

Basic DNA Terminology

1. Nucleotides (bases) → the basic building blocks of DNA/RNA
 - a. There are four nucleotides in DNA (A, T, C, and G) and they pair together to hold each double-strand of DNA together (like rungs in a ladder)
 2. Single nucleotide polymorphism (SNP) → a difference at a single base position in the DNA
 - a. The most common type of genetic variation among the human population
 3. Short tandem repeats (STRs) → DNA regions with core repeated units that are commonly 2–6 bases in length
 - a. Y-STRs → STRs found in the y-chromosomes (males only)
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Annotated Bibliography

1 Familial Searching and Partial Matching

Debus-Sherrill, S. & Field, M. B. (2019). Familial DNA searching – an emerging forensic investigative tool. *Science & Justice*, 59(1), 20–28.

<https://doi.org/10.1016/j.scijus.2018.07.006>.

Familial DNA searching (FDS) is the deliberate search of a DNA database using specialized software to detect and statistically rank a list of potential candidates in the DNA database who may be close biological relatives (e.g., parent, child, sibling) to the unknown individual contributing the evidence DNA profile. All of this is then combined with lineage testing (mtDNA, Y-STR) to confirm or refute biological relatedness. Partial matching (PM) is the moderate stringency search of a DNA database using the routine search parameters within CODIS that results in one or more partial matches between single-source and non-degraded DNA profiles that share at least one allele at each locus. This indicates a potential familial relationship between the known individual in the DNA database and the unknown individual contributing the evidence DNA profile. While both

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processes are very similar, familial DNA searching is used more than partial matching for most types of forensic cases. However, familial DNA searching presents several ethical and legal controversies including privacy issues, issues surrounding racial/ethnic bias, public distrust/discomfort, and problems with family dynamics. While individuals whose DNA profiles are in CODIS have no expectation of privacy due to being convicted for a previous crime, the family members of these individuals that are not in CODIS should not be subject to searching because—as nonoffenders—they legally still have their right to privacy. Familial DNA searching also has a disproportionate impact on minority communities, due to the existing overrepresentation of racial and ethnic minorities in CODIS. Finally, the use of familial DNA searching during an investigation may interfere with an individual's social understanding of his or her family (e.g., if unknown paternity/adoption is uncovered or if it was not known that a family member was convicted of a crime). In addition to these ethical and legal complications, there are also several logistical issues with the use of familial DNA searches including practical challenges related to resources and costs, training and education needs, and the development and approval of policies to regulate its use.

Fortier, A. L., Kim, J., & Rosenberg, N. A. (2020). Human-Genetic Ancestry Inferences and False Positives in Forensic Familial Searching. *G3 (Bethesda)*, 10(8), 2893–2902.
<https://doi.org/10.1534/g3.120.401473>.

In forensic familial searching, a query DNA profile (collected from the scene) is tested against a law enforcement database (i.e., CODIS) to determine if the query profile represents a close relative of a database entrant. One challenge for familial search is that the calculations may require specification of allele frequencies for the unknown

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population from which the query profile has originated. The choice of allele frequencies affects the rate at which non-relatives are erroneously classified as relatives, and allele-frequency misspecification can substantially inflate false positive rates compared to use of allele frequencies drawn from the same population as the query profile. In familial identification, a true relative of the contributor of the query profile has only a partial match at the typed loci. As a result, close relatives of database entrants can be exposed to inappropriate forensic investigation when they have not contributed to query profiles. Accurate understanding of the magnitude of false positive rates in familial search is important for discussions regarding appropriate forensic application of the technique. To study properties of the false positive rate in familial identification, it is necessary to focus on the choice of allele frequencies used as part of familial-search likelihood ratio (LR) calculations. This is because the allele frequencies used in likelihood ratio calculations ultimately affect the probability that a database entrant and the query profile are identified as related, and their misspecification can influence false positive rates. This paper proposes that the allele-frequency misspecifications that produce the highest false positive rates can be avoided by using an ancestry-inference step in the familial search procedure. Forensic genetic profiles, even with the relatively limited marker sets they typically have, contain considerable information about genetic/biogeographic ancestry. Provided the estimated ancestry information is reasonably accurate, extreme misspecifications and the high false positive rates that result from them can be avoided.

Kim, J., Mammo, D., Siegel, M.B., & Katsanis, S. H. (2011). Policy implications for familial Searching. *Investigative Genetics*, 2(22), 1–9. <https://doi.org/10.1186/2041-2223-2-22>.

