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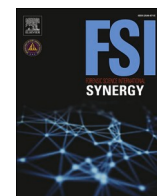
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Law enforcement use of genetic genealogy databases in criminal investigations: Nomenclature, definition and scope

Oliver M. Tuazon^{a,*}, Ray A. Wickenheiser^b, Ricky Ansell^{c,e}, Christi J. Guerrini^d, Gerrit-Jan Zwenne^a, Bart Custers^a

^a Center for Law and Digital Technologies (eLaw), Institute for the Interdisciplinary Study of the Law, Leiden Law School, Leiden University, Kamerlingh Onnes Building, Steenschuur 25, 2311 ES, Leiden, the Netherlands

^b New York State Police Crime Laboratory System, Forensic Investigation Center, 1220 Washington Avenue, Building #30, Albany, NY, 12226-3000, USA

^c Swedish Police Authority, National Forensic Centre, SE-581 94, Linköping, Sweden

^d Baylor College of Medicine, Center for Medical Ethics and Health Policy, Houston, TX, 77030, USA

^e Department of Physics, Chemistry and Biology, Linköping University, Sweden

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ABSTRACT

Although law enforcement use of commercial genetic genealogy databases has gained prominence since the arrest of the Golden State Killer in 2018, and it has been used in hundreds of cases in the United States and more recently in Europe and Australia, it does not have a standard nomenclature and scope. We analyzed the more common terms currently being used and propose a common nomenclature: investigative forensic genetic genealogy (iFGG). We define iFGG as the use by law enforcement of genetic genealogy combined with traditional genealogy to generate suspect investigational leads from forensic samples in criminal investigations. We describe iFGG as a proper subset of forensic genetic genealogy, that is, FGG as applied by law enforcement to criminal investigations; hence, investigative FGG or iFGG. We delineate its steps, compare and contrast it with other investigative techniques involving genetic evidence, and contextualize its use within criminal investigations. This characterization is a critical input to future studies regarding the legal status of iFGG and its implications on the right to genetic privacy.

1. Introduction

Improved DNA analysis technology and public interest in using genetics for genealogical research has increased the size of publicly available databases. Many datasets of previously difficult-to-access genealogical information, such as census information and obituaries, have become available online, enabling remote genealogical research fueled by these large direct-to-consumer genetic databases.

Recently, the technique of finding genetic relatives using commercial databases has been applied to finding the perpetrators of unsolved criminal cases. The combination of genetic comparison of crime scene perpetrator DNA profiles in public databases and genealogical research has provided a solution to the growing list of cold cases whose perpetrators left DNA traces at the crime scene, where law enforcement still could not produce suspect investigational leads after using other DNA-based methods, including searches of their own law enforcement DNA databases. By law enforcement we mean the official governmental

agency responsible for solving crimes, and more specifically in the context of this article, in identifying perpetrator/s who left their DNA at the crime scene under investigation.

Although the technique's first reported use was in early 2018, it became widely known for its use in identifying the Golden State Killer (GSK) later that year [1,2]. Its first reported use in Europe was in Sweden a year later to solve a 15-year old double-murder case in Linköping [3,4]. In 2023, Norwegian law enforcement reported its use in finding a suspect in a 20-year old murder case [5], and the Australian police in solving a couple of cold cases [6]. In the same year, the Dutch government announced its plan to use the technique to solve two cold cases in a pilot study [7,8], as well as in New Zealand [9].

Despite expanding use of the technique, its terminology and definition remain unsettled. Its short history is plagued by uncertainty around what to call it. Commenting on its use in the GSK case, Guerrini et al. gave it a description more than a name when they referred to it as the 'forensic use of genetic genealogy databases' [10]. It was later referred

* Corresponding author.

E-mail address: o.m.tuazon@law.leidenuniv.nl (O.M. Tuazon).

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to simply as ‘forensic genealogy’ or ‘forensic investigative genealogical searching’ [1]. Two dominant terms are currently used interchangeably to refer to the same method, ‘forensic genetic genealogy’ (FGG) [11–13] and ‘investigative genetic genealogy’ (IGG) [14–16]. In 2019, the U.S. Department of Justice introduced yet a third term in its interim policy on the technique: ‘forensic genetic genealogical DNA analysis and searching’ (FGGS) [17]. More recently, the U.S.-based National Technology Validation and Implementation Collaborative (NTVIC) distinguished FGG from IGG and combined them into one term, forensic investigative genetic genealogy (FIGG) [18,19].

As the technique gains popularity and acceptance worldwide, we propose the use of a unified terminology not only for academic purposes but also for ordinary usage, and more specifically for law enforcement use. A common nomenclature will promote harmony and avoid confusion across borders and will facilitate further research and progress in this area, for example, in assessing the technique’s impact on the right to genetic privacy.

We agree with NTVIC on the use of FIGG, given that the nomenclature contains all the elements that are essential to the technique, particularly ‘forensic’ and ‘investigative’, one of which is missing in either FGG or IGG. However, we propose that the word ‘investigative’ comes first, making it function as a qualifier or an adjective to the term FGG. FGG can then be used as a more general term for the forensic use of genetic genealogy databases in identifying individuals, say, those of unidentified human remains (UHR) outside criminal investigations. Its use by law enforcement in criminal investigations follows the same method but for a different purpose—to produce suspect investigational leads, thereby making it a proper subset of FGG. In this regard, we use the term ‘investigative FGG’ (iFGG) when the technique is used for that specific purpose of solving crimes by law enforcement. When applied to a court setting, FGG is the proper term to be used when proving inheritance, identification of missing persons or some other non-investigative application, whereas iFGG is the proper term to be used for criminal investigations—including identification of UHRs within those investigations—by law enforcement. This distinction is important because processing of DNA-related data by law enforcement, or by any public authority for that matter, has its own specific privacy standards to follow [20–22].

This is the first paper *to date* that reviews and analyzes the current terminologies being used to refer to the law enforcement use of genetic genealogy databases in criminal investigations, and at the same time proposes a common terminology and defines its scope. For the purpose of clarifying its scope and to avoid confusion, we reviewed other DNA-based methods currently used by law enforcement in criminal investigations, tracing them from the origins of the use of DNA in forensics. In that way, we illustrate that iFGG is currently law enforcement’s method of last resort in solving crimes. We begin with a short background on the use of DNA in criminal investigations (section 2), which includes DNA profiling in the context of law enforcement and the types of DNA databases that may be accessible to them. We then discuss other DNA-based methods that may be employed by law enforcement before resorting to iFGG (section 3). The reasons for the use of iFGG as a more acceptable nomenclature vis-à-vis the existing ones are then presented (section 4), followed by a clarification of the actual steps involved using iFGG in conjunction with other DNA-based law enforcement tools (section 5).

