
Kinship Analysis and Forensic Genealogy: A Survey

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Abstract

The survey paper explores the interconnected techniques of kinship analysis, forensic genealogy, STR analysis, genetic genealogy, familial searching, and DNA profiling, which are integral to modern forensic science and genealogy. These methodologies collectively enhance the ability to establish familial relationships, identify individuals, and solve crimes by analyzing genetic markers. The paper is structured to provide a comprehensive examination of these techniques, beginning with an introduction to their significance in forensic applications. It delves into the historical development and evolution of these methodologies, highlighting advancements such as nanopore sequencing and the integration of STR and SNP genotyping. The survey also addresses the ethical and legal considerations associated with the use of genetic data in forensic contexts, emphasizing privacy concerns, regulatory frameworks, and the necessity of informed consent. By integrating genealogical research with forensic DNA analysis, the paper underscores the transformative impact of these techniques in resolving cold cases and enhancing public safety. The survey concludes by reflecting on future directions, recommending enhancements in sequencing protocols and ethical frameworks to ensure the continued advancement of kinship analysis and forensic genealogy while maintaining ethical standards and societal values.

1 Introduction

1.1 Importance of Kinship Analysis and Forensic Genealogy

Kinship analysis and forensic genealogy are crucial methodologies in contemporary forensic science, significantly aiding in crime resolution and individual identification. These techniques extend their utility to complex scenarios, such as victim identification in armed conflicts, by analyzing genetic markers to establish familial relationships when traditional identification methods are inadequate [1, 2].

The incorporation of genetic genealogy into law enforcement has markedly improved the resolution of cold cases, exemplified by high-profile investigations like the Golden State Killer case, which utilized public genetic databases to identify potential relatives of unidentified individuals, thereby solving long-standing cases [3, 4]. However, the application of direct-to-consumer genetic data in forensic contexts raises significant ethical, legal, and privacy concerns, warranting careful consideration of policy implications [5].

Moreover, Y-chromosome DNA profiling has proven essential in cases where autosomal DNA profiling is insufficient, such as in sexual assault cases and paternity disputes [6]. This underscores the vital role of kinship analysis in elucidating genealogical relationships defined by birth and parentage [7].

While advancements in forensic genetic genealogy (FGG) demonstrate its efficacy in resolving cold cases, they also introduce challenges regarding implementation and ethical considerations [8]. Balancing the benefits of genetic genealogy for law enforcement with the associated legal and ethical concerns remains a critical area for ongoing research and discourse [9].

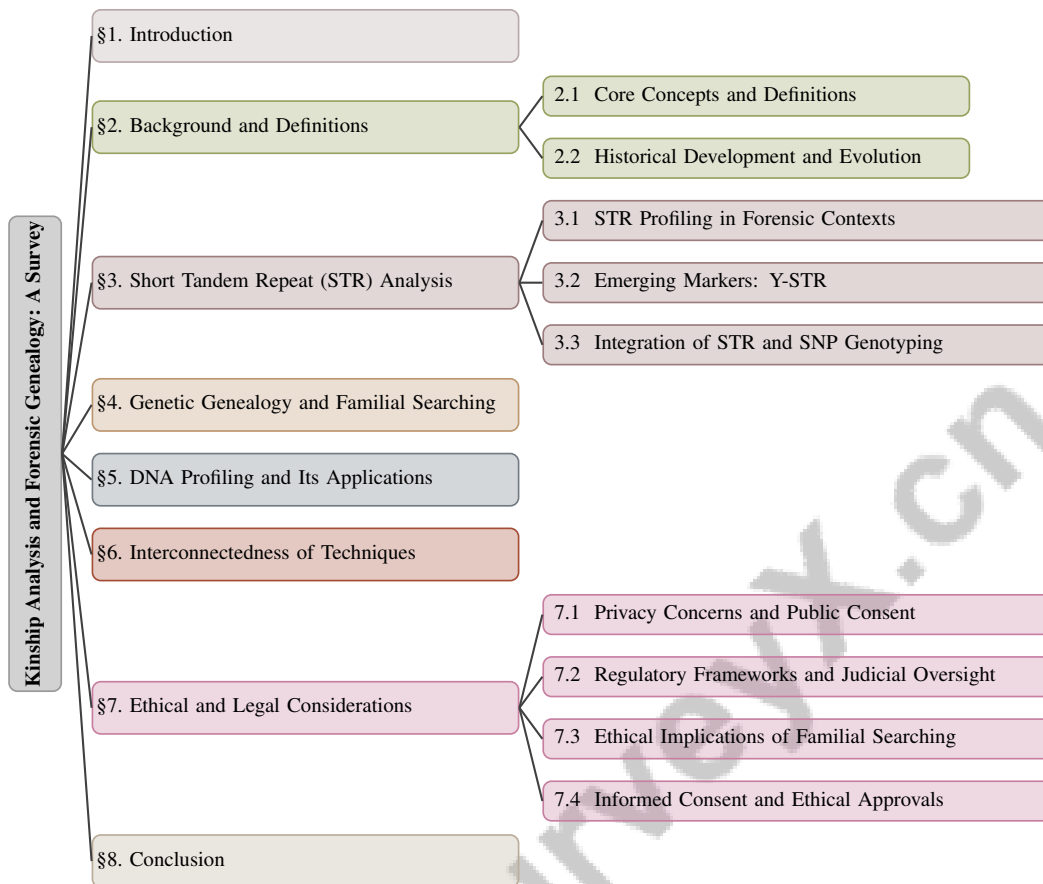


Figure 1: chapter structure

1.2 Structure of the Survey

This survey is meticulously organized to provide a thorough exploration of kinship analysis and forensic genealogy, elucidating their significance in forensic science and genealogy. The introductory section establishes the foundational importance of these methodologies in modern applications. Following this, Section 2 offers background information and definitions, detailing core concepts and tracing the historical evolution of these techniques.