In criminal investigations, familial searching is the intentional search of an offender DNA database for inexact matches between DNA evidence profiles and offender and arrestee DNA profiles. Upon the identification of one or more partial match profiles, law enforcement may investigate purported family members of the partial matches as suspects. The FBI determined that familial searching policies should be decided by individual states; however, there is an expectation of reasonable uniformity among states regarding the use of CODIS. Familial searches involve one of two statistical analyses to interpret the data from molecular analysis. The identity by state (IBS) statistical analysis infers genetic similarity based on the number of matching markers between two profiles, regardless of how individual markers are inherited. Forensic analysts rank matches based on the highest number of matched markers to the lowest number (26 shared alleles = a full match). The kinship index statistical analysis (likelihood ratio) compares the probability of two profiles' being from related sources to the probability of the two profiles' being unrelated. Calculation of the likelihood ratio (LR) allows investigators to rank the individuals within the pool of candidates according to the probability that the evidence profile is related to the CODIS profile. With the increasing usage of familial searching in criminal investigations, there have been calls to create policy to better regulate the method. When developing policies for familial searching, legislators should take into account the impact of familial searching on select populations and the need to minimize personal intrusion on relatives of individuals in the DNA database. Because familial searching falls within state jurisdiction and not federal, the FBI does not have the

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authority to create a uniform policy/set of policies regulating familial searches. With this in mind, the following suggestions for regulating familial searching have been proposed: The FBI could be granted authorization to develop software to search CODIS for biological relatives. National policy could be established to type Y-STR markers for all prospective CODIS profiles. A national advisory consortium could be established to guide the development of statistical tools for familial searching. A national advisory consortium could be established to review cases and serve an ethics advisory function in policy implementation. States could determine for which crimes familial searching is appropriate. States could determine whose DNA profiles can be used to conduct a familial search.

Ram, N. (2011). Fortuity and Forensic Familial Identification. *Stanford Law Review*, 63(4),751–812. <https://www.jstor.org/stable/23058881>.

A “partial” match in a forensic context refers to two genetic profiles (one from a crime scene sample and the other from CODIS) that share some, but not all, of the thirteen core DNA loci that comprise a CODIS profile. A basic understanding of the science and history of DNA matching is necessary for an informed exploration and critique of state policies governing partial matching. In the United States, the most common form of forensic DNA typing examines 13 STR loci in the genome (DNA profiling/fingerprinting). These thirteen loci thus yield a total of 26 data points, each of which have multiple alleles. An individual inherits fifty percent of their genetic material from each parent and is expected to have roughly fifty percent of her genes in common with any full sibling. As a result, there is a significant probability that such close genetic relatives will also share a significant number of STR alleles. Children will share, at

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minimum, 13 alleles with each parent. Siblings, with the same mother and father, on average share 16.7 alleles. When searching CODIS for familial matches, an investigator must determine how strictly to constrain the match parameters. The CODIS software enables searches at three levels of specificity: high, moderate, and low stringency. A high-stringency search requires identity (exact match) both in number and type of all 26 of the alleles in the two samples. A moderate-stringency search returns matches in which the profile has all 26 alleles of the submission, but the submission contains additional material as well. A low-stringency search returns matches in which at least one allele is present even though the profile has additional alleles that the sample does not, or vice versa. Currently, two methods are jointly used for searching for known sibling pairs in mock offender databases: degree of allele sharing (how many alleles are the same between two individuals) and kinship matching (determining if individuals are biologically related). Partial matching methods presently have a significant rate of false positives (i.e., supposed genetic relatives who, upon analysis, turn out not to be related). Partial matches may be uncovered either fortuitously (by chance) or deliberately. While fortuitous partial matches appear in routine database searches, deliberate partial matches are the product of an intentional database search for such matches. Fortuitous partial matches may turn up as the result of lower-stringency search parameters. Most states have distinguished between fortuitously and deliberately discovered partial matches. This imposes significant structural and transparency costs, yet is supported by neither logic nor principle. As a result, much of the existing literature on partial DNA matching is focused on if such evidence should be admissible in court and, if it is continuously used in criminal investigations, to what extent should partial DNA matching be regulated.

Weir, B.S. (2004). Matching and partially matching DNA profiles. *Journal of Forensic Sciences*, 49(5), 1009–1014. PMID: 15461102.

The DNA profiles of two individuals can have 0 (a mismatch), 1 (a partial match), or 2 (a match) pairs of alleles that are the same at each locus. The likelihood ratios for two individuals having a specified degree of relationship versus being unrelated also depend on the numbers of matching and partially matching loci, but even unrelated pairs of individuals can have likelihood ratios that support hypotheses of relatedness. As the number of loci used for forensic profiling grows, the probability that a random person will have any specific profile will decrease. The forensic problem of interest is the probability that an untyped person has a profile given that has already been seen in a forensic database (a match to CODIS). As the size of forensic databases continues to grow, the numbers of matching loci for any two profiles in the data also grows, and it is of interest to predict how much matching is to be expected by chance. The degree of matching depends on the relationship among the people for whom the profiles are determined, and account must be taken of the relationships caused by the shared evolutionary history of humans as well as those for members of the same family. Therefore, it is necessary to create some statistical model to predict the likelihood of DNA matches/partial matches being a result of chance in order to reduce false-positive results. There is also a need to separate data relating to partial matches from data relating to complete matches, as both of these categories indicate two different degrees of allele sharing. Because partial matches and complete matches indicate different amounts of shared alleles, and differing degrees of relatedness to be inferred as a result, any statistical models created to predict likelihood of relatedness and/or likelihood of

matching-by-chance should have differing parameters based on if a match is partial or complete.

