing persons identification. However, these advancements raise ethical and privacy concerns, particularly regarding genetic data misuse and public trust erosion. The survey underscores the importance of robust legal frameworks and ethical guidelines to safeguard privacy rights while leveraging genetic technologies for effective forensic investigations. Future directions include enhancing data security, integrating omics technologies, and developing comprehensive legal standards. Continued research, standardization, and training are vital for addressing challenges and ensuring forensic genetics' efficacy and ethical application in legal contexts.

1 Introduction

1.1 Concept of Forensic Genetics

Forensic genetics applies genetic analysis within legal contexts, focusing on genetic findings related to activity level propositions [1]. This multidisciplinary field utilizes genetic principles to address legal inquiries, especially when conventional autosomal DNA profiling is insufficient [2]. It involves studying genetic markers, including X-chromosomal markers, relevant in both population and forensic genetics [3]. Forensic genetics also incorporates genetic information from databases, such as those used in genetic genealogy, to aid criminal investigations [4].

The discipline is crucial in population history and genome-wide association studies, demonstrating its broader applicability beyond traditional forensic settings [5]. The concept of 'relevant population' in Bayesian forensic inference is vital for presenting forensic evidence in court, emphasizing the need to contextualize genetic findings within appropriate population frameworks [6]. Forensic genetics is fundamental in identifying individuals and resolving complex legal cases, thereby enhancing the legal system's ability to address identity and kinship issues.

1.2 Significance in Legal Investigations

Forensic genetics is essential in contemporary legal investigations, primarily due to its ability to analyze extensive genetic data, which enhances suspect identification and crime resolution [7]. The field has expanded to encompass nonhuman DNA analysis, presenting both novel opportunities and challenges [8]. Integrating forensic genomics into criminal investigations leads to more accurate and efficient case resolutions, promoting just outcomes [9]. The estimation of individual admixture (IA) from genetic data is particularly noteworthy, providing insights into genetic backgrounds that aid in resolving complex legal cases [5].

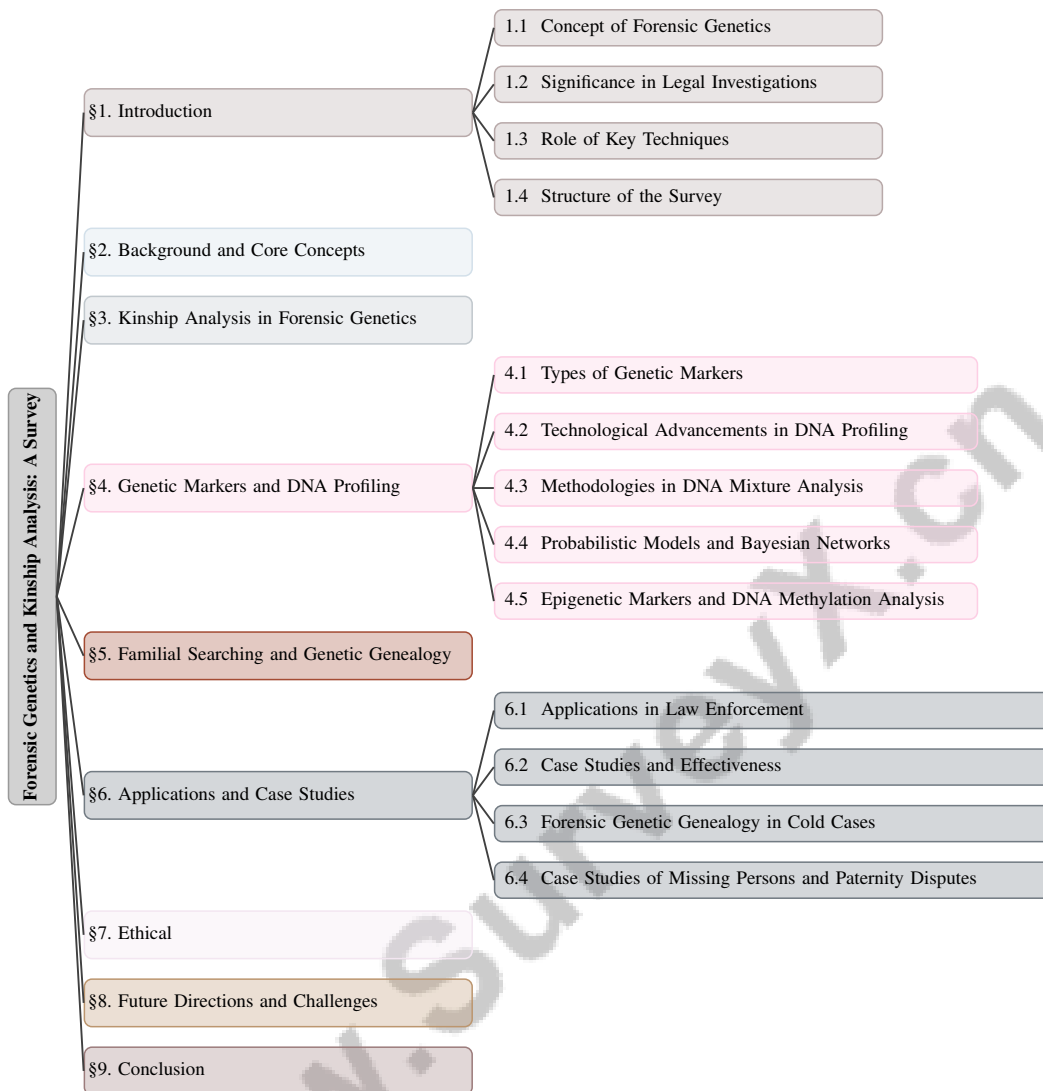


Figure 1: chapter structure

Beyond source attribution, forensic genetics enables transparent inference, which is crucial for informed judicial decision-making [1]. Reliable genetic evidence is vital for crime-solving and individual identification, underscoring the field's importance in legal proceedings. It also addresses critical variables such as DNA transfer, persistence, prevalence, and recovery (DNA-TPPR), which are essential for understanding DNA evidence nuances in criminal investigations [10].

However, the use of genetic genealogy, particularly through direct-to-consumer genetic testing services, raises significant privacy and consent concerns that must be managed within legal frameworks. These services analyze genetic data to identify familial relationships, which, while beneficial for crime-solving, necessitate careful consideration of ethical and privacy implications. The challenge of defining a relevant population in Bayesian forensic analysis complicates the assessment of evidence linking suspects to crimes, highlighting the need for precise forensic practices [6].

1.3 Role of Key Techniques

Kinship analysis is fundamental in forensic genetics, employing genetic markers to elucidate familial relationships and lineage. The use of X-chromosomal markers offers unique insights due to their distinct inheritance patterns, differing from autosomes and Y chromosomes [3]. This technique is particularly useful when direct parental DNA is unavailable, requiring indirect inference of familial

connections [11]. Y-STR haplotyping is invaluable for identifying paternal lineage, leveraging the Y chromosome's inheritance to trace male ancestry in forensic samples [2].

Genetic markers, including single nucleotide polymorphisms (SNPs), are vital in DNA profiling, enabling precise individual identification and resolution of complex forensic cases [12]. The inclusion of SNPs enhances genetic data resolution, providing a robust framework for forensic inference. Moreover, genetic genealogy methodologies have transformed forensic investigations by utilizing direct-to-consumer genetic databases to trace lineage and identify suspects, while raising ethical considerations regarding informed consent and privacy [4].