Section 3 focuses on Short Tandem Repeat (STR) analysis, a cornerstone technique in forensic identification. It examines methodologies and applications while emphasizing emerging markers like Y-STR, DIP-STR, and SNP-STR, which are particularly beneficial for analyzing low-quantity DNA samples and interpreting mixed DNA profiles where traditional methods may falter. This section discusses how Y-STR and DIP-STR markers help detect minor contributors in imbalanced DNA mixtures, while SNP-STR markers enhance genetic profiling accuracy, providing superior discrimination power and exclusion rates compared to standard STRs [10, 11]. The integration of STR and SNP genotyping in forensic contexts is also addressed, showcasing advancements in the field.

Section 4 delves into genetic genealogy and familial searching, highlighting the role of public genetic databases in forensic investigations while addressing ethical and privacy considerations inherent in these practices and their effectiveness in resolving cold cases.

In Section 5, the survey analyzes various DNA profiling techniques, comparing their effectiveness with other genetic markers and exploring their diverse applications in forensic contexts, particularly in establishing identity and kinship relationships. It also highlights recent advancements such as genetic genealogy and familial searching, which have been pivotal in solving cold cases while raising significant ethical issues [12, 13]. Section 6 discusses the interconnectedness of these techniques,

focusing on the integration of genealogical research with forensic DNA analysis and the incorporation of emerging technologies.

Ethical and legal considerations are thoroughly examined in Section 7, which addresses privacy concerns, regulatory frameworks, and the ethical implications of familial searching. This section emphasizes the necessity of informed consent and ethical approvals in the application of genetic genealogy techniques. The survey concludes with Section 8, summarizing key points discussed and reflecting on future directions and potential advancements in the field, particularly regarding ethical considerations. The following sections are organized as shown in Figure 1.

2 Background and Definitions

2.1 Core Concepts and Definitions

Kinship analysis and forensic genealogy rely on pivotal methodologies such as Short Tandem Repeat (STR) profiling, a fundamental technique in forensic DNA genotyping that amplifies specific DNA regions using polymerase chain reaction (PCR) [14]. This method is crucial for both forensic investigations and the authentication of biological samples, like mouse cell lines [15]. However, STR profiling struggles with distinguishing closely related individuals, particularly in single-cell analyses post-whole genome amplification (WGA) [2], and often fails to resolve profiles in unbalanced DNA mixtures [10].

Conversely, Single Nucleotide Polymorphism (SNP) genotyping provides enhanced resolution for examining genetic variations, vital for forensic genetic genealogy (FGG) and kinship dynamics [16]. SNP profiles facilitate detailed genetic relationship construction, crucial for lineage tracing and familial connections. The Y chromosome, with its unique inheritance pattern, is an essential tool for tracing patrilineal inheritance and understanding paternal lineages [17].

Genetic genealogy employs genetic data from direct-to-consumer tests in genealogy databases to trace familial relationships and identify individuals [13]. Utilizing centimorgan similarity scores and concepts like 'identical by descent', it enhances complex case resolution [18]. Familial searching, leveraging genetic databases to find relatives of unidentified individuals, supports criminal investigations and victim identification, especially in challenging scenarios like armed conflicts [1].

The synergy of genealogical research with forensic DNA analysis is vital for resolving cold cases and identifying unknown victims [8]. The concept of 'relevant population' in Bayesian forensic inference is crucial for interpreting forensic evidence in legal contexts [19]. The complexities of genealogy require an analytical framework that acknowledges genealogical categorizations and identity politics [7].

Understanding DNA transfer mechanisms is essential for accurate forensic evidence interpretation, as these factors significantly influence the reliability of genetic analyses in criminal cases [20]. The integration of these genetic tools underscores the interdisciplinary nature of kinship analysis and forensic genealogy, necessitating a comprehensive grasp of both technical aspects and broader implications in forensic science.

2.2 Historical Development and Evolution

The evolution of kinship analysis and forensic genealogy is closely tied to genetic research advancements and the demands of forensic science. Initially, challenges arose in interpreting unbalanced mixed DNA samples, which conventional methods struggled to address [10]. Technological innovations have driven a shift towards more sophisticated approaches, adapting to complex forensic scenarios.

A pivotal development in forensic genetics is nanopore sequencing, which revolutionized genetic data collection and analysis by offering a portable, cost-effective solution for on-site forensic analysis [14]. This marks a significant transition from traditional laboratory techniques. The historical context of forensic Y-chromosome research, particularly legislative changes in the Netherlands, highlights the increased use of Y-chromosome DNA in forensic investigations [6].

The Y chromosome has been key in genetic genealogy, especially in understanding patrilineal inheritance across domains like family history and anthropology [17]. Despite advancements, the

focus on female lines in kinship analysis has overlooked male kin, representing a gap in the historical narrative [21].