2 Traditional Profiling Methods

Comte, J., Baechler, S., Gervais, J., Lock, E., Milon, M. P., Delémont, O., & Castella, V.

(2019). Touch DNA collection – Performance of four different swabs. *Forensic Science International: Genetics*, 43(102113), 1–9.

<https://doi.org/10.1016/j.fsigen.2019.06.014>.

A collaborative study was performed by police forensic units, a DNA laboratory, and a forensic institute to compare the performance of four different swabs in controlled and quasi-operational conditions. Swabbing is the most versatile and one of the most frequently used methods of collecting touch DNA from crime scene evidence (i.e., clothing, surfaces). The technical characteristics of the swabs, such as the type and layout of the fibers as well as the size of the head are likely to influence collection and release of biological material efficiency. The four swab devices tested in this study are: COPAN 4N6FLOQSwabs™ Genetics, Puritan FAB-MINI-AP, Sarstedt Forensic, and the reference Prionics evidence collection kit. Also, three substrates (surfaces) having well-contrasted characteristics and being routinely used for DNA sampling by police forensic units were chosen: cover-less steering wheels of different materials (leather, hard plastic, imitation leather), screwdriver handles, and shirt/t-shirt collars worn for at least one day. In terms of the utility of swabs (durability, efficiency of handling, etc.), the COBAN swab heads were highly durable and the shaft offered an appreciated combination of flexibility and rigidity. Also, the breaking point of the head was

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appreciated by the laboratory as it facilitated the cutting of swabs. Regarding the Sarstedt swab, cotton fibers seemed to be tighter and did not absorb sterile water as well as the others. Also, its shaft was judged to be slightly too pliable. Concerning the Puritan swab, both the opening and the closure of the tube were considered unsafe and presented a potential risk for contamination because the shaft is not attached to the cap of the tube. There was also not enough room for labeling/writing on this tube. With all of this, the COBAN swab was rated best in terms of utility. The COBAN swab also had the largest percentage of DNA recovery from the various substrates, leading the study to conclude that the COBAN swab is overall the most efficient for trace DNA collection at the scene, However, the study did find that DNA collected on the COBAN swab had a higher rate of degradation after 12 days than the other three swabs did, meaning that COBAN swabs are not suited for DNA storage.

Kayser, M. (2017). Forensic use of Y-chromosome DNA: a general overview. *Human*

***Genetics*, 153, 125–133. <https://link.springer.com/article/10.1007/s00439-017-1776-9>.**

The human Y-chromosome is widely used in forensic DNA analysis, particularly in cases where standard autosomal DNA (from the numbered chromosome) profiling is not informative. A Y-chromosomal gene fragment is applied for inferring the biological sex of a crime scene trace donor. Haplotypes (a set of DNA variants inherited together on a single chromosome) composed of Y-chromosomal short tandem repeat polymorphisms (Y-STRs) are used to characterize paternal lineages of unknown male trace donors. Haplotypes (profiles) of Y-chromosomes are especially suitable when males and females have contributed to the same sample (i.e., samples from vaginal swabs in sexual assault cases). Y-STR haplotyping applied in crime scene investigation can exclude male

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suspects from involvement in crime, identify the paternal lineage of male perpetrators, highlight multiple male contributors to a sample, and provide investigative leads for finding unknown male perpetrators. The standard for Y-STR profiling is currently Sanger-based methods using capillary electrophoresis (CE) to type STRs based on DNA fragment lengths. Because Y-STR haplotypes are shared between paternally related men belonging to the same paternal lineage, Y-STR haplotyping is also suitable for solving paternity disputes of male offspring, other types of paternal kinship questions, and for familial searching. The strength of probability of paternity will depend on the allelic frequency of the Y-STR haplotype observed. The same applies in kinship analysis where the paternal relationship of one or more males is to be established or tested from hypotheses based on family record/archive information. Y-STR profiling is suitable to male identification cases involving human remains, such as in disaster victim and missing person identification where only distant relatives are available, as well.

Roewer, L. (2013). DNA fingerprinting in forensics: past, present, future. *Investigative Genetics*, 4(22), 1–10. <https://doi.org/10.1186/2041-2223-4-22>.

Forensic genetic fingerprinting (DNA fingerprinting, DNA profiling) is the comparison of the DNA in a person's nucleated cells with that identified in biological matter found at the scene of a crime or with the DNA of another person for the purpose of identification or exclusion. In the classical DNA fingerprinting method, radiolabeled DNA probes containing minisatellite (small DNA sequences lacking proteins) or oligonucleotide (made up of multiple nucleotides) sequences are hybridized to DNA that has been digested with a restriction enzyme. The sequences are separated by agarose electrophoresis (electrical current) and immobilized on a membrane either by Southern