Familial DNA searching and forensic genetic genealogy have become powerful tools for law enforcement, particularly in solving cold cases. These techniques utilize high-density SNP profiles to infer distant familial relationships, connecting individuals through shared genetic traits [12]. However, the use of genetic genealogy databases by law enforcement necessitates careful ethical scrutiny regarding access rights and consent issues [13].

Incorporating DNA methylation analysis into forensic genetics provides additional insights, offering reliable biomarkers for age estimation, which enhances investigative processes by revealing individuals' biological ages [14]. DNA databases play a crucial role in the criminal justice system, serving as repositories of genetic information that facilitate offender identification and prosecution [9]. Collectively, these techniques underscore the precision and effectiveness of forensic genetics, reinforcing its critical role in modern legal investigations.

1.4 Structure of the Survey

This survey is structured to comprehensively examine forensic genetics and its application in legal investigations. It begins with an introduction to forensic genetics, emphasizing its significance and key techniques, including kinship analysis, genetic markers, DNA profiling, familial searching, and genetic genealogy. Following the introduction, the survey explores the historical development and fundamental principles of forensic genetics, highlighting its importance in the criminal justice system. It outlines the evolution of forensic genetic technologies, which have become increasingly integral to criminal investigations over the past 35 years. Key terminology and concepts are defined, showcasing forensic genetics' role in identifying biological evidence, such as DNA profiles, and discussing its implications for civil and criminal case resolution. Recent advancements in genetic genealogy techniques and their applications in cold case investigations, particularly in the UK, are also addressed, where publicly available genetic data can enhance forensic inquiries [9, 15].

Subsequent sections delve into specific areas of forensic genetics. Section 3 focuses on kinship analysis, discussing methods, applications, challenges, and limitations. Section 4 examines genetic markers in DNA profiling, including types of markers, technological advancements, methodologies in DNA mixture analysis, and the use of probabilistic models and Bayesian networks, alongside epigenetic markers and DNA methylation analysis.

Section 5 addresses familial searching and genetic genealogy, exploring investigative techniques and the significant ethical and privacy dilemmas they present. This section highlights the implications of law enforcement's access to public genealogy databases, such as GEDmatch, utilized in solving cold cases like the Golden State Killer investigation. It also discusses legal challenges posed by non-forensic genetic data use, potential privacy intrusions, and broader societal concerns regarding consent and the protection of genetic information [16, 7, 15, 17]. Section 6 presents applications and case studies, emphasizing forensic genetics' role in law enforcement, effectiveness of case studies, and applications in resolving missing persons and paternity disputes.

Section 7 provides a thorough examination of the ethical, legal, and social implications of forensic genetics, detailing issues such as legal and ethical considerations surrounding public genetic databases, regulatory challenges of integrating forensic genomics in law enforcement, security vulnerabilities of these databases, and broader social impacts, including public trust in genetic data usage. This section highlights the evolving landscape of forensic genetics, particularly regarding recent advancements like genetic genealogy and trait prediction, which raise significant privacy and consent issues extending beyond criminal justice [18, 7, 17]. Section 8 identifies future directions and challenges in forensic genetics, including emerging technologies, interdisciplinary applications, and the importance of standardization and training.

The conclusion summarizes key points discussed, reinforcing the importance of forensic genetics and the need for ongoing research and ethical considerations. The survey evaluates current research categorization into police genealogy and predictive forensic genetics, particularly concerning their ethical, legal, and social implications [18]. The following sections are organized as shown in Figure 1.

2 Background and Core Concepts

2.1 History and Evolution of Forensic Genetics

Forensic genetics has evolved significantly, transitioning from basic DNA profiling to sophisticated forensic genomic applications leveraging extensive genetic datasets [7]. This transformation began with the advent of DNA profiling techniques, which have become indispensable in legal contexts [19]. The field has expanded to include nonhuman genetic evidence, reflecting its adaptability and the technological progress that has broadened its scope [8]. Advances in genetic technologies have enhanced the precision and applicability of forensic genetics, influencing modern crime governance strategies and investigative practices [9]. Historical perspectives on genetic privacy and public attitudes towards genetic data have shaped the field's development, underscoring the importance of ethical considerations [20]. The integration of personalized medicine and omics into forensic methodologies represents a significant milestone, demonstrating the field's capacity to incorporate cutting-edge scientific advancements [21]. Additionally, data sharing in human ancient DNA studies has been pivotal for scientific advancement, presenting both challenges and opportunities for forensic genetics' development [22]. These developments illustrate the dynamic nature of forensic genetics and its continuous evolution in response to technological innovations and societal needs.

2.2 Key Concepts and Definitions

Forensic genetics is anchored in several key concepts essential for its legal applications. Kinship analysis, utilizing genetic markers, is fundamental for determining familial relationships, particularly in complex cases involving collateral relatives [3]. Genetic markers such as short tandem repeats (STRs), single nucleotide polymorphisms (SNPs), and insertions-deletions (INDELs) are crucial for DNA profiling, enabling precise individual identification. Familial DNA searching extends traditional DNA profiling by identifying potential relatives of a DNA sample donor, playing a vital role in criminal investigations through indirect familial connections [7]. This technique closely relates to forensic genetic genealogy, which combines genetic data analysis with genealogical research to trace familial ties and identify individuals through shared genetic traits. Specific genetic markers, such as the Y chromosome and mitochondrial DNA (mtDNA), trace patrilineage and matrilineage, respectively, providing insights into genetic genealogy and trait inheritance through parental lines. These markers are particularly useful for identifying male contributors in mixed samples. DNA methylation analysis is employed to estimate biological age, offering valuable identification information [14].

Forensic genetics also involves analyzing DNA mixtures, utilizing statistical models like the multi-variate Dirichlet-multinomial distribution and the α -correction to interpret complex DNA evidence [23]. Bayesian networks enhance the sensitivity of inferences drawn from forensic genetic data by accounting for population heterogeneity [24]. The field benefits from concepts such as haplotypes and population dynamics, explained through models like the Fisher-Wright model, which are crucial for understanding population genetic structures and their forensic implications [25]. The integration of personalized medicine and molecular autopsy into forensic investigations reflects the field's adaptability to incorporate scientific advancements [21]. Ethical considerations, including informed consent and privacy rights, are critical in forensic genetics, particularly concerning genetic genealogy and law enforcement's use of genetic data. Challenges associated with data sharing, such as potential misuse and conflicts of interest, further emphasize the need for ethical vigilance in forensic genetic research and applications [22].

In recent years, the field of forensic genetics has evolved significantly, particularly in the area of kinship analysis. This evolution is underscored by a variety of methods and applications that have emerged, each accompanied by distinct challenges. To elucidate these complexities, Figure 2 presents a comprehensive overview of the hierarchical structure of kinship analysis. This figure categorizes traditional and modern techniques, as well as advancements in omics technologies, while also addressing the critical issue of data sharing within legal contexts. Furthermore, it delineates the

challenges that researchers face, which are classified into computational and methodological issues, ethical and quality concerns, and potential solutions. This visual representation not only enhances our understanding of kinship analysis but also underscores its significance in forensic investigations, illustrating the intricate interplay between technological advancements and ethical considerations in this vital field.