Socio-political factors, particularly related to armed conflicts, have further shaped kinship analysis and forensic genealogy techniques, driving the development of advanced methods for victim identification and familial searching [1]. The complexity of DNA transfer mechanisms, categorized into direct and indirect, underscores the intricacies of forensic investigations [20].

Forensic genetic genealogy (FGG) has emerged as a transformative practice in law enforcement, involving stages such as DNA sample collection, SNP analysis, genealogical matching, and legal investigation [8]. This evolution reflects increased collaboration among forensic specialists and genealogists. However, using public genetic datasets and genealogy records for law enforcement poses ethical and legal challenges, necessitating careful examination of the implications [5].

The historical development of kinship analysis and forensic genealogy also includes advancements in understanding crossover events and their impact on DNA inheritance [18]. This theoretical perspective situates genealogical relatedness within broader social and political contexts, examining its implications for collective identity [7]. Despite challenges, the field continues to advance, paving the way for more comprehensive and precise forensic investigations.

In recent years, the application of Short Tandem Repeat (STR) analysis has become increasingly significant in forensic genetics, leading to a deeper understanding of its hierarchical structure. As illustrated in Figure 2, this figure delineates the various aspects of STR analysis, emphasizing key categories such as profiling applications, emerging markers, and the integration of STR and SNP genotyping. Each of these categories is meticulously divided into subcategories that provide insights into specific advancements, challenges, and future directions within the field. This structured representation not only enhances our comprehension of the current landscape of forensic genetics but also underscores the importance of continued research and innovation in this area.

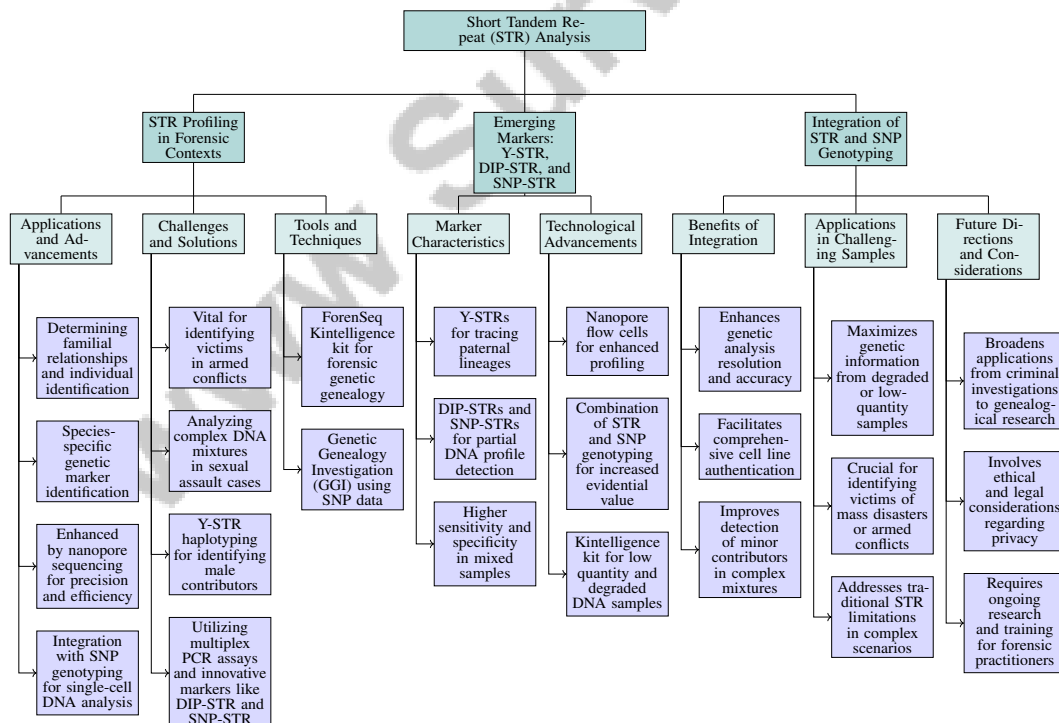


Figure 2: This figure illustrates the hierarchical structure of Short Tandem Repeat (STR) analysis in forensic contexts, highlighting key categories such as profiling applications, emerging markers, and the integration of STR and SNP genotyping. Each category is further divided into subcategories that detail specific advancements, challenges, and future directions in forensic genetics.

3 Short Tandem Repeat (STR) Analysis

3.1 STR Profiling in Forensic Contexts

STR profiling is pivotal in forensic science for determining familial relationships and individual identification. Utilizing PCR to amplify DNA regions with repeating sequences, this technique is validated for diverse applications, including species-specific genetic marker identification [15]. Its strength lies in generating unique genetic profiles crucial for resolving complex forensic cases. Advances in sequencing, notably nanopore sequencing, have enhanced STR profiling's precision and efficiency, expediting forensic investigations [14]. The integration of length-based STR and sequence-based SNP genotyping has improved single-cell DNA analysis post-WGA, aiding in distinguishing closely related individuals [2].

STR profiling is vital where traditional methods are insufficient, such as identifying victims in armed conflicts [1]. It excels in analyzing complex DNA mixtures, crucial in sexual assault cases for differentiating multiple contributors [6]. Y-STR haplotyping offers superior discriminatory power for identifying male contributors, surpassing autosomal STR profiling's effectiveness. Innovative methodologies like multiplex PCR assays have expanded STR capabilities, enabling simultaneous detection of multiple SNP-STRs and the Amelogenin marker, enhancing DNA mixture analysis [11]. DIP-STR and SNP-STR markers further improve detection and differentiation of partial DNA contributions, enhancing forensic accuracy [10].