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blotting (separating DNA sequences based on size) or immobilized directly in the dried gel. The polymerase chain reaction (PCR) method replaced the traditional fragment length-bases analysis methods in the 1990s because PCR had improved sensitivity, genotyping specificity, and speed. With the PCR-method, DNA profiling started to focus specifically on characterizing and analyzing short tandem repeats (STRs) to discriminate an individual's DNA from a mixed crime scene sample. Currently, forensic DNA profiling is performed using a panel of multi-allelic STR markers which are structurally analogous (similar) to the original minisatellites but with much shorter repeat tracts, making them easier to amplify and multiplex with PCR. In the United States, the FBI set the standard for STR analysis and DNA profiling in CODIS by naming 13 forensically significant STR loci. The probability that two individuals will have identical markers at each of 13 different STR loci within their DNA exceeds one out of a billion, which is what makes this number significant for forensic evidence. In addition to STRs, lineage markers (found on Y chromosomes and in mitochondrial DNA) have special applications in forensic genetics. Y chromosome analysis (Y-STR analysis) is very helpful in cases where there is an excess of DNA from a female victim and only a low proportion from a male perpetrator (e.g., sexual assault cases). Mitochondrial DNA (mtDNA) is of importance for the analyses of low level nuclear DNA samples, namely from unidentified (typically skeletonized) remains, hair shafts without roots, or very old specimens where only heavily degraded DNA is available. Information on the biogeographic origin of an unknown DNA could also be retrieved from a number of ancestry informative SNPs/markers (AISNPs or AIMs) on autosomes or insertion/deletion polymorphisms. However, estimation of biogeographic ancestry is relatively controversial due to its

methodology being based on European populations. It is also controversial due to biogeographic ancestry being treated as equivalent to race/ethnicity in non-scientific contexts (i.e., criminal cases), which can be unethical if it leads to racial/ethnic prejudice or bias. Finally, it is likely that DNA sequencing will soon replace methods based on fragment length analysis. With the emergence of next generation sequencing (NGS) technologies, the body of forensically useful data can potentially be expanded and analyzed quickly and cost-efficiently.

Verdon, T. J., Mitchell, R. J., & van Oorschot, R. A. H. (2014). Evaluation of tapelifting as a collection method for touch DNA. *Forensic Science International: Genetics*, 8(1), 179–186. <https://doi.org/10.1016/j.fsigen.2013.09.005>.

Tape-lifting is a technique commonly used to collect biological material (often touch/trace DNA shed from skin) for forensic analysis. A pressure-sensitive adhesive tape is applied to the test surface, leading to the direct transfer of particles on the surface to the tape. The use of tape-lifting for collection of touch DNA from fabrics is routine in many forensic laboratories. However, there is a lot of ambiguity and inconsistency of data relating to the effectiveness of different types of tapes for tape-lifting, the amount of tape-lifting required to generate a useful profile, and whether or not tape-lifting is more effective than swabbing from various substrates. Currently, the two tapes most commonly used in forensic casework are Scotch Magic tape and Scenesafe FAST minitape; these two tapes have different adhesive properties/strength. These two tapes were compared and evaluated in a study to determine which (if either) tape is best suited for forensic casework, and if tape-lifting overall is an effective method of DNA collection. Results of the study found that the mean percentage of DNA recovery was higher (with statistical

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significance) with the Scenesafe FAST tape than with the Scotch Magic tape for all surfaces except for one. The study also found that donor allele counts from the Scenesafe FAST tape were statistically significantly higher than from the Scotch Magic tape when sampling cotton and polyester surfaces; however, there was no statistically significant difference in allele counts from flannel surfaces. Both tape-lifting methods were also compared to swabbing, which is another method of trace DNA collection commonly used in forensics. Results of these comparisons found that Scenesafe FAST tape-lifting was the best method for collecting touch DNA from polyester, cotton, and poly-cotton blend surfaces. However, swabbing was the most effective method for collecting trace DNA from flannel surfaces. Overall, the results show that Scenesafe FAST tape is more effective in collecting quality trace DNA than Scotch Magic tape, likely due to Scenesafe FAST tape having better adhesive strength. It should also be noted that the biological material more deeply embedded in the surface will be harder to collect than that on the surface. As a result, after the surface material is removed by initial tape-lifting, collection of the remaining material may be beyond the capabilities of the tape and would require other methods such as swabbing.

3 Forensic Genetic Genealogy

García, Ó. (2021). Genealogía forense. Implicaciones sociales, éticas, legales y científicas.

Revista Española de Medicina Legal, 47(3), 112–119.

<https://doi.org/10.1016/j.reml.2020.06.001>.

Forensic genealogy (FGG) has three methodologies: the probabilistic (likelihood) method, the segment method and the KING method (exploratory approaches). Regardless of the method used, it is crucial to adequately interpret DNA results in order to make the best possible inference about the genetic relationship between two individuals. One major problem with forensic genealogical data interpretation is the companies doing genealogical testing do not all analyze the same number of SNPs, so sometimes a sample containing little or degraded DNA will not have maximum efficacy. This issue can be resolved by imputation, which is a method that consists of inferring DNA results that are common in different populations, but have not been tested directly in the user's DNA. During imputation, the absent DNA is predicted (imputed) based on the DNA in adjacent locations by relying on the principle of linkage disequilibrium (DNA locations being inherited together in groups). While imputation alleviates some logistical issues with forensic genealogy, several ethical concerns still surround its use in criminal investigations. These concerns include the use of public DNA databases by law enforcement agencies, lack of validation and formal education/training, potential for data loss and/or hacking genetic data, revelation of health and/or ancestry information without an individual's consent, and more. To address these concerns, the U.S. Department of Justice (DoJ), the American Society of Crime Laboratory Directors (ASCLD), and the Scientific Working Group on DNA Analysis Methods (SWGDM) have published similar

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proposals for the criteria to be considered before undertaking analysis using forensic genealogy. These criteria include limiting the use of forensic genealogy to serious offenses (e.g., murder, sexual assault), using high throughput technology that is able to perform molecular analyses with high accuracy and precision, and using forensic genealogy only as a last resort (i.e., after all other forensic analyses failed to provide any investigative leads).