2. DNA in criminal investigations

The decoding of the human genome is the object of the Human Genome Project (HGP), which released the first-ever human genetic blueprint in 2003 [23,24], although a complete human genome sequence was only published in April 2022 [25]. Every person’s genome is unique to them. It combines their parents’ genetic material into a new biological identity. Although some studies have shown that rare mutations happen after the zygote has split into two, they are hard to detect in

usual forensic DNA investigations [26–28]. At the same time, DNA is for the most part (more than 99%) also shared by all human beings [29]. It is this dual characteristic of both being individual and shared that allows its exploitation for law enforcement use in criminal investigations. McGonigle uses the word ‘dividual’ or ‘dividuality’ to refer to the ‘shared nature of genomic data’ [30].

2.1. Origins of the forensic use of DNA

Prior to the Human Genome Project, the uniqueness of DNA was explored using various techniques, including Sir Alec Jeffreys’ DNA fingerprinting method as reported in the mid-1980’s [31]. Jeffreys put his DNA profiling discovery to practical use in 1986 when the police asked him to apply it in solving the sensational Pitchfork rape and murder cases. The initial results led to the exoneration of a man who wrongly confessed to the murder of one of the victims, and to the eventual conviction of Pitchfork himself [32,33].

Following the success of the forensic application of Jeffrey’s DNA fingerprinting method, law enforcement officers have been keen to exploit its use in more criminal investigations, specifically in identifying unknown criminals who left biological material at crime scenes. The term ‘biological material’ is used because DNA can only be extracted from crime scenes that contain cells or tissues left by the criminal perpetrator, the most common of which is called ‘touch or trace DNA’ [34,35].

The development of various DNA-based techniques in criminal investigations went and continue to go hand-in-hand with breakthroughs in DNA technology. Over time, they become less resource intensive—with more automated systems that facilitate quicker interpretation of results—and less costly, while increasing their sensitivity and the level of information they provide. Newer techniques used by law enforcement are based on genetic profiles comprised of short tandem repeats (STRs), and more recently, single nucleotide polymorphism (SNPs).

2.2. DNA profiling in law enforcement: STRs and SNPs

Owing to the uniqueness of a person’s DNA, save for monozygotic twins, DNA profiling has become a standard method in criminal investigations. The current international method of choice measures the presence of variant STRs in specific positions (‘loci’; singular, ‘locus’) of the DNA [36], mainly in its non-coding regions [37]. The number of STR loci amplified depends on the national system, usually between 13 and 23 loci. Since these DNA databases are built over time, the older entries contain fewer STR loci data, which obviously provide less information, and less chances of getting matches using indirect matching techniques, especially for familial searching. A case in point: The Netherlands Forensic Institute upgraded its marker systems from 10 to 15 to the now 23 STR markers [38].

STR profile alleles are converted into numerical equivalents that are stored in law enforcement DNA databases. The most commonly used database system is the Combined DNA Identification System (CODIS) developed in the USA [39]. One disadvantage of STR-based databases is that profile matching is limited to samples available in these databases, and it has a limited capacity for familial searching in a sense that only close relatives—such as parents/offspring and full siblings—can be linked to the unknown sample [15]. This is one reason why the GSK case remained unsolved for 40 years, aside from limited uploading of DNA profiles in CODIS as well as limited cross comparisons among states. It was argued, however, that the GSK could have been identified a decade earlier given that his brother had prior conviction but DNA collection and familial searching laws were still non-existent at that time [40].

The piling-up of cold cases despite use of STR profile data led law enforcement officers to seek other ways of exploiting the inherent identifiability of DNA data through single nucleotide polymorphisms (SNPs). SNPs are variations in the DNA sequence involving a single nucleotide—that is, either a guanine (G), adenine (A), thymine (T) or

cytosine (C)—and whose specific alterations or ‘variations’ may be used to ‘distinguish people for purposes of biological relationship testing’ [17]. Barring a mutation, these SNPs are inherited intact by genetic relatives. Hence, they are useful in tracing genetic relationships.

Law enforcement agencies usually outsource the generation of SNP profile data to external laboratories, whose accreditation requirement depends on prevailing legislation. These are then used for the identification of human remains or uploaded to third-party commercial DNA databases for SNP matching. The power of SNP profile matching rests on its ability to predict long distance familial relationships (see Table 2 for a detailed comparison). The total amount of DNA shared is expressed in centimorgans (cM) [16], where a higher value means a higher degree of familial closeness (Table 3). For example, individuals share 50% of their DNA with their biological parents, which on average means around 3485 cM. Compare that to a first cousin who shares an average of 866 cM, and a second cousin and a third cousin who share an average of 229 cm and 73 cM, respectively, with the individual in question. In this way, the GSK was traced from his third cousin who uploaded his/her SNP profile data in GEDmatch [41]. Wickenheiser claims that it is a ‘probable 4th cousin’ [1].

As DNA sequencing becomes cheaper, whole genome sequencing (WGS) and massive parallel sequencing (MPS) have been developed for the generation of more accurate STR and SNP profile data for forensic use. In most cases using iFGG, SNP profile data have been generated using microarray genotyping [42]. However, this method requires more DNA than is usually present in crime scene samples. Forensic samples are usually in the form of ‘touch DNA’, which contain low amounts of DNA [43]. In that regard, WGS has been reported to be a good alternative to produce SNP profile data for iFGG [42]. The DNA Doe Project makes use of WGS, from which SNP datasets are generated for upload to GEDmatch [40].

Along the same line, massive parallel sequencing (MPS) has been developed that is specifically targeted to improving the generation of both STR and SNP profile data [44,45]. MPS also provides a solution to problems of low quantity and degraded DNA gathered from crime scenes [46]. A difference in a single base—ACTGACTG vs. ACTGACCG—is clearly distinguishable in MPS but not in standard STR amplification and capillary electrophoresis, which measures fragment lengths rather than individual base pair composition [47]. MPS also promises to provide a more accurate estimation of the number of contributors in a mixed DNA sample, which is usually the case among crime scene samples [48]. MPS can also be used for other forensic purposes, like externally visible characteristics (EVCs) and biogeographical ancestry (BGA), whose legal implications still have to be sorted out [49].

Table 1

Size of law enforcement (LE) and third-party (3P) DNA databases (arranged in descending order, as of October 2023).

Database	Country of origin	Year launched	DNA markers	Size (min.)
AncestryDNA (3P)	United States	2012	SNP	23 M
National DNA Index (LE)	United States	1998	STR	20.6 M
23andMe (3P)	United States	2006	SNP	14 M
MyHeritage (3P)	Israel	2003	SNP	7.4 M
National DNA Database (LE)	United Kingdom	1995	STR	7.1 M
FamilyTreeDNA (3P)	United States	1999	SNP	2 M
GEDMatch (3P)	United States	2010	SNP	1.4 M

Sources for the US and UK forensic DNA databases: NDIS [le.fbi.gov/science-and-lab-resources/biometrics-and-fingerprints/codis/codis-ndis-statistics]; NDNAD [www.gov.uk/government/statistics/national-dna-database-statistics]; Sources for the third-party DNA databases: M. O’Brien’s ‘Who has the largest DNA database? (2023)’ [www.dataminingdna.com/who-has-the-largest-dna-database/], the International Society of Genetic Genealogy Wiki [https://isogg.org/wiki/] and the respective company websites. [last accessed 3 November 2023].