Tools like the ForenSeq Kintelligence kit, targeting 10,230 SNPs for forensic genetic genealogy, exemplify integrating SNP data to enhance genetic profile resolution and familial assessments [16]. Genetic Genealogy Investigation (GGI) utilizing anonymized SNP data in databases like GEDmatch underscores STR profiling's utility in solving cold cases and identifying unknown individuals [13].

3.2 Emerging Markers: Y-STR, DIP-STR, and SNP-STR

Emerging markers like Y-STR, DIP-STR, and SNP-STR are enhancing forensic genetics. Y-STRs are effective for tracing paternal lineages and clarifying contributions from multiple male donors in mixed samples, particularly in sexual assault and paternity disputes [10, 6, 17]. DIP-STRs and SNP-STRs surpass traditional STR limitations, improving detection and interpretation of partial DNA profiles in mixed samples [10]. These markers exhibit higher sensitivity and specificity, identifying minor contributors in imbalanced mixtures, a frequent forensic challenge [11].

Advancements in sequencing technologies, such as nanopore flow cells for genotyping forensic STRs, have enhanced profiling accuracy and efficiency [14]. The combination of STR and SNP genotyping significantly increases the evidential value and accuracy of kinship analysis, particularly in single-cell contexts [2]. Practical applications like the Kintelligence kit utilize targeted PCR to amplify low quantity and degraded DNA samples, enhancing kinship analysis reliability [16]. These advancements underscore the transformative potential of emerging technologies in forensic genetics, paving the way for more precise analyses.

3.3 Integration of STR and SNP Genotyping

The integration of STR and SNP genotyping marks a significant advancement in forensic investigations, enhancing genetic analysis resolution and accuracy. This approach combines STR profiling's high discriminatory power with SNP genotyping's detailed genetic information. The multiplex PCR assay exemplifies this integration, facilitating comprehensive cell line authentication and forensic identification [15].

STR profiling generates unique genetic profiles through repetitive DNA sequence amplification, increasingly enhanced by SNP genotyping. This combination allows precise analysis of genetic variations, crucial in forensic contexts where mixed DNA samples can obscure results. SNP-STR markers excel in detecting minor contributors in complex DNA mixtures, achieving high resolution even at low ratios, thus improving forensic identifications compared to traditional STR methods [14, 10, 11, 15]. SNPs provide critical insights into lineage and ancestry, enabling detailed genetic relationship construction essential for familial connection tracing and individual identification.

This integration is valuable for challenging samples, such as degraded or low-quantity ones, maximizing genetic information extraction from compromised samples and enhancing forensic analysis

robustness. It is crucial in identifying victims of mass disasters or armed conflicts, where biological sample degradation hinders traditional techniques [1, 19]. The integration addresses traditional STR limitations, especially in complex scenarios involving unbalanced DNA mixtures or closely related individuals. This innovative approach enhances minor contributor detection in mixed samples, significantly improving forensic genetic profiling accuracy and reliability. Studies show SNP-STR markers provide superior discrimination and exclusion power compared to standard STRs, invaluable in challenging forensic analyses [10, 11].

The synergy between STR and SNP genotyping not only enhances forensic analysis precision but also broadens potential applications, from criminal investigations to genealogical research. As forensic science progresses, integrating innovative techniques like DNA transfer analysis, forensic genetic genealogy, and genealogy databases will be crucial for improving genetic relationship assessments and individual identifications. These advancements promise deeper insights into DNA presence at crime scenes while raising ethical and legal considerations regarding privacy and data protection, necessitating ongoing research and training for forensic practitioners to navigate these complexities effectively [20, 12, 3].

4 Genetic Genealogy and Familial Searching

4.1 Integration of Public Genetic Databases in Forensic Investigations

The integration of public genetic databases into forensic investigations has revolutionized crime resolution and identification of potential relatives. Leveraging extensive data from direct-to-consumer genetic tests, familial searching queries DNA databases to identify relatives of specific individuals, proving more effective than traditional methods [22]. The ability to correlate multiple DNA matches is crucial for accurately identifying potential relatives, enhancing the resolution of complex cases [18].

These databases, rich in DNA profiles from diverse individuals, enable cross-referencing to detect genetic similarities and infer relationships. Incorporating prior information and forensic evidence refines suspect populations, significantly improving identification accuracy [19]. This approach is particularly advantageous in scenarios where traditional methods are inadequate, such as mass disaster victim identification or solving cold cases.

The creation and management of datasets, including those using DNA from common cell lines, provide a foundation for developing and validating forensic methodologies, ensuring the reliability of genetic analyses [15]. As forensic genetics evolves, the role of public genetic databases remains pivotal, offering new pathways for crime-solving and individual identification through familial connections.

4.2 Ethical and Privacy Considerations

The use of genetic genealogy and familial searching in forensic investigations raises significant ethical and privacy concerns. The potential misuse of genetic data and legal complexities surrounding DNA evidence remain critical issues [5]. The proliferation of direct-to-consumer genetic testing exacerbates privacy concerns, particularly regarding services that can identify individuals from anonymous data [23]. This has led to increased scrutiny of privacy policies of genetic genealogy companies, which often allow law enforcement access to user data, raising questions about consent and data protection [24].