Granja, R. (2023). Citizen science at the roots and as the future of forensic genetic genealogy. *International Journal of Police Science & Management*. 1–12.

Forensic genetic genealogy (FGG) is an interdisciplinary practice that combines genomics and computer database technologies, as well as traditional and genetic methods of genealogical research to identify unknown individuals by reconstructing their ancestral lineages and drawing out their family trees. In the United States, forensic genetic genealogy investigations fall into four broad categories: criminal investigations (~ 80%), investigations to identify unknown deceased individuals (~19%), identification of mass disasters and identification of living persons (~ 1%). Because forensic genetic genealogy is dependent on the complexity of the search required to identify an individual, the amount of work and resources put into each investigation are highly variable. In addition to this logistical issue, there are several ethical issues and sociolegal controversies surrounding forensic genetic genealogy. The ethical controversies and social implications of using forensic genetic genealogy include: an enlargement of the type of information that can be retrieved from DNA, an expansion of populations involved in law enforcement searches, issues associated with consent, and negative impacts on public trust. Legal and/or policy regulation of the use of forensic genetic genealogy is also

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difficult, since companies involved with direct-to-consumer genetic testing have different policies and practices with varying degrees of restrictions and cooperation. For these reasons, it is recommended that citizen science be utilized as the framework for doing forensic genetic genealogy. Citizen science refers to the substantial increase in citizens engaged in scientific endeavors, which includes some combination of contractual projects, contributory projects, collaborative projects, co-created projects, and collegial contributions. Understanding the challenges posed by forensic genetic genealogy is of paramount importance in understanding the role of citizen science because these challenges help us to consider and critically engage with the potential implications for the forensic epistemic culture of having citizens with very diverse educational training and professional skills actively involved with and contributing to this type of investigative work.

Kling, D., Phillips, C., Kennett, D., & Tillmar, A. (2021). Investigative genetic genealogy: Current methods, knowledge and practice. *Forensic Science International: Genetics*, 52, 1–23. <https://doi.org/10.1016/j.fsigen.2021.102474>.

Investigative genetic genealogy (IGG) is the process whereby dense SNP data—commonly comprising more than half a million markers—are employed to infer distant (degrees of relatedness exceeding that of first cousins) relationships. More specifically, investigative genetic genealogy involves methods of relationship matching and SNP analysis on an enlarged scale that are used in a forensic setting to identify a suspect in a criminal investigation or a missing person. This process results in an inference of relatedness, which has the primary aim of determining whether regions of DNA are shared identical by descent (through common ancestry). Investigative genetic genealogy is often

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compared to familial searching, which is the search of the database conducted after a routine search for the purpose of potentially identifying close biological relatives of the unknown forensic sample associated with the crime scene profile. However, key differences in the two processes include the source of genetic data being used for comparison and the resulting ethical and (potential) legal implications. Genetic data used in familial searches is gathered from state or local indices (CODIS) that are already in the possession of law enforcement. Investigative genetic genealogy, on the other hand, primarily uses genetic data obtained from commercial genealogical databases hosted by private companies (e.g., Ancestry, GEDMatch, 23andMe). Because individuals whose DNA profiles are in state/local indices have already been convicted of a crime, they legally are considered to have forfeited their rights to privacy. Therefore, familial searching has far fewer privacy implications than investigative genetic genealogy, which extends searches to both close and distant relatives who are in a genealogy DNA database (not all of whom have given consent for their profiles to be used). The primary concerns around investigative genetic genealogy are the lack of transparency on part of law enforcement and that many of the technical details around the analysis of forensic DNA for long-range familial searching are still not in the public domain.

Kling, D. & Tillmar, A. (2019). Forensic genealogy—A comparison of methods to infer distant relationships based on dense SNP data. *Forensic Science International: Genetics*, 42, 113–124. <https://doi.org/10.1016/j.fsigen.2019.06.019>.