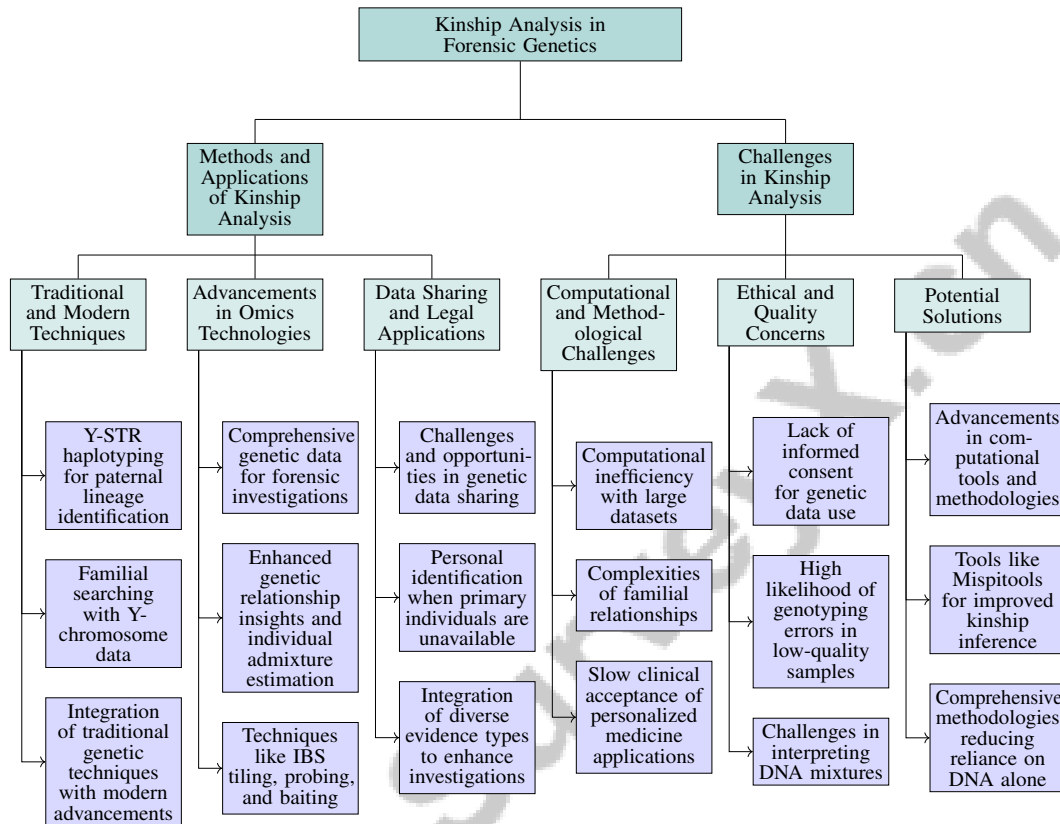


Figure 2: This figure illustrates the hierarchical structure of kinship analysis in forensic genetics, categorizing the methods and applications alongside the challenges faced. It highlights traditional and modern techniques, advancements in omics technologies, and data sharing within legal contexts. Challenges are divided into computational and methodological issues, ethical and quality concerns, and potential solutions, emphasizing the complexity and importance of kinship analysis in forensic investigations.

3 Kinship Analysis in Forensic Genetics

3.1 Methods and Applications of Kinship Analysis

Kinship analysis in forensic genetics employs diverse methodologies to elucidate familial relationships, integrating traditional genetic techniques with modern advancements. Y-STR haplotyping is pivotal for identifying paternal lineages, leveraging the Y chromosome's inheritance patterns to trace male ancestry and identify contributors in forensic samples [2]. This is complemented by familial searching approaches using Y-chromosome data to establish genealogical connections [2].

Advancements in omics technologies have enriched kinship analysis by providing comprehensive genetic data for forensic investigations [21]. These methodologies enhance genetic relationship insights and individual admixture estimation, improving the precision of genetic inferences [5]. Techniques such as IBS tiling, probing, and baiting exploit genetic genealogy services to identify shared segments, revealing critical genotype information in forensic contexts [26].

Data sharing methods in ancient DNA research have offered valuable insights into the challenges and opportunities of genetic data sharing [22]. Such knowledge is crucial for optimizing data sharing practices and ensuring forensic investigation integrity. These methodologies underscore kinship analysis's essential role in forensic genetics, particularly in complex legal cases. They facilitate personal identification when primary individuals are unavailable, integrating diverse evidence types, including genetic markers and genealogical databases, to enhance forensic investigations. The growing demand for sophisticated kinship analysis highlights its importance in ensuring judicial fairness and advancing investigative techniques [27, 28, 15, 11, 16]. By merging traditional and modern genetic tools, kinship analysis continues to evolve, providing increasingly precise insights into familial relationships.

As illustrated in Figure 3, the hierarchical structure of kinship analysis in forensic genetics is depicted, emphasizing the integration of genetic techniques, data sharing methods, and their applications. The first image presents ROC curves for two pedigrees, illustrating the trade-off between true positive and false positive rates in kinship determination, thereby emphasizing accuracy in genetic profiling. The second image depicts a family tree, foundational for tracing lineage and understanding genetic inheritance patterns, which aids in identifying familial links crucial for forensic and genealogical studies. The third image explores probabilistic relationships between genetic events, highlighting the analytical complexity required in kinship analysis. Collectively, these examples underscore the diverse techniques that make kinship analysis indispensable in forensic science [28, 15, 23].

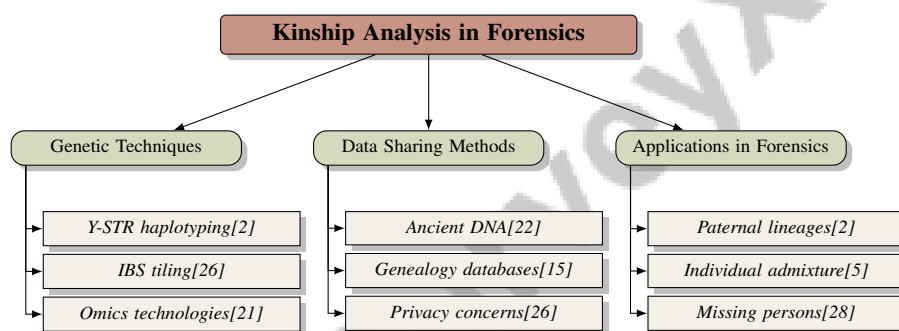


Figure 3: This figure illustrates the hierarchical structure of kinship analysis in forensic genetics, highlighting the integration of genetic techniques, data sharing methods, and their applications. Key genetic techniques include Y-STR haplotyping, IBS tiling, and omics technologies, which are essential for identifying familial relationships. Data sharing methods focus on ancient DNA, genealogy databases, and privacy concerns. The applications in forensics emphasize tracing paternal lineages, estimating individual admixture, and assisting in missing persons cases.

3.2 Challenges in Kinship Analysis

Kinship analysis in forensic genetics faces several challenges that hinder accurate familial relationship determination. A major issue is the computational inefficiency of existing methods, which struggle with the dynamic nature of Identity-By-Descent (IBD) graphs, especially when handling large datasets [29]. This is compounded by the complexities of familial relationships, where benchmarks often fail to account for the high probability of misidentification among distant relatives [30].