While familial searching effectively identifies potential suspects, it poses ethical dilemmas in balancing public safety with privacy rights [12]. The risk of false conclusions from chance database matches complicates the identification of true relatives, potentially implicating innocent individuals [22]. Furthermore, the use of genetic genealogy databases by law enforcement without data contributors' consent raises significant ethical and legal concerns.

The disproportionate representation of racial minorities in DNA databases complicates the ethical landscape, necessitating a nuanced understanding of the power dynamics and social relations that genealogy can reveal [25, 7]. Ethical considerations must address potential discrimination and advocate for equitable forensic practices.

4.3 Applications and Effectiveness in Solving Cold Cases

Genetic genealogy has become a crucial tool in forensic investigations, particularly for solving cold cases. By utilizing extensive genetic databases to identify potential relatives of unknown individuals, law enforcement gains innovative avenues for identifying previously unknown suspects [9]. The success of genetic genealogy in resolving cold cases is evident in high-profile investigations where traditional methods had failed, offering closure to families and enhancing public safety [8].

Empirical studies highlight the effectiveness of genetic genealogy in forensic contexts. For instance, research in the UK identified four out of ten volunteers using genetic genealogy techniques, demonstrating the method's potential for broader forensic use [13]. This success is attributed to the integration of SNP panels, which enhance evidential value in kinship analysis and expand forensic applications [2].

Y chromosome analysis plays a vital role in genetic genealogy, particularly for genealogical inquiries and historical remains identification, demonstrating its effectiveness in solving complex cases involving paternal lineage tracing [17]. Advanced assays, such as those capable of detecting SNP-STR haplotypes from minor DNA contributors, exemplify the superior forensic efficiency of these markers over standard autosomal STRs [11].

Familial searching protocols enhance genetic genealogy's effectiveness by accurately identifying first-degree relatives while minimizing misidentification risks, ensuring the reliability of forensic investigations [25]. The integration of these techniques has been instrumental in resolving cold cases, providing closure for families of missing persons [4].

The application of genetic genealogy in solving cold cases marks a significant advancement in forensic science, equipping law enforcement with powerful tools to tackle unsolved cases and improve public safety. Ongoing research aims to refine methodologies, revealing the potential for these techniques to revolutionize forensic investigations, particularly in addressing long-standing cold cases. Recent applications of direct-to-consumer genetic databases have led to breakthroughs in identifying suspects and missing persons, highlighting the effectiveness of tracing familial connections. However, these advancements also raise critical ethical and privacy concerns, necessitating collaborative efforts among forensic scientists, bioethicists, law enforcement, and genetic genealogists to establish responsible guidelines for their use [3, 4, 13].

5 DNA Profiling and Its Applications

5.1 Overview of DNA Profiling Techniques

DNA profiling encompasses several critical techniques for individual identification and kinship analysis in forensic science. Short Tandem Repeat (STR) analysis remains a cornerstone method, leveraging the amplification of DNA regions with repeating sequences to generate distinct genetic profiles. Despite its widespread use, STR analysis faces challenges with complex repeat patterns that can impact genotyping accuracy, as evidenced by nanopore sequencing experiments [14].

Emerging as a potent complement to STR analysis, Single Nucleotide Polymorphism (SNP) genotyping offers enhanced resolution of genetic variations, particularly beneficial in forensic genetic genealogy for tracing familial connections. The Kintelligence kit exemplifies this approach's efficacy, validated for high-quality SNP profiles from low DNA input samples, underscoring its forensic relevance [16].

Advancements in sequencing technologies, notably nanopore sequencing, have augmented DNA profiling by facilitating high-throughput STR analysis with improved accuracy, particularly in loci with homopolymers. These innovations lower sequencing costs and broaden forensic applications, enabling more detailed genetic analyses in criminal investigations [12, 20, 14, 5, 26]. However, achieving uniform genotyping accuracy across all loci remains challenging, necessitating ongoing methodological refinement.

The interplay between established techniques like STR analysis and novel methodologies such as SNP genotyping and nanopore sequencing is reshaping DNA profiling in forensic science. While STRs are effective for individual identification, SNP genotyping excels in analyzing degraded or low-quantity DNA samples, especially in complex mixtures. Furthermore, advancements in nanopore sequencing,

with new flow cell designs, promise to enhance STR genotyping accuracy, though challenges with homopolymers persist in routine forensic workflows. As forensic genomics evolves, integrating these approaches can enhance investigative capabilities, utilizing public genetic datasets for forensic genetic genealogy, while also raising ethical and legal considerations [20, 14, 10, 5]. This integration not only improves genetic analysis resolution and accuracy but also expands forensic applications from criminal investigations to genealogical research.