Table 2

Short tandem repeat (STR) vs. single nucleotide polymorphism (SNP) profile data.

	STR	SNP
No. of markers	13–23	more than 600,000 ^a
DNA detected	2–6 base pair repeats	single nucleotide changes
Coverage	mainly non-coding regions	entire human genome ^a
Relatives detected	mainly, siblings, parents, aunts/uncles	up to 5th cousin and beyond
Data derived	mainly identification markers	identification markers, plus health, genetic predisposition and other trait markers ^a
Law enforcement activity	directly performed	usually outsourced
Database where stored	law enforcement DNA databases	third-party DNA databases

^a QIAGEN recently released *ForenSeq Kintelligence* (targeted amplicon sequencing) using only 10,230 SNP markers, none of which are known health-related SNPs [89].

Table 3

Estimated amount of shared DNA given some biological relationships.

Relationship with the sample	Percentage	cM (average)
Parent, child	50%	2376–3720 (3485)
Sibling	50%	1613–3488 (2613)
Grandparent/grandchild	25%	984–2462 (1754)
Uncle/aunt, niece/nephew	25%	1201–2282 (1741)
Great grandparent, great grandchild	12.5%	485–1486 (887)
1st cousin	12.5%	396–1397 (866)
2nd cousin	3.125%	41–592 (229)
3rd cousin	0.781%	0–234 (73)
4th cousin	0.195%	0–139 (35)

Sources: B.T. Bettinger’s shared cM Project version 4.0 [thegeneticgenealogist.com/2020/03/27/version-4-0-march-2020-update-to-the-shared-cm-project/]; and the International Society of Genetic Genealogy Wiki [isogg.org/wiki/Autosomal_DNA_statistics]. [last accessed 3 November 2023]

2.3. DNA databases and criminal investigations

Each kind of DNA profile is associated with its own category of DNA database. Whereas STR profiles populate law enforcement DNA databases, SNP profiles populate third-party or commercial databanks.

2.3.1. Law enforcement DNA databases

The unprecedented success of the use of DNA profiling in criminal investigations led to the eventual legislation and creation of forensic DNA databases in national systems [50], starting with the United Kingdom in 1995 [51]. Over the years, various countries have set up their own databases with the intention of providing a speedy means of solving criminal cases where the perpetrator has left DNA traces at the crime scene. Among these is the National DNA Index (NDIS) of the United States, whose CODIS expert system is used by many countries, including the Netherlands and Sweden. These national databases are crucial for cross-border cooperation in combatting cross-border crime and terrorism, as highlighted in the EU Council’s Prüm Decision in 2008 [52]. According to the latest Global DNA Profiling Survey of the INTERPOL, 70 among the 89 countries that use DNA profiling in criminal investigations have their own national forensic DNA databases [53]. In Europe, the UK has the largest databank. It comprises more than 7 million individual profiles, whereas the US has more than 20 million profiles (Table 1). In 2009, the US Federal Bureau of Investigation (FBI) improved CODIS to make it functional for the automatic exchange of data as mandated by the Prüm Directives [54–56].

Countries differ in terms of the kinds of individuals included in their DNA databases, which are then named according to the profiles they retain. Among these are offender databases, those of missing and

unidentified persons, as well as reference and elimination databases. Offender databases usually contain STR profile data of convicted individuals. There is no consensus as to whether arrestees' DNA samples should be stored in offender databases prior to conviction. A landmark case in the United States, *Maryland v. King*, with a close 5-4 voting at the Supreme Court, considered the taking of DNA samples from arrestees as part of the ordinary booking procedure in a criminal investigation, similar to taking fingerprints [57]. The European Court of Human Rights appears to require conviction as a prerequisite prior to long-term retention of STR profile data in national forensic DNA databases in its landmark decision, *S. and Marper vs. the United Kingdom* [58], although in the Netherlands, DNA profiles may be collected from suspects and stored temporarily while awaiting the final judgment of the court in a criminal case. In a later case, *Gaughran vs. the United Kingdom*, ECtHR ruled that mere conviction does not necessarily allow indiscriminate retention of DNA profiles [59].

Offender databases have two key contributions to criminal investigations. One, in the absence of suspects, STR profile data from crime scene samples may be scanned and compared with those stored in these databases. Whether or not one agrees with the theory of recidivism—that past offenders tend to recommit crimes [60]—offender databases are useful in informing whether DNA at crime scenes are associated with other crimes. Two, they can be used for familial DNA searching where investigation leads may surface through the subject's close relatives whose STR profile data are present in the database.

Databases of missing persons and unidentified human remains are usually maintained, if there is a suspected crime involved, like in the Netherlands. A couple of years ago, INTERPOL launched 'I-Familia', a global DNA database to help identify missing persons [61]. Reference or population databases, on the other hand, are used for the calculation of allelic frequencies and the consequent determination of their statistical significance in a given population. Finally, elimination databases contain STR profile data of law enforcement personnel, whose DNA may have been inadvertently transferred to the crime scene samples in the course of their work. These are referred to as forensic DNA elimination databases (EDB) [62].

The extension of database coverage to an entire population for criminal investigation purposes describes a universal forensic DNA database [21]. The comprehensive nature of its coverage inevitably gives rise to questions of privacy, although it has been argued that a universal database is not necessarily privacy intrusive given that only non-coding regions of the DNA are stored in the database—a notion that has been challenged recently [63]. Universal databases also make DNA collection more equal as opposed to current law enforcement DNA databases that tend to skew towards cultural minorities. Although it has been claimed the universal DNA databases may be 'inevitable' [64], it is not clear whether it has been implemented even in countries that have attempted to legislate on it, mainly due to its perceived privacy overreach [65].

2.3.2. Genomic big data and third-party DNA databases

The ability of iFGG to generate faster and more reliable suspect investigational leads is fueled by the accumulation of genomic big data brought about by the increase in consumers of direct-to-consumer genetic testing (DTC-GT) companies. Genomic big data is referred to here as the generation and subsequent retention of genomic data in databases whose analysis can no longer be performed manually but must instead be performed by computers, whether such computers make use of artificial intelligence (AI) or not.