Integrating personalized medicine into kinship analysis introduces further challenges, as the slow clinical acceptance of these applications complicates the adoption of new methodologies [21]. Ethical concerns arise from the lack of informed consent for using genetic data by law enforcement, potentially increasing surveillance and distrust among minority groups [18].

Moreover, the high likelihood of genotyping errors in low-quality DNA samples, such as those from degraded biological traces, can significantly distort accurate genetic profiles. This distortion complicates forensic analysis and undermines the reliable interpretation of DNA mixtures, where multiple contributors' profiles may be obscured. Techniques like shotgun DNA sequencing can mitigate these challenges by analyzing all DNA fragments in a sample, enabling SNP profiles from compromised samples. However, the stochastic nature of low-quality DNA can lead to frequent locus drop-outs and insufficient SNP genotypes, complicating the accurate representation of individual contributions. Probabilistic models and likelihood ratio calculations are essential in forensic genetics

to account for potential genotyping errors and enhance DNA evidence interpretation [31, 24, 1, 32, 33]. These challenges are particularly problematic when the number of contributors and their relative contributions are unknown, leading to potential inaccuracies in DNA evidence interpretation.

Despite these obstacles, advancements in computational tools and methodologies offer potential solutions. Tools like Mispitools, which integrate genetic and non-genetic evidence, significantly improve kinship inference accuracy by systematically evaluating diverse evidence lines—such as pigmentation traits, biological sex, and age—through a likelihood ratio approach. This comprehensive methodology reduces reliance on DNA alone, enhancing investigative workflows in missing persons searches and addressing kinship analysis complexities, particularly in cases where key individuals are unavailable for testing [12, 28, 11, 32, 30]. Addressing these challenges requires ongoing research, improved methodologies, and enhanced training for forensic practitioners to ensure effective kinship analysis in forensic genetics.

4 Genetic Markers and DNA Profiling

4.1 Types of Genetic Markers

In forensic genetics, genetic markers are crucial for identifying individuals and understanding familial ties. Single Nucleotide Polymorphisms (SNPs) are prevalent due to their abundance and ability to provide detailed genetic insights. The selection of markers and reference populations is vital for accurate admixture estimation [5]. Short Tandem Repeats (STRs), with their high polymorphism, are instrumental in differentiating individuals, especially in complex kinship analyses [11]. Y-chromosomal STRs (Y-STRs) are particularly useful for tracing paternal lineages, offering advantages in cases lacking direct paternal DNA [2]. The emphasis on appropriate marker selection and reference populations is essential for resolving complex cases and enhancing forensic precision [5].

Advancements in computational methodologies, such as the Hash-based Framework for Dynamic Graphs (HFDG), have improved the efficiency of analyzing complex genetic relationships [11]. As forensic genetics advances, integrating innovative technologies and methodologies will enhance investigation precision, including applications to nonhuman genetic material, while addressing ethical and legal considerations. The development of novel genetic markers and analytical techniques positions forensic genetics as a vital tool in crime prevention and resolution [8, 7, 1].

4.2 Technological Advancements in DNA Profiling

Technological advancements in DNA profiling have greatly improved the precision and reliability of genetic analysis, offering promising solutions for forensic investigations. The integration of novel chip technology and omics research has increased the accuracy and efficiency of genetic profiling [19]. Shotgun DNA sequencing, which analyzes all DNA fragments in a sample, is particularly beneficial for low-quality samples from crime scenes or ancient remains [31]. This technique enhances the detection and interpretation of mixed DNA samples, improving forensic analysis accuracy.

As illustrated in Figure 4, the evolution of DNA profiling technologies reflects the field's adaptability to integrate scientific breakthroughs, such as genealogy databases for criminal investigations and personalized medicine applications. This figure highlights not only the novel techniques and applications but also the ethical considerations that accompany these advancements. The adaptability of these technologies addresses emerging ethical concerns and extends genetic analysis applications beyond human DNA [16, 1, 21, 8]. As these technologies progress, they will further bolster forensic genetics capabilities, ensuring continued relevance and efficacy in legal contexts.

4.3 Methodologies in DNA Mixture Analysis

DNA mixture analysis is critical in forensic genetics, addressing complexities from multiple individuals in a single sample. The Multiple Population Evolutionary Algorithm (MEA) employs a guided mutation operator to enhance deconvolution, providing a robust framework for resolving individual contributions [33]. The multivariate Dirichlet-multinomial distribution (MDM) evaluates allele count probabilities in mixed samples, with γ -correction enhancing reliability in diverse populations [23]. These methodologies integrate advanced statistical techniques, improving the accuracy of identifying contributors and strengthening DNA evidence in legal investigations.

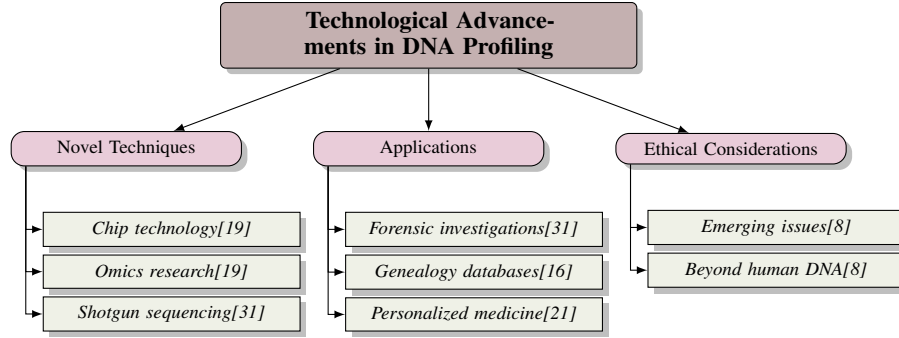


Figure 4: This figure illustrates the key technological advancements in DNA profiling, highlighting novel techniques, applications, and ethical considerations. It emphasizes the integration of chip technology and omics research, the application in forensic investigations and genealogy databases, and the ethical implications of extending genetic analysis beyond human DNA.

The integration of probabilistic models and personalized medicine approaches is set to revolutionize forensic analysis, allowing tailored interpretations that consider unique case specifics [8, 1, 21, 10, 14]. As forensic genetics evolves, developing and refining these methodologies will remain pivotal in addressing challenges posed by complex DNA evidence.

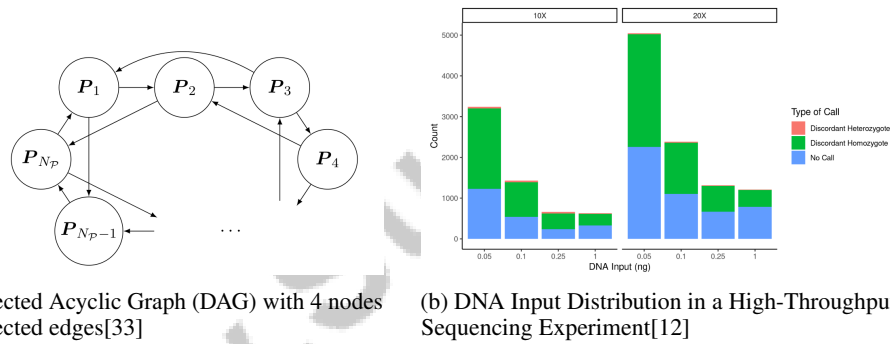


Figure 5: Examples of Methodologies in DNA Mixture Analysis

As illustrated in Figure 5, the exploration of genetic markers and DNA profiling has significantly advanced methodologies in DNA mixture analysis. The DAG illustrates the structured nature of data processing, emphasizing directed pathways in analyzing complex genetic information. The bar chart of DNA input distribution provides insights into variability in DNA input levels and categorizing samples into different genetic calls, highlighting the intricate methodologies used in DNA mixture analysis.