Forensic genetic genealogy (FGG) has multiple methodologies, each of which have their advantages and disadvantages. Genealogy, in general, is the inference of familial relationships by assigning the degree of relatedness between two individuals

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(classification). In a forensic context, it is vital to correctly determine familial relationships and classify degree of relatedness because misclassification can lead to false accusations—all of which could result in wrongful conviction. There are three methodologies for determining relatedness used in forensic genetic genealogy: the method using likelihood, and the KING and segment methods (both of which are exploratory in approach). In the method using likelihood, the likelihood ratio (LR) is the conditional probability of observing some genetic marker data for a set of individuals and some precise hypothesis about the relationship between the individuals. The likelihood ratio is used as a means to measure the weight of the genetic evidence, rather than being serving as the evidence itself. The advantage of the likelihood method is that the likelihood ratio is computer-generated, and is less subject to the biases and interpretations of a forensic analyst. However, because the likelihood ratio is computer-generated based on allele frequencies, results are highly sensitive to the position of genetic markers (genetic maps) and can potentially lead to false classifications if this factor is unaccounted for. An exploratory approach to determining genetic relatedness relies on characterization of the alleles themselves rather than a computation of their frequencies. Because of this, the data produced by both exploratory methods is used as the evidence in a forensic investigation and not as a weight of evidence. The KING method counts the number of shared alleles identical by state for each marker and averages over a large number of markers yielding a measure of the degree of relationship. The segment method estimates the total length of shared genomic segments by measuring segments along the chromosomes where a pair of individuals shares at least one allele along the complete segment. Both the exploratory methods have the advantage over the likelihood method

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for classifying non-related/extremely distantly related individuals. However, the likelihood method is best for classifying the degree of relatedness between related individuals.

Machado, H. & Silva, S. (2022). Investigative Genetic Genealogy: An Ethical and Privacy Assessment Framework Tool Is Needed. *Forensic Science Review*, 34(4), 17–19.

Forensic genetic genealogy (FGG)/investigative genetic genealogy (IGG) is different from traditional DNA profiling (fingerprinting) in several ways. Traditional DNA profiling uses short tandem repeats (STRs) to match a crime scene sample against DNA profiles that are already in the law enforcement's database. Traditional DNA profiling is highly regulated, and has set standards for molecular analyses and application of resulting data in a legal context. Investigative genetic genealogy, on the other, is very unregulated and is based on matching single nucleotide polymorphisms (SNPs) from a crime scene sample to similar SNPs found in the databases of private genealogy companies (e.g., 23andMe, Ancestry) in hopes of identifying an individual(s) that is related to the perpetrator of a crime. As a result, forensic genetic genealogy creates concerns surrounding consent and privacy. While consumers may consent to uploading their genetic profiles to sites, their biological relatives have not consented to their now indirect inclusion in these databases. Private genealogy companies require the informed consent of the consumer submitting their DNA samples and include a disclosure that their genetic data can be used by law enforcement. However, the focus on individual-based consent narrows ethical discussion by shielding other substantive political and societal issues from critical scrutiny (e.g., public interest issues, societal good, state power, and oversight mechanisms). There are also concerns around how transparent private

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genealogy companies are about sharing data with law enforcement agencies, the criteria for law enforcement to access data from these companies, quality assurance (policies surrounding sharing genetic information with third-parties and health insurers), and balancing individual's right to privacy with the need to insure public safety by identifying perpetrators of crime (proportionality). All of these concerns require consideration of the adoption of a broader ethical and privacy assessment approach to investigative genetic genealogy, such that the process is developed in partnership with all relevant professionals and stakeholders. There also needs to be guidance to limit activity in investigating potential distant relatives, as well as clear indications related to the kind of genetic information that would be revealed in the analysis in addition to genealogical information.

Ram, N. Guerrini, C. J., & McGuire, A. L. (2018). Genealogy databases and the future of criminal investigation. *Science*, 360(6393), 1078–1079.

<https://doi.org/10.1126/science.aau1083>.

The use of forensic genetic genealogy (FGG) by law enforcement agencies raises questions surrounding its legal implications. Because this is a relatively new means of investigation, it is difficult to determine to what extent evidence gained from forensic genetic genealogy should be used in a criminal court. Opponents of using forensic genetic genealogy argue that using genetic data from private genealogy companies to search for a suspect is a violation of one's Fourth Amendment protections against warrantless search and seizure. However, because the genetic data is from databases where individuals voluntarily give their DNA, the Fourth Amendment does not apply to forensic genetic genealogy as there is no expectation of privacy. The Genetic Information Nondisclosure

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Act (GINA) only protects genetic information from being used by health insurers; therefore, this protection also does not apply to forensic genetic genealogy being performed by law enforcement agencies. It has also been argued that the use of data from genealogical databases by law enforcement during criminal investigations should be considered a violation of the Health Insurance Portability and Accountability Act (HIPAA), which protects an individual's health information from health agencies, insurance companies, and other stakeholders. However, the private companies that host genealogical data require that consumers agree to private policy disclosures prior to submitting their DNA samples. The disclosures for a majority of these private genealogical companies explicitly state that consumers' genetic data will be given to law enforcement, without a warrant, if required during a criminal investigation. By agreeing to these disclosures and submitting to the genealogical databases, consumers no longer have a legal right to privacy protections. Because no legal grounds currently exist to prohibit the use of forensic genetic genealogy by law enforcement agencies, it is recommended that some policy/policies be put in place to better regulate forensic genetic genealogy. A policy that is legally similar to the Stored Communications Act (1986), but applicable to genetic data and information, would be most appropriate for regulating forensic investigative genealogy.