The power of DNA-based searching methods like iFGG generally depends on the size of the corresponding DNA database, and more specifically, to the presence of the relevant target population (the presence of genetic relatives) in the database. These databases are not currently maintained by law enforcement. However, their use by law enforcement came to the fore upon the arrest of the GSK in April 2018 [1,41]. Some of these databases are maintained by direct-to-consumer

genetic testing (DTC-GT) companies such as AncestryDNA 23andMe, MyHeritage and Family Tree DNA (FTDNA), and may be referred to as 'commercial DNA databases'. The database used to track down the GSK, GEDmatch, is not maintained by a DTC-GT but it is an open platform where DNA data from various DTC-GT companies may be uploaded for comparison, and may also be referred to as a 'public genealogy DNA database'. Collectively, all of them may be referred to as 'third-party DNA databases' or 'non-law enforcement DNA databases' given that they are not originally intended for law enforcement use.

DTC-GT companies constantly build up their DNA databases as more consumers participate. Although it does not currently allow law enforcement use of its database, the key player among DTC-GT companies is AncestryDNA. It boasts more than 23 million genomic profiles in its database, which is more than NDIS (Table 1). The lowering cost and greater speed in generating genomic big data—thanks to breakthroughs in DNA sequencing technology—further help increase the size of these databases, and consequently, the power of iFGG.

Absent a major public backlash and a subsequent mistrust in the current system of entrusting one's genomic data to commercial companies, there is no other likely direction but a continued growth in these databases. However, even if a majority of the population decide not to have themselves tested by these DTC-GT companies, it is enough for one genetic relative to allow law enforcement use of their genomic data to potentially identify them. More concretely, it has been claimed that a DNA database only needs to cover two percent (2%) of the target population for it to produce a third cousin match akin to that of the GSK case [41]. It has been predicted that the U.S. database only needs to contain around three million Americans of European descent for someone of that ethnicity to have a 99% probability of a match with a third cousin using this technique [41]. Although this claim may be theoretically true, one has to be careful not to 'equate the probability of a match with the probability of identification' [66]. As discussed in Section 5, law enforcement use of iFGG is more complex and may require 'thousands of hours of research' [66], in the form of background research, triangulation, family-tree building, among others.

3. DNA-based methods used by law enforcement prior to iFGG

Prior to resorting to investigative forensic genetic genealogy (iFGG), law enforcement usually makes use of various DNA-based methods in their effort to identify perpetrators of a given crime. To help differentiate these methods from iFGG and to appreciate the scope of possible law enforcement activity prior to its final use of iFGG, a quick look into these various methods is now in order. These methods are not discussed exhaustively given that they are not the main topic of this article. They are discussed in reference to iFGG.

3.1. STR profile matching

The most common and available method used by law enforcement is what is generally called STR-profile matching. It makes use of STR profile data generated from crime scene samples and compares them to the STR profile of a person of interest or data stored in law enforcement DNA databases.

The initial step in STR profile matching is to look for a direct match between the crime scene sample and samples from suspects (or victims) of the crime under investigation. When there is no available suspect or when a direct match does not result, the crime scene DNA is then searched for matches among the samples stored in the database (Step 1, Fig. 1). Failure to achieve a direct match, and in special situations provided by law, law enforcement may proceed to indirect matching.

3.2. Familial DNA searching (FDS)

Familial DNA searching involves a 'deliberate search' of a forensic DNA database for partial matches between the crime scene sample and

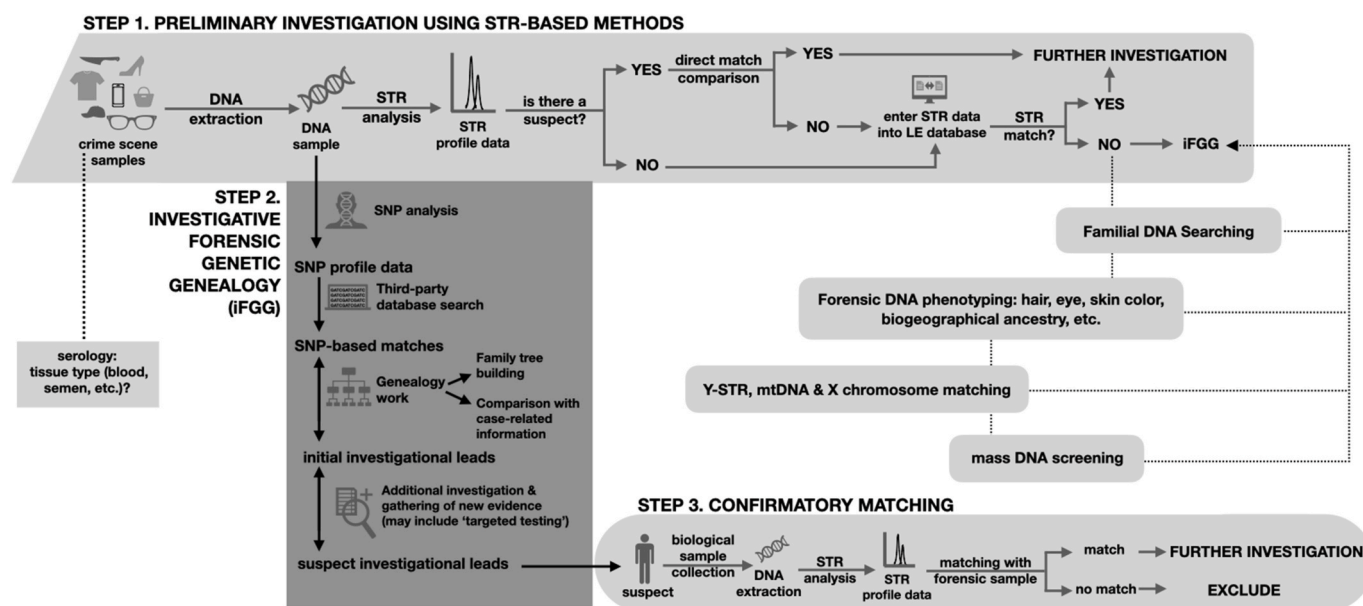


Fig. 1. DNA-based methods used by law enforcement in criminal investigations relevant to investigative forensic genetic genealogy (iFGG). Prior to iFGG, a preliminary investigation using STR-based methods is conducted (Step 1). When relevant, a serology or similar test to assess the type of biological material is carried out. A direct match comparison is first carried out with the STR profile data of suspects, if there are any. STR profile data generated from crime scene samples (also referred to as 'forensic samples') are entered into the law enforcement database. They are subjected to STR matching among the profiles retained in the database should there be no direct match with available suspects. Only when there is a failure to generate relevant STR matches will there be a resort to iFGG (Step 2). On a case-to-case basis, and depending on the relevant legislation, available resources, and capability of law enforcement, other methods are employed prior to final resort to iFGG, such as familial DNA searching (FDS), forensic DNA phenotyping (FDP), and Y-STR, mt-DNA and X chromosome matching; legal and financial considerations may also allow resort to mass DNA screening, which is fed by the same DNA-based methods, mainly by STR and Y-STR matching. The results of iFGG will have to be confirmed through direct matching between the STR profiles of the suspect and that of forensic sample, which should generate a full matching profile; otherwise, it shall be excluded (Step 3).

those present in the database [67]. It is used in countries such as the Netherlands, Sweden, United Kingdom, Australia, New Zealand, and some states in the USA [68,69]. The method also makes use of STR profile data. It is based on the assumption that the more STR-based alleles individuals share, the more related they are to each other, at least biologically. In this regard, the US National Institute of Justice describes FDS as the 'process of running additional CODIS searches in pursuit of biological relatives' [70]. Hence, the necessity of resorting to FDS is only evaluated by law enforcement once initial STR profile matching is not successful.