4.4 Probabilistic Models and Bayesian Networks

Probabilistic models and Bayesian networks are integral in forensic genetics, especially for DNA analysis. These models allow forensic scientists to manage uncertainties and complexities associated with DNA evidence, offering a structured framework for probabilistic reasoning. Bayesian networks are crucial in forensic evaluations, facilitating systematic assessment of evidence weight in legal contexts [1].

Advancements in probabilistic modeling for DNA analysis include the MEA and MDM methods. The MDM method significantly enhances DNA mixture interpretation by evaluating allele count probabilities and accounting for positive correlations among alleles [23]. Bayesian networks complement these models by integrating evidence sources and enabling probabilistic reasoning at the activity level. This approach allows forensic analysts to evaluate hypotheses' likelihood, considering uncertainties and dependencies in DNA data [1].

The integration of probabilistic models and Bayesian networks enhances forensic evidence accuracy and credibility. As these methodologies evolve, they are expected to play a crucial role in interpreting complex DNA evidence, leading to more accurate legal outcomes. The continuous advancement of forensic genetic models reflects the discipline's commitment to enhancing evidence accuracy and credibility, facilitating nuanced evaluations of DNA transfer and persistence. As the field expands, it demonstrates a commitment to employing cutting-edge techniques that bolster evidence reliability in judicial processes [23, 1, 21, 10].

4.5 Epigenetic Markers and DNA Methylation Analysis

Epigenetic markers, particularly DNA methylation, are emerging as valuable tools in forensic genetics, offering insights beyond traditional genetic markers. DNA methylation, involving the addition of a methyl group to cytosine bases, regulates gene expression without altering the DNA sequence, making it a valuable biomarker for forensic applications [14]. In forensic investigations, DNA methylation analysis offers an additional layer of information for estimating biological age, crucial in identifying unknown remains or assessing unknown perpetrators [14]. This technique enhances forensic scientists' ability to infer individuals' ages based on DNA samples, improving analysis accuracy.

Moreover, DNA methylation analysis addresses challenges associated with traditional genetic markers, such as distinguishing between identical twins, who share the same genetic sequence but differ in DNA methylation patterns [14]. As forensic genetics evolves, incorporating epigenetic markers like DNA methylation is expected to enhance forensic analysis precision and reliability. By providing additional genetic information layers, epigenetic markers improve forensic investigations' accuracy and depth, contributing to effective legal outcomes. Ongoing research and development of epigenetic methodologies highlight their transformative potential in forensic genetics, enabling precise age estimation and biological trace analysis. The adoption of these innovative techniques promises to enhance evidence interpretation and overall forensic analysis efficacy, including integrating personalized medicine approaches and improving the field's adaptability to cutting-edge techniques [8, 1, 21, 14].

5 Familial Searching and Genetic Genealogy

5.1 Techniques of Familial Searching and Genetic Genealogy

Familial searching and genetic genealogy are integral to forensic genetics, offering crucial insights for resolving complex cases. Familial searching involves querying DNA databases to identify potential relatives of individuals whose genetic material is present at crime scenes, utilizing Kinship Indices to evaluate genetic relationships between the target and database members. This method significantly enhances investigative capabilities, exemplified by the Golden State Killer case, where forensic DNA profiles were integrated with genealogical databases like GEDmatch [16, 27]. This approach is particularly beneficial when direct DNA matches are lacking, allowing exploration of indirect familial links that might otherwise go unnoticed.

A prominent application of familial searching is identifying male relatives via Y-chromosome DNA analysis, advantageous in sexual assault cases and paternity disputes [2]. The Y chromosome's unique inheritance patterns facilitate tracing paternal ancestry and identifying male contributors, even in the absence of direct paternal DNA.

Forensic genetic genealogy extends familial searching by merging genetic data analysis with genealogical research, leveraging direct-to-consumer (DTC) genetic services that allow users to upload genetic data for genealogical exploration. Although effective for lineage tracing and individual identification through shared genetic traits, these services raise significant privacy issues due to potential unauthorized access to sensitive genetic data [26].

Familial searching and genetic genealogy have proven particularly effective in solving cold cases and identifying missing persons. The integration of advanced forensic techniques, especially through genealogy databases like GEDmatch, has significantly enhanced law enforcement's investigative capabilities, facilitating the resolution of cold cases and identification of unknown remains. This progress prompts ethical and privacy considerations regarding personal genetic data use in criminal investigations [16, 9, 21, 13]. As these methodologies advance, they are expected to play an increas-

ingly vital role in forensic investigations, offering innovative solutions to complex legal challenges while necessitating careful ethical and privacy considerations.

5.2 Ethical and Privacy Concerns

The use of familial searching and genetic genealogy in forensic investigations raises substantial ethical and privacy concerns, particularly regarding potential genetic data misuse and implications for individual privacy rights. The use of genetic genealogy by law enforcement has sparked debates on the ethical and legal ramifications of accessing genetic data without explicit consent, especially when identifying individuals not involved in criminal activities [20]. This practice challenges traditional privacy norms, as genetic data shared via DTC genealogy services can be exploited to infer sensitive genetic information, raising concerns about re-identifying individuals from ostensibly anonymous datasets [32].

The risk to genetic privacy in DTC genealogy databases is exacerbated by potential adversaries exploiting uploaded datasets to infer genotypes, compromising the anonymity of individuals who have shared their genetic data for genealogical purposes [26]. This concern is pressing given the societal and legal implications of genetic privacy breaches, which can lead to unauthorized access to personal genetic information and potential misuse by third parties.

Moreover, familial searching poses significant misidentification risks, particularly among distant relatives, potentially leading to wrongful investigations and legal consequences for individuals erroneously linked to crime scenes [30]. Such errors highlight the need for stringent ethical guidelines and robust legal frameworks governing familial searching techniques in forensic investigations.

Public perception and societal norms significantly influence legal interpretations of privacy, affecting the acceptance and regulation of genetic genealogy practices by law enforcement [17]. As forensic genetics progresses, it is crucial to balance the benefits of genetic genealogy in crime-solving with the ethical imperative to protect individual privacy rights. Achieving this balance requires comprehensive policies and regulations addressing the ethical and privacy concerns associated with genetic data use in forensic contexts, ensuring advancements in genetic technologies are leveraged responsibly and ethically.

6 Applications and Case Studies

6.1 Applications in Law Enforcement

Forensic genetics has become integral to law enforcement by enhancing crime-solving capabilities through advanced genetic evidence analysis. Key advancements include refined DNA transfer mechanism understanding, which optimizes evidence collection and analysis, ensuring DNA evidence integrity and facilitating accurate inferences regarding genetic material's presence and origin at crime scenes [10]. Technologies like the ForenSeq Kintelligence kit further support law enforcement by generating reliable SNP profiles from low-input samples, crucial for cases with limited genetic material [12]. Such technologies are indispensable in linking suspects to crime scenes via robust genetic profiles.