4 Forensic Pathology

De Boer, H. H., Berger, Ce. E., & Blau, S. (2021) Providing a forensic expert opinion on the

“Degree of force”: Evidentiary considerations. *Biology*, 10(12), 1–9.

<https://doi.org/10.3390/biology10121336>.

In forensic cases where the cause of death is blunt-force trauma (injury caused by an impact to the body) or certain types of asphyxiation (suffocation), forensic pathologists and forensic anthropologists are asked to assess the “degree of force” by law enforcement. The forensic pathologist’s/anthropologist’s assessment of the degree of force is often used by law enforcement investigators to determine whether an individual’s manner of death is a homicide or accident. The opinion of the degree of force provided by forensic practitioners is perceived as particularly helpful in court during a criminal trial, which is why forensic practitioners are asked for this assessment frequently. This perceived value of opinions on the degree of force appears to be based on three assumptions, namely that proportional relationships exist between (i) the intent of an offender/assailant and the amount of force they use, (ii) the amount of force that an offender/assailant uses and the amount of force that is actually transferred to the body of the decedent (deceased), and (iii) the amount of force that is applied on the body of the decedent and the severity of injury. From a methodological perspective, however, assessing the degree of force for a blunt-force trauma or asphyxia has several theoretical and practical difficulties. Primarily, estimations of force alone are inadequate due to high subjectivity both in terms of the durability of a victim’s body and the characteristics surrounding the impact. An injury that is mild when inflicted on one individual’s body can be fatal when inflicted upon another individual’s body. Also, an impact applied at one

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velocity (speed) to a particular part of the body could lead to minor injury while that same type of impact applied either to a different part of the body or at a higher velocity could result in death. Because so much subjectivity is involved in a forensic concept of “force”, many forensic pathologists/anthropologists are ultimately unable to give an answer that would be legitimate enough to serve as evidence of a homicide. Many forensic practitioners opt to respond with non-answers (e.g., “no comment”), or with strict scientific answers such as “the force was sufficient to result in skeletal injury”. Occasionally, however, a forensic practitioner might actually attempt to assess degree of force in a way that leads to an inference of a specific manner of death (i.e., homicide). These assessments are based on subjective interpretation of injuries; therefore, it is recommended that such assessment not be admissible in a criminal court.

Lunn, M. (2017). Cause and Manner of Death. *Essentials of Medicolegal Death Investigation*. Oxford, UK: Elsevier Academic Press.

The medicolegal death investigator is responsible for determining how an individual died, as well as all of the circumstances surrounding that individual’s death. In order to do this, the medicolegal death investigator must establish the mechanism of death, cause of death (COD), and manner of death (MOD). Mechanism of death is the physiological disturbance(s) and change(s) that is caused by the overarching cause of death, and is often not specific to the particular injury/disease that caused the individuals’ death. Cause of death relates to the natural disease(s) and/or the injury/injuries causing the physiological change(s) leading to death. The cause of death could either have occurred just before the death event (acute) or the result of a chronic disease or injury. There are hundreds of different diseases and injuries, as well as thousands of combinations of

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diseases and/or injuries, that fall under types of cause of death. Determination of the manner of death is based on the circumstances surrounding the case (from the evidence), and therefore relies on the medicolegal death investigator's ability to accurately capture all of the evidence from the death scene. Lacking adequate information would greatly decrease the medicolegal death investigator's ability to make an accurate assessment of the cause and mechanism(s) of death, all of which could lead to an inaccurate determination of manner of death. Manner of death is determined only after all necessary body examinations and review of the evidence has been completed, making it the last piece of the death certification. There are five categories for manner of death used in the United States: natural, accident, suicide, homicide, and undetermined. Natural deaths, which make up approximately half of the cases evaluated by medicolegal death investigators, are those cases where a disease process(es) is the only reason attributed to an individual's death. A death is ruled accidental when some external event and/or substance contributes to death without foul play; the most common accidental deaths are drug overdoses, motor vehicle accidents, and falls. Suicide is the explicit or implicit intent by the decedent (deceased) to end their own life, or action(s) taken by the decedent that a normal prudent person would understand as an event that would end their life. Homicide is a death caused by the actions of another individual(s) including blunt-force traumas, sharp-force traumas, projectile (gunshot) traumas, strangulation, and more. Finally, a death is ruled undetermined when, even after autopsy, the reasons and circumstances surrounding the death event remain scientifically unknown.

Lunn, M. (2017). *Special Death Investigations. Essentials of Medicolegal Death*

Investigation. Oxford, UK: Elsevier Academic Press.