The resulting search results depend on a pre-determined likelihood ratio threshold value, which may yield few results if it is too high, or many results if it is too low. In any case, the resulting 'candidate list' may include random individuals who may not be related to the criminal perpetrator. Some jurisdictions may allow Y-STR analysis of candidates to reduce the number of adventitious matches. The use of Y-STRs may have the added benefit of detecting full male siblings and all paternal relatives sans mutation.

Using other available evidence at the hands of the police, the list of possible suspects provided by familial searching are then analyzed and vetted. Among other factors, the suspect's age, relationship with the victim, and physical possibility of being present at the crime scene, are evaluated. The most likely perpetrator or perpetrators are subjected to confirmatory matching (Step 3, Fig. 1), which requires a full match with the DNA found at the crime scene—after confirming that the DNA sample was left by the unknown perpetrator—failure of which demands that the suspect be ruled out.

Each legal system has its own set of requirements with respect to when familial searching may be conducted. The additional work required is meant to serve as a legal safeguard to protect the privacy of individuals whose DNA profiles are stored in law enforcement DNA databases. Because of the extra administrative work, privacy concerns and expected lack of success when used, this method is not always

employed by law enforcement (hence, the dash lines in Fig. 1). Thus, familial searching can be, but is not always, part of an investigation involving iFGG.

3.3. Forensic DNA phenotyping (FDP)

In the absence of eyewitnesses to the crime, law enforcement work of sketching out and describing the physical appearance of a perpetrator is extremely challenging. If DNA is left by the criminal perpetrator at the crime scene, however, their external (phenotypic) characteristics may be predicted on the basis of their DNA sequence (genotype) and used to generate investigative leads.

The method of predicting phenotypic characteristics of individuals based on their genetic data is referred to as forensic DNA phenotyping (FDP). Among such characteristics are eye, hair and skin color, which are collectively referred to as externally visible characteristics (EVCs). This method is the basis of the 'Snapshot service' of a company called Parabon NanoLabs [71]. They currently offer this service to law enforcement for criminal investigations, mainly in the United States.

FDP is not yet commonly used by law enforcement due to cost and availability, and in Europe, mainly due to privacy concerns [22]. The results of FDP, whenever employed, are currently used to further narrow down the list of possible suspects, if there are any, or to help point the investigation to a certain direction. It is to be noted that in practice, the analytical results may not turn out as predicted, as when the genotype, for example, says that the perpetrator has blond hair, whereas the actual phenotype turns out to be brown.

3.4. Y-STR, mtDNA and X chromosome searching

Three methods have been recently proposed by Wickenheiser as 'new search key strategies' that use shared DNA among biological relatives to help identify perpetrators who are not enrolled in databases, thereby

theoretically expanding the scope of current forensic DNA databases for the generation of investigational leads [65]. These are Y-STR, mitochondrial (mt) DNA and X chromosome searching.

Y-STR searching is similar to STR profile matching with respect to the use of STRs, only that STRs in this particular case all come from the Y-chromosome. In addition, a Y-STR haplotype is not unique to a specific individual. It provides information about his paternal biogeographical lineage or ancestry. This kind of search is particularly useful when it has been determined that the perpetrator is a male—say in cases of rape, when the perpetrator left behind semen at the crime scene—owing to the fact that only males have Y chromosomes. Absent mutations, Y-STR profiles are transmitted intact along the male line throughout generations. The information that can be derived from it helps narrow down the list of suspects in an investigation [72].

Mitochondrial (mt) DNA is passed on from the maternal line, given that mitochondria are found in the cytoplasm of a cell. During fertilization, an intracellular mechanism of sperm mitochondrial removal has been reported that explains why humans inherit their mitochondria only from their mothers [73]. It was through this scientific fact that it was postulated that all human beings can eventually be traced to one woman called the ‘mitochondrial Eve’ [74]. The method usually consists of sequencing the short mtDNA and comparing its sequence with that generated from the crime scene sample. Whereas Y-STRs provide information on potential paternal lineage, mtDNA provides potential information on the maternal lineage [75].

X chromosomes are present in both males and females. Females have two X chromosomes, whereas males inherit only one coming from their mothers, the other being the Y chromosome (from their fathers). This brief background shows that X chromosomal patterns in males may provide additional information on their maternal lineage. Hence, this information can be an additional searching mechanism to find potential relatives enrolled in the database [76].

These three methods, which reveal either the maternal or paternal lineage of the perpetrator, may provide additional information that can help law enforcement narrow down the list of possible suspects in a crime, whether iFGG is eventually employed or not. Yet, they also entail more cost, time and effort.

3.5. Mass DNA screening

When law enforcement has no suspects and law enforcement database matching is unsuccessful, they may ask people who live near the crime scene, or a certain age group of individuals, to submit DNA samples for comparison. This is called ‘mass DNA screening’. It is not a DNA technique in itself but rather a method of collecting DNA samples from which it earned its other name, ‘DNA dragnet’ [77]. The DNA samples collected are then subjected to DNA profiling.

This method has been criticized for privacy overreach and undue government intrusion upon the lives of the people in the area, which is mitigated by not making the submission of samples mandatory [77]. One may think that the perpetrator will most likely not volunteer to submit a sample. However, such evasion can itself be suspicious, which undermines the voluntary nature of the screening process. At the same time, mass DNA screening has proven to be indirectly useful in solving cold cases as shown in the Pitchfork case, where Sir Alec Jeffreys first applied ‘DNA fingerprinting’ in solving a murder case. The criminal in this case, Pitchfork, asked somebody else to submit DNA on his behalf knowing very well that his DNA would generate a perfect match with that of the crime scene sample. However, he was indiscreet about it while in a public house. The police got wind of this and they turned to Pitchfork himself to provide a sample. A perfect match was detected and the rest was history [32,33]. In the Netherlands, mass DNA screening was applied by the Dutch police in solving the widely-publicized Marianne Vastra [78] and Nicky Verstaappen murder cases [79]. In Sweden, however, mass-DNA screening of more than 6000 individuals did not generate investigational leads, which led law enforcement to try

iFGG [3].