Methodologies like the Multiple Population Evolutionary Algorithm (MEA) effectively resolve complex DNA mixtures, enhancing the evidential value of mixed samples, particularly when one contributor's profile is known [33]. This precision is vital in distinguishing contributors in cases involving multiple suspects. Forensic genetics also extends to nonhuman genetic evidence analysis, broadening investigative scopes and providing insights when human DNA evidence is scarce [8]. The integration of genetic genealogy techniques has revolutionized cold case resolutions, allowing law enforcement to identify individuals in long-unsolved cases by uncovering familial connections through direct-to-consumer genetic databases [15, 27].

Bayesian networks illustrate forensic genetics' impact by integrating diverse evidence types, facilitating comprehensive evaluations of genetic data in criminal identification and disputed paternity cases [24]. This systematic approach enhances forensic analysis reliability, contributing to informed judicial decision-making. Additionally, the fwsim method efficiently simulates large populations, essential for analyzing genetic data related to crimes [25]. As forensic genetics evolves, it plays a critical role in law enforcement, solving crimes, addressing legal issues, and analyzing nonhuman genetic

material in various investigations, including animal attacks and bioterrorism. The integration of innovative sequencing technologies and diverse genetic markers enhances forensic genetics' efficacy, ultimately improving public safety and governance within the justice system [8, 9]. Advanced genetic technologies will continue to bolster forensic investigators' capabilities in navigating complex legal challenges.

6.2 Case Studies and Effectiveness

Forensic genetics has demonstrated effectiveness in resolving intricate legal cases, as evidenced by numerous case studies employing advanced genetic techniques. Y-chromosome analysis has been instrumental in solving cold cases and identifying multiple male contributors in forensic investigations, particularly where traditional autosomal DNA profiling falls short [2]. The application of forensic genetic genealogy (FGG) has also shown remarkable success in solving cold cases and identifying unknown individuals. By leveraging genetic data from direct-to-consumer databases alongside genealogical research, law enforcement agencies have uncovered familial connections, leading to resolutions of long-standing unsolved cases. Maryland's regulatory framework for FGG exemplifies how robust regulation can effectively address the challenges surrounding genetic genealogy [34].

These case studies underscore forensic genetics' transformative impact on legal investigations, showcasing its ability to provide critical insights and facilitate complex case resolutions. The ongoing advancement and integration of sophisticated genetic techniques significantly enhance forensic genetics' efficacy, not only in solving criminal cases but also in preventive measures to avert judicial processes. This evolution encompasses a broader spectrum of applications, including nonhuman genetic material analysis, aiding investigations such as animal attacks, species trafficking, and biocrimes. The rise of public genealogy databases and trait prediction technologies further transforms forensic practices, necessitating careful consideration of privacy and ethical implications surrounding genetic data usage [8, 18, 1, 10, 9].

6.3 Forensic Genetic Genealogy in Cold Cases

Forensic genetic genealogy (FGG) has emerged as a transformative approach in cold case resolutions, equipping law enforcement with powerful tools to uncover familial connections and trace lineage, ultimately identifying suspects in long-unsolved cases. This method utilizes genetic data from direct-to-consumer (DTC) genetic testing services, commonly used for personal genealogical research. By cross-referencing this data with crime scene DNA, investigators can identify potential relatives of unknown suspects, facilitating the resolution of dormant cases [13].

FGG's effectiveness is particularly evident in providing investigative leads when traditional DNA profiling fails to yield direct matches, especially in cases where the suspect's DNA is absent from law enforcement databases. Since the identification of the Golden State Killer in 2018, FGG has been successfully applied to over 100 cases, leading to the arrest of at least thirty individuals for serious crimes, including murder and sexual assault. However, FGG raises significant ethical and privacy concerns regarding the use of personal genetic data without explicit consent [27, 4, 15, 20].

The increasing use of FGG necessitates thorough examination of ethical considerations and the establishment of comprehensive regulatory frameworks. Such frameworks should guide responsible FGG applications in criminal investigations, include accountability provisions for unauthorized genetic information use, and enhance transparency through mandatory public reporting [18, 7, 34, 17]. Balancing the benefits of solving cold cases with ethical obligations to protect privacy rights is essential as advancements in genetic technologies unfold.

As forensic genetic genealogy continues to evolve, its application in cold cases is expected to grow, driven by the increasing accessibility of public genetic databases, such as GEDmatch. These advancements have already facilitated breakthroughs in high-profile cases, leading to suspect identification through familial DNA matching. Law enforcement agencies must navigate new ethical, legal, and privacy considerations as they expand their use of genomic techniques [16, 7, 15]. Developing ethical guidelines and regulatory policies will be crucial in ensuring responsible genetic genealogy use, safeguarding individual rights while enhancing forensic investigations' capabilities in solving cold cases.

6.4 Case Studies of Missing Persons and Paternity Disputes

Forensic genetics has become pivotal in addressing complex cases involving missing persons and paternity disputes through genetic data analysis. The use of genetic markers, such as Short Tandem Repeats (STRs) and Single Nucleotide Polymorphisms (SNPs), is instrumental in these contexts, enabling precise identification and familial relationship determination [10, 1, 8, 14].

In missing persons cases, forensic genetics aids in identifying individuals by analyzing DNA samples from remains and comparing them with profiles from potential relatives, providing crucial leads in long-unresolved cases [7]. This capability has proven essential in solving numerous missing person cases, offering closure to families and supporting law enforcement investigations.

Paternity disputes also benefit significantly from forensic genetics, employing various genetic markers, including STRs, SNPs, and Y-chromosome short tandem repeats (Y-STRs), to establish biological parentage accurately. This advanced technique provides definitive answers in legal disputes and enhances forensic investigations by identifying paternal lineages, particularly in cases involving male contributors, such as sexual assault or missing persons. Utilizing these markers allows forensic experts to exclude male suspects, identify potential perpetrators, and trace ancestry, offering critical insights in criminal cases and familial relationships [31, 2, 15, 12]. Establishing paternity with high precision has significant implications for legal proceedings, influencing child custody cases, inheritance claims, and other familial legal matters.

Forensic genetic genealogy has emerged as a groundbreaking tool, significantly enhancing the resolution of missing persons cases and paternity disputes. By leveraging publicly available genetic data from direct-to-consumer testing platforms, law enforcement can identify individuals linked to unsolved cases, including cold cases that have persisted for decades. Since the Golden State Killer's identification in 2018, genetic genealogy has been applied to approximately 100 cases, demonstrating its effectiveness in identifying missing persons and establishing familial relationships. This method raises important ethical considerations regarding privacy and implications for relatives of DNA testing participants [16, 4, 15]. Integrating genetic data analysis with genealogical research allows forensic genetic genealogy to trace familial connections and identify individuals through shared genetic traits, proving particularly effective when traditional DNA profiling methods are inadequate.

The use of forensic genetics in these contexts underscores the field's critical role in addressing complex legal challenges, providing robust methodologies for individual identification and familial dispute resolution. As forensic science advances, the integration of sophisticated genetic technologies and methodologies is poised to significantly enhance the accuracy and reliability of forensic investigations. This evolution encompasses not only human DNA analysis but also extends to nonhuman genetic material, increasingly utilized to provide critical evidence in various legal contexts, including animal attacks and biocrimes. Incorporating personalized medicine principles and omics technologies further enhances the potential for tailored forensic approaches, leading to more effective and equitable outcomes in the justice system [8, 21].