Ideally, every death should be approached in the same way by the medicolegal regardless of the victim and/or the circumstances surrounding the death event. However, this is often not the case due to the general lack of standardization of death investigation in the United States. In the U.S., medicolegal death investigation typically falls under a medical examiner's (ME) system, a coroner's system, or some combination of both. In medical examiner's systems, the medicolegal death investigator does not certify death, or determine cause/manner of death, unless they are a physician (forensic pathologist) or another healthcare provider (e.g., physician's assistant, forensic nurse, practitioner). The medicolegal death investigator in a coroner's system, on the other hand, almost always certifies death and determines cause/manner of death, even if they are not a physician or certified healthcare provider. This lack of uniformity becomes even more concerning when dealing with special death investigations, which are those death investigations involving vulnerable decedents (children or the elderly), victims of drowning, or deaths in custody. Death in custody refers to the death of a person in the custody of the police, other law enforcement authorities, or in jail/prison. When an individual dies as a result of police/law enforcement action, the medicolegal death investigator should rule the manner of death as homicide. However, ruling such a death as a homicide does not mean that anyone will be legally punished for the in-custody death. The way in which the medicolegal death investigator examines the body will inform them of the mechanism and cause of death, which will ultimately be used in a court setting to determine whether a police officer and/or a law enforcement agency should be legally responsible for a death

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in-custody. However, because medicolegal death investigators have varying qualifications, education levels, and training backgrounds throughout the country, deaths in-custody with very similar circumstances could lead to very different legal outcomes based on the medicolegal death investigator's reports.

Mitchell, R. A., Diaz, F., Goldfogel, G. A., Fajardo, M., Fiore, S. E., Henson, T. V., Jorden, M. A., Kelly, S., Luzi, S., Quinn, M., & Wolf, D. A. (2017). National Association of Medical Examiners Position Paper: Recommendations for the definition, investigation, postmortem examination, and reporting of deaths in custody. *Academic Forensic Pathology*, 7(4), 604–618. <https://doi.org/10.23907/2017.051>.

Deaths in custody refers to those deaths in which the circumstances of the death place the decedent in either direct or indirect contact with law enforcement (such as incarceration, apprehension, and pursuit). The National Association of Medical Examiners (NAME) commissioned an ad hoc committee to provide recommendations for the investigation, examination, and reporting of deaths in custody. It is recommended to define deaths in custody as those deaths that occur under the perceived or physical control or restraint of a law enforcement officer, a correctional officer (including a private correctional officer), or an authorized employee or agent of a district juvenile secure facility or youth residential facility. This definition would also include the deaths of individuals incarcerated in, committed to, or on work release from a jail or correctional facility (including contract facility) or a psychiatric hospital; individuals committed to a juvenile secure facility; and judicial executions. All deaths that occur in the custody of law enforcement, while being pursued by law enforcement, or while detained by law enforcement, must be reported to the medical examiner (ME) or coroner immediately.

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The coroner/medical examiner should determine what body examination (external examination or autopsy) is most appropriate for the case. In addition to examination of the body, the medical examiner/coroner should also consider doing the following: collection of biological evidence (blood, semen, etc.) from the body, radiological examination, toxicological analysis, histological examination, microbial analysis, ancillary testing, and organ/tissue recovery. Only after completing a thorough examination of the body and associated evidence should the medical examiner or coroner execute the death certificate. Certification of deaths in custody may come under increased scrutiny and concerns may arise when the manner of death determination is performed by the agency that is under investigation. In these instances, effort should be made to relinquish this determination to either another investigative body within the government organization, a neighboring medical examiner/coroner, or a truly independent agency should be identified to ensure death investigative transparency and community confidence.

Ruiz, L., Posey, B., Neuilly, M. A., Stohr, M. K., & Hemmens, C. (2018). Certifying Death in the United States. *Journal of Forensic Sciences*, 63(4), 1138–1146.

<https://doi.org/10.1111/1556-4029.13689>.

A comprehensive overview of statutes surrounding death investigation in all fifty states highlights the inconsistencies in death investigation in the United States. Statutes designate a broad range of individuals as responsible for the classification and certification of death. Those vary by state and set of circumstances and can include medical examiners, coroners, pathologists, other physicians, registered nurses, and more. States with coroner's systems do not require medicolegal death investigators to have any

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formal background or education in pathology or medicine. Medicolegal death investigators in coroner's systems are simply trained at hiring/on the job; for most states with this system, the only requirements to become a medicolegal death investigator are being eighteen years of age and holding a high school diploma. Medical examiner's systems, on the other hand, require medicolegal death investigators to hold an advanced medical degree and have training via formal fellowships or medical assistantships. Medicolegal death investigators in medical examiner's systems typically have specialized training in pathology, forensic medicine, and/or human anatomy. Such heterogeneity of the death certification process and personnel in the United States has led to calls for more unified medicolegal standards of qualifications, training, certification, and registration of death investigators. Current state statutes use vague language regarding legal certification of death, which can lead to misinterpretation. This vagueness of statutory language can also make establishing liability in cases of malicious intent and professional misconduct difficult. In addition to general vagueness, language about death certification in state statutes typically does not distinguish between subtypes of medicolegal investigators. A majority of state statutes use the titles "medical examiners" and "coroners" in a broad sense to encompass all investigators, and use unspecified medicolegal jargon. To rectify these issues, it is recommended that current legislation be updated to include more concise and understandable language. It is also recommended that policies establishing some uniform qualification(s) to be a medicolegal death investigator and standards for methods of death investigation be implemented.