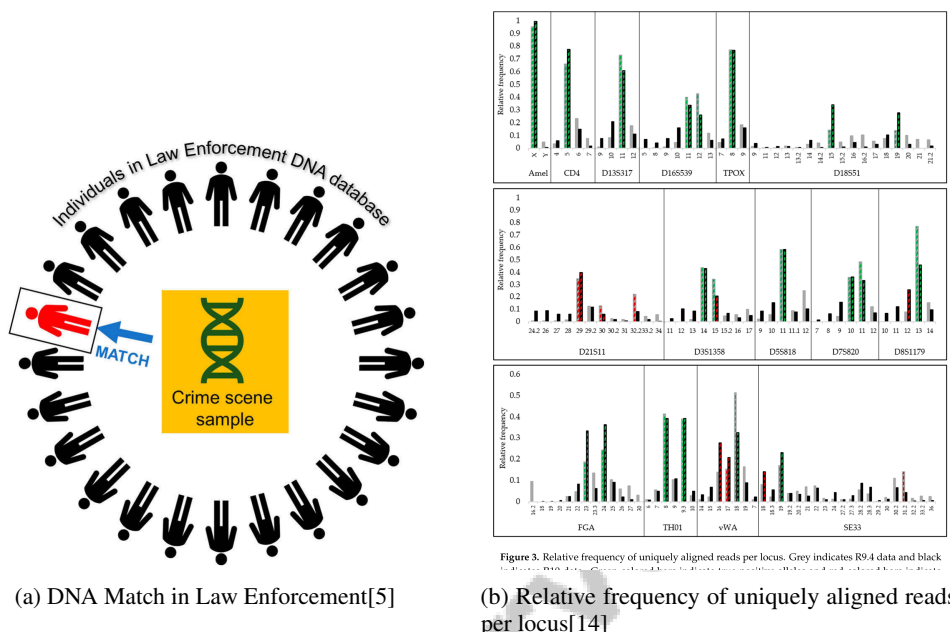


Figure 3: Examples of Overview of DNA Profiling Techniques

As illustrated in Figure 3, DNA profiling has transformed law enforcement and genetic research by providing precise identification techniques based on genetic markers. The first image, "DNA Match in Law Enforcement," depicts the process of matching a crime scene DNA sample with an individual from a law enforcement database, highlighting the critical role of DNA profiling in accurately identifying suspects through genetic evidence. The second image, "Relative frequency of uniquely aligned reads per locus," presents a bar chart that showcases the frequency of uniquely aligned DNA reads across various genetic loci, differentiating data from different genetic markers, including the X and Y chromosomes. Together, these examples underscore the diverse applications of DNA profiling techniques and their significance in forensic investigations and genetic research [5, 14].

5.2 Comparative Analysis with Other Genetic Markers

DNA profiling, notably through STR analysis, has been instrumental in forensic investigations for generating unique genetic profiles. The introduction of SNP genotyping enhances forensic analyses by providing higher resolution insights into genetic variations [16]. Whereas STR profiling focuses on repeating DNA sequences, SNP genotyping examines single base pair changes, offering a detailed genetic landscape crucial for tracing familial connections and constructing complex genealogical relationships [14].

SNP integration into forensic investigations is particularly valuable in scenarios where traditional STR markers may falter, such as with degraded or low-quality DNA samples. SNPs are less prone to degradation and can be analyzed from smaller DNA quantities, making them suitable for challenging forensic contexts [2]. Additionally, SNP panels provide ancestry and lineage insights unavailable through STRs alone, broadening forensic applications to include genealogical and anthropological inquiries.

Y-chromosome STRs (Y-STRs) and mitochondrial DNA (mtDNA) are additional genetic markers crucial in forensic investigations. Y-STRs, inherited through the male line, are useful for tracing

paternal lineages [17], particularly in cases involving male relatives or where autosomal DNA is insufficient [27]. Conversely, mtDNA, maternally inherited, offers insights into maternal lineages and is beneficial when nuclear DNA is unavailable or too degraded.

Despite the unique advantages of each genetic marker, their integration into a comprehensive forensic strategy enhances overall effectiveness. Combining STR, SNP, Y-STR, and mtDNA analyses facilitates a multifaceted genetic profiling approach, accommodating diverse forensic scenarios and improving identification accuracy and reliability. As forensic science advances, integrating genetic markers informed by DNA transfer dynamics and forensic genetic genealogy will be crucial in addressing contemporary forensic challenges. This evolution will enhance DNA evidence interpretation accuracy, improve suspect identification through familial searching techniques, and navigate ethical and legal complexities arising from extensive genomic datasets. Continued research and robust guidelines will be essential to maximize these tools' potential while safeguarding privacy and ensuring responsible law enforcement use [20, 12, 5, 8].

6 Interconnectedness of Techniques

6.1 Integration of Genealogical Research with Forensic DNA Analysis

The fusion of genealogical research with forensic DNA analysis represents a significant advancement in forensic science, enhancing both individual identification and the resolution of complex cases. This integration is significantly supported by advanced sequencing technologies, such as nanopore sequencing, which improve genetic profiling accuracy and efficiency [14]. Legal frameworks and familial involvement are critical, with Manera et al. underscoring the necessity of legal backing and ethical guidelines for the responsible use of genealogical data in forensic settings [1]. Formici et al. further explore these ethical dimensions, proposing a framework for evaluating the effectiveness and ethical considerations of DNA analysis in criminal investigations [12].

Tools like the Kintelligence kit, discussed by Peck et al., provide reliable SNP profiles from low input and degraded samples, crucial for challenging forensic cases [16]. Familial DNA searching methods, as evaluated by Slooten et al., systematically assess relatedness, reducing the likelihood of false matches [22]. The MVN-FG method, integrated with traditional genealogical research, enhances familial relationship assessments, offering a robust forensic framework [18].