7 Ethical, Legal, and Social Implications

The intersection of forensic genetics with legal systems raises intricate ethical, legal, and social challenges. These issues primarily revolve around privacy rights and the responsible utilization of genetic data in law enforcement. The following subsections delve into these considerations, emphasizing privacy, consent, and potential data misuse.

7.1 Legal and Ethical Considerations

Forensic genetics' role in legal investigations has sparked debates over privacy and legal frameworks, particularly concerning the use of genealogical DNA databases without explicit consent. This practice raises significant Fourth Amendment concerns, challenging the legal and ethical boundaries of genetic data usage [16]. The utilization of these databases by law enforcement, while providing valuable leads, also poses risks of privacy violations and data misuse [16]. Unauthorized access and potential adversarial exploitation of uploaded datasets further threaten the anonymity of individuals who share genetic information for genealogical purposes [26].

Genetic markers such as Y-chromosomal DNA present unique ethical dilemmas by potentially revealing sensitive familial relationships, necessitating careful consideration of their forensic application [21]. Establishing guidelines for using these markers is crucial to prevent misuse. Comprehensive policies must balance crime-solving benefits with privacy rights protection, ensuring ethical application of genetic technologies [16]. Robust ethical guidelines and regulatory frameworks are essential for the responsible evolution of forensic genetics, promoting just outcomes in legal contexts.

7.2 Legal Frameworks and Regulatory Challenges

The integration of genetic genealogy into legal investigations presents regulatory challenges, particularly with outdated frameworks like the third-party doctrine, which inadequately address privacy concerns [17]. This doctrine allows warrantless searches of shared information, risking privacy rights by permitting unauthorized genetic data access [16]. The lack of legal protections against such searches underscores the need for updated regulatory measures to safeguard privacy in forensic genetics.

Comprehensive frameworks must balance genetic genealogy's investigative benefits with privacy rights protection. A survey by [13] categorizes genetic genealogy companies' privacy policies, highlighting the need for standardized transparency and accountability policies in forensic data use. Establishing robust regulatory frameworks is crucial for responsibly harnessing forensic genetics advancements. Comprehensive legal frameworks and explicit guidelines governing genetic data use can enhance public trust in genetic technologies, ensuring ethical utilization in legal settings. Accountability mechanisms, such as damages for wrongful disclosure and transparency mandates, are vital for fostering informed public discourse and confidence at the intersection of genetics and law enforcement [18, 7, 34, 8].

7.3 Security and Vulnerabilities in Genetic Databases

Genetic database security is a critical concern in forensic genetics due to the sensitive nature of stored information. These databases are vulnerable to unauthorized access and misuse, posing risks to individual privacy and forensic investigation integrity [26]. The potential for adversaries to exploit these vulnerabilities, especially in forensic and genealogical contexts, underscores the need for robust security measures [32].

Data breaches compromise privacy and public trust in genetic technologies. The risk of sensitive genotype information being inferred from uploaded datasets further necessitates strong security measures [26]. The use of direct-to-consumer genetic databases in forensic investigations introduces additional vulnerabilities, emphasizing the need for comprehensive security protocols to prevent unauthorized access and misuse.

Developing and enforcing robust data protection policies and security frameworks is imperative to ensure genetic databases' confidentiality and integrity. Advanced encryption, access controls, and regular security audits are essential to mitigate unauthorized access and data breaches. Emphasizing database security is crucial for bolstering public trust and promoting responsible genetic data use in forensic investigations, particularly with advancements like direct-to-consumer genetic genealogy databases that raise significant privacy concerns [18, 8, 7, 4, 20].

7.4 Social Impacts and Public Trust

Forensic genetics' integration into legal investigations has significant social impacts, particularly concerning public trust and perceptions of genetic data use. These technologies can help resolve cold cases and mitigate systemic biases in criminal investigations by providing access to diverse genetic data pools. However, using genealogical databases such as GEDmatch raises urgent questions about privacy, representation, and criminal justice fairness [16, 9, 6].

Public trust in forensic genetics is vital for its development and acceptance. Misuse of genetic data and ethical and privacy concerns about accessing genealogical databases without consent can undermine confidence in forensic investigations [13]. Re-identification risks from ostensibly anonymous datasets further emphasize the need for transparency and robust data protection measures [32].

Maintaining public trust in forensic genetics is essential for the field's continued development and acceptance. Addressing the ethical, legal, and privacy issues arising from genetic information

integration in criminal investigations is crucial. This includes navigating complexities introduced by techniques like genetic genealogy, which utilize public databases to identify suspects and relatives, raising significant concerns about consent, data security, and potential misuse. As forensic genetics evolves, a coordinated policy response is necessary to safeguard public confidence in forensic and medical genetics [18, 7, 9, 8]. Transparent communication about forensic genetics' benefits and risks and robust privacy protections are essential for fostering public confidence in genetic technologies and their application in legal contexts.

Collaborative efforts among policymakers and stakeholders are crucial for developing comprehensive policies and regulatory frameworks addressing forensic genetics' social impacts and public trust issues. Prioritizing privacy rights protection and ensuring responsible and ethical genetic data use in forensic investigations are essential. By promoting public trust and addressing societal concerns about privacy and consent, forensic genetics can evolve to enhance justice system outcomes. As forensic genetics increasingly intersects with medical genetics, a comprehensive approach to ethical and regulatory challenges is vital for genetic data use in criminal investigations [18, 7, 9, 8].

8 Future Directions and Challenges

The future of forensic genetics offers significant growth opportunities due to technological advancements and a deeper understanding of genetic data within legal contexts. As law enforcement increasingly utilizes genomic techniques like forensic genetic genealogy and extensive public genetic datasets, it is vital to address both the advancements in suspect identification and ethical, legal, and privacy concerns. Additionally, the broadening scope of forensic genetics to include nonhuman genetic material, pertinent to wildlife trafficking and biocrimes, requires thorough examination of the implications for societal norms and legal standards [8, 7].

8.1 Future Directions and Research

Forensic genetics is poised for significant advancements through ongoing research and technological innovation. A key focus is developing countermeasures to prevent privacy breaches in genetic databases, such as improved algorithms for detecting shared segments to avert unauthorized access [26]. Ensuring genetic information security is crucial as the field progresses.

Enhancing data sharing practices is vital for improving genetic data accessibility and reliability. By advancing archiving tools and promoting open science, researchers can foster collaboration and transparency in forensic investigations [22], driving scientific innovation and improving forensic analyses.

Integrating omics technologies into forensic genetics presents substantial research opportunities. These methodologies offer detailed genetic profiles, enhancing forensic analyses' accuracy and depth [21]. Leveraging omics allows forensic scientists to gain deeper insights into genetic relationships, improving precision and expanding the field's capabilities.

Establishing comprehensive legal standards for genetic data use in law enforcement is another crucial research area, balancing crime-solving benefits with civil liberties protection [16]. Clear legal frameworks will guide the responsible use of genetic data in forensic contexts.