4. Towards a common nomenclature and definition of iFGG

Stephen Busch, a former FBI agent working on iFGG, admitted that the method has gone through ‘several name transitions over the years’ [80], such as genetic genealogy, forensic genealogy, forensic genetic genealogy, investigative genetic genealogy, and more recently, forensic investigative genetic genealogy. As the method continues to gain acceptance worldwide, we believe that it is important to have a common terminology and definition to avoid confusion across borders, and to facilitate both academic exchange among those who do research in this field and its law enforcement use ‘between jurisdictions’ [81]. Further, common terminology will facilitate legal analysis of the technique with respect to, e.g., privacy rights [20]. We analyzed below the various terminologies that have been used to refer to the method vis-à-vis our proposed nomenclature: investigative forensic genetic genealogy (iFGG), which we define as ‘the use by law enforcement of genetic genealogy combined with traditional genealogy to generate suspect investigational leads from forensic samples in criminal investigations’ (Table 4).

4.1. iFGG is more specific than genetic genealogy

One of the earliest articles published reporting the use of a ‘novel forensic approach’ to identify the GSK was authored by Guerrini et al. a few months after his reported capture in April 2018 [10]. It did not name the method but only referred to it in its abstract as ‘police access to genetic genealogy databases’ [10], and in the main text, ‘forensic use of genetic genealogy databases’ [10] without giving it a special name. Later, in a review article in *Nature Genetics* on genomic data sharing,

Table 4
Comparison of relevant terminologies. The definitions provided are not exhaustive. They are only meant to exhibit the differences among the various terminologies. FIGG is not included in the table given that NTVIC used it to combine both FGG and IGG (their definitions are found in the article). IGG and iFGG are synonyms but we argue that iFGG is a more comprehensive and appropriate term to refer to ‘the use by law enforcement of genetic genealogy combined with traditional genealogy to generate suspect investigational leads from forensic samples in criminal investigations’.

Term	Definitions	Examples
Genealogy	the study of tracing an individual’s ancestors and relatives <i>in general</i> ; a.k.a. ‘traditional genealogy’	traditional family tree building using oral histories, birth certificates and baptismal records
Genetic genealogy (GG)	the study of tracing an individual’s ancestors and relatives using genetics <i>in particular</i>	family tree building using DNA data for the determination of an individual’s genetic heritage
Forensic genealogy (FG)	the use of traditional genealogy in forensics	‘disputed inheritance, identification of military personal and citizenship claims’ [15]
Forensic genetic genealogy (FGG)	the use of genetic genealogy combined with traditional genealogy <i>in forensics in general</i>	identification of unidentified human remains outside criminal investigations
Investigative genetic genealogy (IGG)	the use of genetic genealogy combined with traditional genealogy <i>in forensics, specifically in criminal investigations</i>	identification of criminal perpetrators and unidentified human remains within criminal investigations
Investigative forensic genetic genealogy (iFGG)	a synonym for IGG but it provides a complete picture of the technique as the use of FGG in criminal investigations, that is, investigative FGG	identification of criminal perpetrators and unidentified human remains within criminal investigations

Bonomi et al. referred to the technique used to capture the GSK as long-range familial search [82], which may also be attributed to Y-STR and mtDNA searching for their capacity to find familial matches over generations. We avoid using ‘long-range familial search’ in referring to iFGG as it may be confounded with familial DNA searching using STRs and Y-STRs, mtDNA and X-chromosome haplotypes. In the same vein, Greytak et al. argued that ‘the use of the phrase “long-range familial searching” unnecessarily conflates genetic genealogy with familial searching of law enforcement databases when the two methods are fundamentally different’ [66], although one may argue that they instead form part of a continuum.

Bonomi et al.’s article also referred to ‘genetic genealogy’. However, the term ‘genetic genealogy’ is too broad to refer to the method. It is inadequate in the sense that it can refer to the determination of genetic relatedness via genealogy without any particular reference to the involvement of law enforcement. Kennett, for example, defined ‘genetic genealogy’ as ‘the term used to describe the combination of genealogical research with DNA records to form conclusions about relationships’ [40]. It is the proper term that refers to what Greytak et al. described as having ‘enabled thousands of individuals who have lost their biological identity through adoption, abandonment, anonymous gamete donation, misattributed parentage, etc., to regain their genetic heritage’ [83]. The same authors distinguished that method from its use in ‘identify[ing] DNA from suspected perpetrators’ [83], which we refer to specifically as iFGG (Table 4).

4.2. iFGG is not only forensic genealogy

Forensic genealogy was the term used in one of the most relevant articles reporting its use in the identification of the GSK [1]. Wick-enheiser described the method as ‘forensic genealogical searching’ whereby ‘the DNA profile found at the scene of the crime is searched against DNA profiles from individuals known not to be the perpetrator for genetic similarities consistent with originating from a related family member’ [1]. Later, the same author would refer to it as investigative genetic genealogy (IGG) [65].

Forensic genealogy is inadequate in referring to iFGG as it may only mean, *sensu stricto*, the forensic use of traditional genealogy, which is the use of ‘documentary records and oral histories to trace families backwards in time’ [83]. In that regard, Glynn warned that the method should ‘not be confused with Forensic Genealogy, which has long been in existence and typically uses non-DNA genealogical methods, albeit DNA evidence can sometimes be used to confirm conclusions’ [67]. The word ‘genetic’ is crucial because it is what differentiates it from traditional genealogy, given its exploitation of the *dividual* nature of the genetic material, the DNA, in tracing genetic relatedness. Hence, it is an essential part of the method, not a mere option. In this regard, the U.S. NIJ’s Forensic Technology Center of Excellence simply described the method as ‘a technique that combines traditional genealogy research with DNA analysis’ [70].

As to cases that pertain to these methods, forensic genealogy is used more for non-criminal cases like ‘estate and probate cases to identify/find heirs, the identification of living descendants of fallen soldiers for their repatriation, and other historical investigations’ [67], whereas iFGG is specifically aimed at producing suspect investigational leads in criminal investigations. In this regard, Kling et al. noted that forensic genealogy ‘has a distinct meaning in US genealogical circles and relates to all questions of a legal nature that require genealogical analyses, including disputed inheritance, identification of military personal and citizenship claims’ [15]. In either case, both genetic genealogy and forensic genealogy may still be used to refer to iFGG but they have to be combined with other phrases to make them complete, such as ‘genetic genealogy for forensic investigations’, ‘forensic genealogy for investigations using genetic databases’ or ‘law enforcement use of genetic genealogy’. However, they do not capture the technique as a nomenclature but only as a description, as iFGG does (Table 4).

4.3. iFGG is a proper subset of FGG

The most common nomenclatures in academic literature referring to the method are ‘forensic genetic genealogy’ (FGG) and ‘investigative genetic genealogy’ (IGG). Both terms are sometimes used interchangeably within the same article [4,15,65,67], although some articles exclusively use only either FGG [13] or IGG [14,84]. We will tackle FGG in this subsection and IGG in the next one.