Caswell et al.'s two-sex kinship model explores gender differences in kinship dynamics, highlighting the complex interplay between genealogical research and forensic DNA analysis [21]. Brümmer et al. stress the importance of consensus on population assumptions to ensure reliable analyses [19]. Rohlf et al. introduce a simulation-based approach to evaluate familial search processes, focusing on misidentification risks across diverse populations [25].

6.2 Integration with Emerging Technologies

The integration of emerging technologies into forensic genealogy and kinship analysis has significantly enhanced forensic capabilities, enabling more precise and efficient analyses. Advances in sequencing technologies, particularly nanopore sequencing, have transformed genetic data collection, providing portable and cost-effective solutions for on-site forensic analysis, thereby facilitating rapid and accurate genetic information processing [10].

Emerging markers such as Y-STR, DIP-STR, and SNP-STR are crucial in enhancing sensitivity and specificity for low DNA quantities and complex mixtures, addressing traditional STR analysis limitations [10]. These markers are particularly valuable in scenarios involving degraded or low-quality DNA samples.

The integration of advanced analytical frameworks and privacy policies in genetic genealogy companies highlights the ethical considerations of emerging technologies. The categorization of privacy policies based on law enforcement access underscores the need for robust regulatory measures to protect privacy while ensuring effective forensic practices [24]. This framework calls for transparent regulations to safeguard individual rights.

The integration of technologies like forensic genetic genealogy and advanced kinship analysis marks a substantial evolution in forensic science, providing law enforcement with innovative tools to tackle complex cases. This advancement enhances forensic investigation accuracy while raising critical

ethical and legal considerations regarding privacy and genealogical database use, as demonstrated by high-profile cases like the Golden State Killer’s apprehension through public genealogical data [3, 12]. As these technologies advance, their forensic applications will play a crucial role in enhancing capabilities while ensuring ethical genetic information use.

7 Ethical and Legal Considerations

7.1 Privacy Concerns and Public Consent

The integration of genetic data in forensic investigations intensifies privacy concerns and underscores the necessity for public consent, central to the ethical discourse in forensic genetic genealogy (FGG) and familial DNA searching. A significant challenge is the lack of informed consent from individuals whose genetic data is used, raising ethical and privacy issues [4]. Relatives of consented individuals are vulnerable to privacy violations, as their genetic information can be indirectly accessed through familial links [3]. The inclusion of minors in genetic databases adds complexity regarding consent and data protection [4].

The labor-intensive nature of FGG investigations and potentially incomplete genealogical data further complicate the ethical landscape, necessitating robust guidelines to address privacy and consent issues [8]. The absence of legal protections for those whose genetic data is in non-forensic databases exacerbates these challenges, risking privacy rights violations [3]. Establishing clear legal frameworks is essential for safeguarding individual privacy while effectively utilizing genetic data in forensic contexts.

Brümmer et al. highlight the importance of considering relevant population data for interpreting genetic evidence, crucial for ensuring forensic investigations’ accuracy and fairness, especially with diverse population samples [19]. Rohlf et al. provide guidance for law enforcement practices and policy decisions regarding familial searching, advocating for informed consent and ethical considerations [25].

Institutional bioethics committees play a vital role in addressing privacy concerns and integrating ethical considerations into genetic research and forensic investigations [27]. Their approval underscores the importance of ethical oversight in genetic data collection and use. As forensic genetics evolves, prioritizing informed public consent and establishing transparent guidelines are crucial for maintaining public trust and realizing genetic data integration benefits without compromising ethical standards.

7.2 Regulatory Frameworks and Judicial Oversight

The use of genetic information in forensic genealogy necessitates robust regulatory frameworks and judicial oversight to ensure ethical compliance and protect individual rights. The rapid advancement of forensic genetic genealogy (FGG) highlights the urgent need for comprehensive regulations governing DNA database use, addressing ethical implications of genetic surveillance, and maintaining public trust in forensic practices [12].

A critical aspect of these frameworks is implementing measures to prevent genetic data misuse. Recommendations like rate limiting and anomaly detection for API queries are essential to mitigate attacks on genetic databases, ensuring security and integrity of sensitive information [23]. These safeguards are vital to prevent unauthorized access and misuse, protecting privacy.

The regulatory landscape of FGG requires categorization and oversight to address complex ethical and legal challenges associated with genetic data use. A proposed framework emphasizes judicial oversight and informed consent, ensuring transparent and ethical collection and use of genetic information [28]. This framework advocates for judicial involvement in overseeing genetic technologies’ application, ensuring accountability and adherence to ethical standards.

Public discourse on ethical implications of genetic surveillance is crucial in shaping regulatory frameworks. Engaging stakeholders, including legal experts, ethicists, and the public, in discussions about genetic data usage risks and benefits can inform policy development balancing crime-solving capabilities with privacy rights. This collaborative approach is vital for creating effective, ethically sound regulations ensuring responsible use of advancements in forensic genealogy. As law enforcement increasingly utilizes public genetic datasets for investigations, addressing associated ethical,

legal, and privacy challenges is essential. By fostering stakeholder collaboration, we can establish a regulatory framework balancing public interest in solving crimes with the need to protect privacy rights, promoting transparency and accountability in forensic genetic genealogy [3, 28, 5, 12].