Future research should prioritize robust genetic data protection policies, addressing ethical considerations surrounding genetic information use in law enforcement [18]. Emphasizing ethical guidelines and data protection measures safeguards individual rights while utilizing genetic technologies to resolve complex legal cases.

As forensic genetics advances, exploring new methodologies and technologies will be pivotal. Integrating personalized medicine and advanced genetic analyses underscores the field's dynamic nature and transformative potential. Innovations like omics technologies enhance molecular autopsies, identify genetic predispositions to sudden death, and analyze nonhuman genetic material for broader applications in criminal justice. These advancements address complex legal challenges and promise more effective and equitable outcomes within the justice system [16, 21, 8].

8.2 Interdisciplinary Applications and Expanding Capabilities

Forensic genetics' interdisciplinary applications are transforming scientific and legal investigations, offering innovative solutions across various fields. Integrating forensic genetics with personalized medicine can revolutionize both domains by providing tailored approaches to individual identification and health assessments [21]. This integration enhances forensic analyses through detailed genetic profiles for more accurate investigations.

The field is also expanding its capabilities through advanced statistical and computational methodologies, such as probabilistic models and Bayesian networks, facilitating complex DNA evidence interpretation [1]. These tools enhance genetic analyses' reliability, contributing to more informed judicial outcomes.

Forensic genetics' application in population genetics and evolutionary biology provides insights into genetic structures, informing forensic investigations and broader scientific research [25]. This capability enhances our understanding of human history and evolution, offering perspectives on genetic diversity and inheritance patterns.

Developing methodologies for analyzing nonhuman DNA illustrates forensic genetics' expanding capabilities, providing critical insights in environmental forensics and wildlife conservation [8]. These applications demonstrate the field's versatility in addressing a wide range of scientific and legal challenges beyond traditional human identification.

As the field evolves, integrating cutting-edge technologies and interdisciplinary approaches will be vital in enhancing forensic genetics' capabilities. Ongoing research in personalized medicine and molecular autopsy emphasizes these innovations' transformative potential for forensic investigations. Employing advanced techniques such as genomics, transcriptomics, and metabolomics allows forensic science to tackle complex legal challenges more effectively. These advancements facilitate identifying genetic predispositions to fatal conditions, improve death determinations, and raise critical ethical and legal questions regarding genealogy database usage in criminal investigations. The evolution of forensic methodologies promises to enhance the justice system's ability to deliver fair outcomes while safeguarding individual privacy rights [16, 21].

8.3 Standardization and Training

Standardization and training in forensic genetics are essential for ensuring analyses' accuracy, reliability, and consistency. Establishing uniform procedures and protocols for collecting, analyzing, and interpreting genetic data is crucial for maintaining forensic investigation integrity and facilitating results comparison across laboratories and jurisdictions. Standardized practices reduce errors and biases during analyses, enhancing genetic evidence reliability in court. This approach supports thorough evaluations within specific cases and improves interpretative frameworks through advanced probabilistic models like Bayesian networks. Adhering to rigorous training and quality assurance protocols allows forensic scientists to provide clearer, more robust assessments of genetic evidence, bolstering judicial processes with transparent and well-founded inferences [8, 7, 1, 18].

Training is vital for forensic practitioners, enhancing their ability to utilize genetic technologies and methodologies effectively in investigations and preparing them to analyze a broader range of genetic materials, including nonhuman DNA. This relevance is increasingly important in cases like wildlife trafficking, bioterrorism, and food fraud. Comprehensive training ensures practitioners can accurately interpret DNA transfer dynamics and provide informed opinions on evidence, improving overall forensic investigation efficacy [8, 10]. Training programs should encompass both theoretical and practical aspects of forensic genetics, covering DNA extraction, profiling techniques, data interpretation, and the ethical and legal considerations associated with genetic analyses. Ongoing education and professional development opportunities are essential for keeping practitioners abreast of the latest advancements and best practices.

The integration of advanced technologies into forensic genetics underscores the need for specialized training. As the field advances, practitioners must develop proficiency in state-of-the-art tools and methodologies, including probabilistic models, Bayesian networks, and omics technologies. These techniques are crucial for enhancing analyses' accuracy and reliability, particularly in evaluating forensic genetic findings based on activity level propositions. As analytical sensitivity increases, forensic biologists must adeptly interpret data and communicate findings to support judicial processes,

ensuring robust and transparent inferences regarding trace evidence transfer. Specialized training and adherence to international reporting guidelines are vital for effective legal decision-making [19, 23, 11, 24, 1]. Training programs should therefore be designed to address these emerging technologies, preparing forensic scientists to meet modern investigative challenges.

Moreover, standardization and training are critical for fostering public trust in forensic genetics. Implementing standardized protocols and ensuring specialized training in areas like DNA transfer and activity level propositions underscore the field's commitment to scientific rigor and ethical responsibility, enhancing forensic evidence reliability in legal proceedings. This approach enables forensic scientists to provide well-informed opinions based on robust data interpretation methods, such as probabilistic genotyping and Bayesian networks, essential for accurately assessing the context and implications of their findings in court. Continuous training and competency testing are vital for maintaining high standards in evidence evaluation, ultimately reinforcing forensic evidence credibility in the judicial system [10, 1, 18]. As forensic genetics expands its capabilities and applications, ongoing development of standardized practices and comprehensive training programs will be essential for maintaining the field's integrity and advancing its contributions to justice and public safety.

9 Conclusion

The evolution of forensic genetics has marked a significant leap forward in the realm of legal investigations, diversifying from traditional DNA profiling to encompass an array of sophisticated methodologies. This includes the utilization of genetic markers such as Single Nucleotide Polymorphisms (SNPs) and Short Tandem Repeats (STRs), which are indispensable for accurate individual identification and resolving complex forensic cases. The incorporation of nonhuman DNA analysis and advanced techniques like shotgun DNA sequencing has further bolstered the precision and dependability of forensic practices.

Kinship analysis remains a cornerstone of forensic genetics, employing cutting-edge techniques such as Y-STR haplotyping to elucidate familial connections and trace lineages. Despite its critical role, this area faces challenges, including computational limitations and ethical issues surrounding genetic data use. These challenges necessitate ongoing research and the refinement of methodologies, alongside enhanced training for forensic experts.

The advent of genetic genealogy and familial searching has revolutionized forensic investigations, offering innovative solutions for cold cases and missing persons through the scrutiny of consumer genetic databases. These approaches have demonstrated remarkable efficacy in intricate legal scenarios, underscoring forensic genetics' vital contribution to modern law enforcement. Nonetheless, the forensic application of genetic genealogy presents substantial ethical and privacy concerns, particularly regarding genetic data misuse and erosion of public trust.

As forensic genetics continues to advance, it stands at a critical juncture where technological and methodological innovations are reshaping its landscape and expanding its capabilities. The integration of omics technologies, probabilistic models, and Bayesian networks has enhanced the accuracy and reliability of forensic analyses, facilitating the resolution of complex legal challenges. However, these developments highlight the necessity for comprehensive legal frameworks and ethical standards to govern the use of genetic data, safeguarding individual privacy rights.

Addressing the existing gaps in current forensic methodologies is imperative, along with the development of more effective genetic markers to propel the discipline forward. The establishment of international guidelines is also crucial, ensuring the responsible utilization of genetic genealogy databases in law enforcement, balancing the benefits of forensic genetics with the need to protect public safety and individual privacy.

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