In her review of FGG, Glynn identified the two predominant ways in which the method is used, that is, ‘in investigations to identify perpetrators of violent crimes’ and in ‘identify[ing] unidentified human remains (UHRs)’ [67]. Ertürk et al. also described it similarly as ‘a new approach to solving violent crimes and identifying human remains’ [12]. These descriptions underline the broad scope of FGG, that is, it is the proper term to be used whenever genetic genealogy databases are used for the purpose of identifying an unknown sample, including its use by law enforcement to identify unknown criminal victims and to identify unknown decedents whose death is not attributed to a crime.

The U.S. NIJ’s Forensic Technology Center of Excellence uses the term FGG for the method’s use in all cases where traditional genealogy research is combined with DNA analysis. However, we prefer to highlight its investigative use by law enforcement in solving crimes not only because it has brought FGG to prominence starting with the GSK case, but also because it can have very specific implications on the right to privacy. The latter is precisely the subject of the task given to the Biometrics and Forensics Ethics Group (BFEG) in the UK, that is, ‘to consider the feasibility of the use of genetic genealogy resources for the identification of suspects in criminal cases in the UK’ [20]. Among its findings include the need to clearly establish whether it is in keeping with Article 8 of the European Convention of Human Rights (ECHR) or the right to respect for private life. Given that FGG’s investigative use is just one aspect of the method, it is technically its proper subset (Fig. 2), hence, we propose that we specifically name it investigative FGG (iFGG).

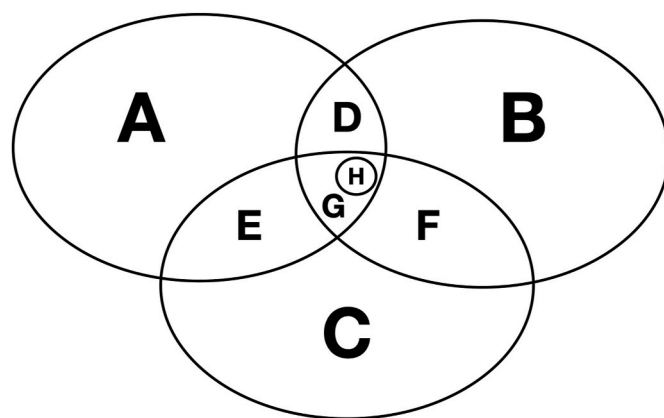


Fig. 2. A Venn Diagram illustrating the relationships among various fields related to forensic genetic genealogy (iFGG). It shows that iFGG is a proper subset of FGG [iFGG $\subset$ FGG]. It also illustrates why it is better to use iFGG over IGG.

LEGEND

- A: Genetics
- B: Genealogy
- C: Forensics
- D: Genetic genealogy
- E: Forensic genetics
- F: Forensic genealogy
- G: Forensic genetic genealogy
- H: Investigative forensic genetic genealogy; also investigative genetic genealogy.

4.4. iFGG, not just IGG

Compared to FGG, we find IGG to be a more accurate term for the technique. Although it was mentioned in the previous subsection that FGG and IGG are sometimes used interchangeably in academic literature, the preferred term appears to be IGG when the report specifically pertains to the identification of a criminal suspect, as when its first use in Europe was reported [3].

In the same vein, the Scientific Working Group on DNA Analysis Methods (SWGDM) also prefers using IGG, although it acknowledges other terms currently being used to refer to the same method, such as ‘forensic genealogy, forensic genetic genealogy, forensic genetic genealogical DNA analysis and searching, genetic genealogy and investigative genealogy’ [16]. Another professional organization, the International Society of Genetic Genealogy (ISOGG) also prefers using IGG although it acknowledges that it is sometimes known as FGG [85].

What is of interest in the nomenclature is the word ‘investigation’, which highlights its use by law enforcement in solving crimes (criminal investigations). Although the same word may also loosely refer to scientific research—for that matter, scientific investigation—we have not encountered any article on IGG using it in that manner. It always refers to law enforcement investigation, specifically in solving crimes.

At the same time, we argue that the word ‘forensic’ should be kept in its nomenclature to highlight its dual scientific and legal orientation. When used as an adjective, as in this case, ‘forensic’ is defined by Merriam-Webster as ‘relating to or dealing with the application of scientific knowledge to legal problems’ [86]. That definition captures what the entire method is about in a general sense. It applies scientific knowledge—specifically, in genetics and genealogy—in solving a legal problem, that is, the identification of suspect in a crime. Hence, although IGG may already capture the method under study better than FGG, it will be more accurate to keep the word ‘forensic’ in the nomenclature, hence, iFGG (Table 4).

Although the focus of this article is on the law enforcement use of genetic genealogy databases for criminal investigations, it would be worth mentioning, albeit briefly, the proper terminology for plain recreational use of genetic genealogy databases to identify one’s ancestry. Given the definition of ‘forensic’ we provided above, it cannot properly be put under the category of FGG, but may be simply referred to as recreational genetic genealogy (RGG).

4.5. FIGG and iFGG

We also consider forensic investigative genetic genealogy (FIGG) as a favorable alternative nomenclature given that it contains the two essential words—forensic and investigative—one of which is missing in either FGG or IGG. FIGG has been previously considered as a synonym for both FGG and IGG, although without an explanation for the use of the term [87]. In February and October 2023, the National Technology Validation and Implementation Collaborative (NTVIC)—a professional organization based in the United States that sets and shares ‘minimum standards and best practices’—gave FIGG a different twist by providing a separate definition for FGG and IGG and then put them together into one [18,19].

NTVIC considered FGG as ‘the process of developing a DNA SNP profile to be specifically uploaded into a genealogical database’, whereas IGG as ‘the investigative component of FIGG, that includes the upload of a SNP profile into a genealogical database, the creation of a family tree, and the investigation of leads’ [19]. They considered both FGG and IGG as ‘two components’ of FIGG [18], which they define as ‘a procedure that combines genetic testing with traditional genealogical research to generate leads in investigations of unsolved violent crimes and unidentified human remains’ [19]. We note that NTVIC’s definition is an attempt to provide a measurable system for purposes of collaboration across the United States. Their new classification of FGG and IGG as two components of FIGG may serve this technical purpose.

On the other hand, as noted in the previous subsection, we agree to the use of the term FGG by the U.S. National Institute of Justice’s Forensic Technology Center of Excellence [70], but more in a general sense, that is, the technique’s use outside criminal investigations, say, identification of missing persons or proving inheritance in an estate proceeding. We propose that its application to criminal investigations by law enforcement be defined as the investigative use of FGG, that is, ‘investigative FGG’, or ‘iFGG’ for short. The word ‘investigative’ functions as a qualifier or an adjective to the term FGG. In this regard, we use the term investigative FGG (iFGG) when the technique is used for that specific purpose of solving crimes by law enforcement, thereby making it a proper subset of FGG (Fig. 2).