7.3 Ethical Implications of Familial Searching

The ethical implications of familial searching in forensic investigations are multifaceted, raising significant concerns about privacy, consent, and potential misidentification. Familial searching, which analyzes genetic data to identify potential relatives of individuals of interest, can inadvertently reveal sensitive familial information and disrupt personal relationships. A key challenge is the risk of misattributed paternity and the use of Y chromosome data, raising ethical questions about genetic privacy boundaries and consequences of uncovering unexpected familial connections [4].

The lack of comprehensive oversight in current studies exacerbates these ethical concerns, as biased data usage in criminal investigations can have far-reaching implications. This underscores the necessity for regulations and ethical guidelines to protect privacy rights while leveraging genetic genealogy's potential to solve cold cases [3]. Ensuring DNA match interpretations' reliability is crucial for responsible forensic genealogy, necessitating improvements to address ethical implications and guarantee accurate, fair outcomes.

Moreover, the potential for misidentifying distant relatives during familial searches poses practical and ethical challenges for law enforcement. This risk highlights the need for refined methodologies accounting for multiple potential relatives in databases, ensuring forensic identifications' reliability. User awareness and transparency in privacy policies are critical in addressing ethical implications of police access to genetic information without explicit consent. Discrepancies in transparency regarding genetic data practices, combined with a lack of user awareness about data utilization, emphasize the urgent need for clear communication about ethical, legal, and privacy implications of employing genetic data in forensic investigations. This is particularly relevant as law enforcement increasingly leverages advanced techniques like forensic genetic genealogy, utilizing public genetic datasets to identify potential relatives of DNA donors, raising significant ethical concerns about privacy and consent [12, 5].

Finally, while genetic genealogy can foster connections and understanding across diverse identities, it also risks reinforcing exclusivity and essentialist notions of belonging based on ancestry. As forensic genetic genealogy evolves, balancing its powerful capabilities with careful ethical considerations and adherence to legal frameworks is essential to realize these technologies' benefits without compromising individual rights and ethical standards [4].

7.4 Informed Consent and Ethical Approvals

The necessity of informed consent and ethical approvals in applying genetic genealogy techniques is paramount, ensuring individuals' rights and privacy are protected while advancing forensic investigations. Ethical considerations are integral to responsible genetic data use, as evidenced by the ethical approval obtained from the Ghent University Hospital Ethical Committee for studies involving kinship analysis [2]. Such approvals ensure adherence to ethical standards, safeguarding participants' rights and maintaining public trust in genetic research.

The evolving landscape of genetic genealogy necessitates robust consent mechanisms, particularly given changes to platforms like GEDmatch, which now require users to opt-in for law enforcement searches [13]. This shift underscores the importance of transparent and informed consent processes, enabling individuals to make educated decisions regarding their genetic data's use in forensic contexts. Legislative measures mandating judicial supervision for forensic genetic genealogy (FGG) searches and requiring affirmative consent from users further emphasize informed consent's critical role in maintaining ethical standards [28].

Future research should focus on developing more comprehensive consent frameworks, enhancing data security, and examining broader societal implications of genetic genealogy practices within law enforcement [9]. This includes exploring Y-chromosomal data integration with surnames and improving statistical methods for estimating match probabilities, enhancing forensic applications' accuracy and reliability [6]. Additionally, ethical approvals, such as those from Tongji Medical

College's medical ethics committee, highlight informed consent's importance from all genetic studies participants, ensuring ethical compliance and participant protection [11].

8 Conclusion

8.1 Future Directions and Recommendations

Advancements in kinship analysis and forensic genealogy are poised to be significantly influenced by technological progress and the establishment of comprehensive ethical frameworks. Enhancing STR panel designs alongside sequencing protocols is pivotal, particularly in improving nanopore sequencing's accuracy and efficiency for forensic applications. This refinement is essential for elevating genetic profiling techniques and ensuring the dependability of forensic investigations. Moreover, the development of an allelic ladder holds potential for augmenting cell line authentication, thereby enhancing forensic methodologies.

The synergistic integration of SNP genotyping with STR analysis presents promising prospects, especially in intricate kinship scenarios where SNP genotyping offers substantial evidential value. Future research should aim to refine models such as the MVN-FG by incorporating additional biological factors, thereby advancing the precision of forensic genealogy. Expanding kinship models to encompass affinal kin, stepkin, and blended family dynamics, along with considerations for non-binary gender identities, is crucial for fostering inclusivity and aligning with contemporary societal norms.

As the field evolves, ethical considerations remain a central focus, necessitating the development of robust regulatory guidelines and heightened public awareness regarding genetic privacy issues. Future research should prioritize the formulation of international best practices for the ethical use of genetic genealogy in investigations, examining the implications of emerging technologies in genetic testing, and addressing public concerns about privacy. This endeavor includes the creation of clearer privacy policies and enhanced user education about data usage, ensuring a balanced approach between the benefits of genetic genealogy and the protection of user privacy.

The establishment of comprehensive genetic profile databases is imperative for bolstering victim identification efforts, particularly in overcoming socio-political challenges. As the role of Y chromosome analysis evolves, exploring its applications in understanding human history and migration patterns will provide valuable insights into population dynamics.

Future efforts should also focus on developing detailed guidelines for selecting relevant populations in various forensic contexts, thereby ensuring the accuracy and equity of forensic analyses. By addressing these future directions and recommendations, kinship analysis and forensic genealogy will continue to advance, offering potent tools for forensic science while upholding ethical standards and societal values.

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