The creation of a distinct nomenclature when the technique is used by law enforcement facilitates the legal analysis of its acceptability from the point of view of various legal regimes. For example, although the technique has been used successfully by law enforcement in identifying the culprit of a double-murder case in Sweden, the government put it on hold until it is satisfied that it is legally acceptable within its data protection and privacy laws [4]. It then shows that the technique’s specific use in identifying criminal perpetrators—as iFGG—has its own legal repercussions demanding its own legal assessment, whether or not the same may apply to the technique’s use in identifying other unknown individuals—as FGG.

5. Solving criminal cases using iFGG

After clarifying the concept and definition of iFGG, we now discuss the scope of the method and its expected result. Understanding the latter is crucial in future studies involving iFGG—say, its implications on the right to genetic privacy—as it delineates its coverage and therefore its legal implications. As Guerrini et al. noted, some of the misconceptions surrounding iFGG are not actually specific to it, such as surreptitious collection of DNA samples, which has been in practice even before iFGG [14].

5.1. Preliminary investigation

The initial step in the entire process is the gathering of crime scene samples with biological traces, from which DNA can be extracted (Step 1, Fig. 1). In some jurisdictions and depending on the case at hand, serology testing is conducted to assess the fluid type [88]. For example, in rape cases, it is used to assess if the biological sample is semen from the perpetrator. DNA is then extracted from the biological sample, and is subjected to STR analysis. The resulting STR profile data is compared with those of known suspects, if any.

The procedure quickly ends in the case of a direct match. Law enforcement will then conduct further investigation as to whether the suspect may now be brought to court taking into account the other non-DNA evidence within their reach or custody. If there is no known suspect or a no-match, STR profile data from the forensic sample is entered into the database and subjected to a search. In the event of a match, law enforcement conducts further investigation. It is only when there is no full or partial (familial) match that law enforcement might attempt iFGG. There are other DNA-based procedures that law enforcement may resort to (see methods pointed to with dotted lines in Fig. 1) depending on their expertise and financial capacity, within the limits set by law.

5.2. iFGG proper

The previous step shows that a lot of work is conducted by law enforcement before it finally decides to resort to iFGG. In this regard, it may be considered as the current method of last resort after all the other DNA-based methods have failed to produce investigative leads. It mainly consists of the following steps (Step 2, Fig. 1): 1. SNP analysis; 2. Third-party database search; 3. Genealogy work; and 4. Gathering of additional investigation to narrow down the results of genealogy work,

including targeted testing whenever applicable. The end product is a suspect investigational lead, not a confirmed perpetrator *per se*.

5.2.1. SNP analysis

iFGG starts with a DNA sample generated from the crime scene samples. This may be the same sample generated from the forensic samples for STR analysis, although this time it is subjected to extensive SNP analysis. Currently, most law enforcement agencies do not have in-house facilities to subject DNA samples to SNP analysis. It is usually outsourced to an external laboratory that has been properly accredited [18].

The resulting SNP profile data is analyzed and formatted—into what are called SNP datasets—according to the requirements of the database that will be used for searching purposes. The SNP datasets include information concerning the SNP identification number, the chromosome where it is found, its position in reference to the reference genome, and the genotype [4]. Usually, more than half a million SNP positions are used for comparison during the search process. More recently, QIAGEN released *ForenSeq Kintelligence* which uses targeted amplicon sequencing using only 10,230 SNP markers and which avoids medically-related information [89].

5.2.2. Third-party database search

A third-party database search refers to the comparison of SNP profile data generated from the forensic sample with those retained in third-party DNA databases that are open for law enforcement use. For SWGDAM, the method only begins with this step [16]. However, we have shown in section 5.2.1 that it begins with the generation of SNP profile data from a forensic DNA sample (Step 2, Fig. 1). These are then compared with SNP profile data stored in commercial DNA databases. Since law enforcement database searches involve a different type of DNA data—that is, STR profile data instead of SNP profile data—that initial step is necessary before undergoing a third-party DNA database search under iFGG.

Depending on the capability of law enforcement, they themselves can conduct this search, or if supported by appropriate legislation, it may be contracted to a commercial laboratory or a genetic genealogist [4]. Not all private companies allow law enforcement to use their DNA databases in criminal investigations. To date, only three of them explicitly allow their use for that purpose, namely, GEDmatch PRO, FamilyTreeDNA (FTDNA), and a more recent non-profit undertaking called DNA Justice [90]. A private initiative, DNASolves, is an internal database used by its parent company, Othram, Inc., to search for genetic relatives on behalf of law enforcement [67].

The end-product of a third-party DNA database search is a list of potential genetic relatives whose closeness to the forensic sample depends on the amount of SNP data they share, as expressed in centimorgans (cM). This ‘genetic association’ process is based on the assumption that the more DNA data they share, the higher cM values are generated between them [17].

5.2.3. Genealogy work

The list of putative genetic relatives generated in the previous step is vetted by law enforcement with either an in-house or a contracted genealogist to narrow it down to one or a few suspects per SNP profile at hand. From the various SNP-based matches obtained, a family tree is built based on the amount of SNP data they share from which relationships with respect to the forensic sample may be imputed (Column 1 in relation to Column 3, Table 3). One popular tool used to generate family trees using SNP-profile data is ‘The Shared CM Project’, a ‘collaborative citizen scientist’ initiative spearheaded by B. Bettinger [91,92].

Together with publicly available data such as civil and church records, the family tree is built back up to the Most Recent Common Ancestor (MRCA) between the matching individuals and the one generated from the forensic sample, and then built back down to the

present time to include descendants of the MRCA [65]. The family tree is then examined for crossing or overlap of maternal and paternal family lines coinciding within a single family unit. The latter’s progenies are scrutinized for possible suspects that match crime particulars, such as sex, age, possible geographical location during the commission of the crime, past criminal records, circumstances that may connect them to the crime, and other information. Family tree building is usually limited to third cousins, given that an average individual has ‘1000 fourth cousins and 5000 fifth cousins’ [4].

To further refine the data obtained in the foregoing, additional work is performed on the database list vis-à-vis the putative family tree. One such activity is referred to as ‘triangulation’, where intersections among them are searched to discover possible genetic relationships [4].

5.2.4. Additional investigation

The initial investigative lead generated from genealogy work is normally sufficient should it have generated a single or a few suspects. However, additional investigation may be necessary in cases when, at this point in the iFGG process, there are still several possible suspects without a clear direction as to who among them may be subjected to confirmatory testing. Compared to the previous step where the building of a family tree mainly depended on SNP-generated matches and evidence that were within reach of law enforcement, this additional step involves gathering additional evidence to further narrow down the list of investigational leads.

In the event of an impasse in the genealogical search and family-tree building process, a strategy known as ‘third-party, target/targeted or reference testing’ may be performed by law enforcement. This includes asking possible relatives from the specific area of interest to take a test in aid of the investigation. Each jurisdiction has its own rules on target testing. The main purpose of target testing is to eliminate some branches of the family tree and to include others, thereby facilitating the family-tree building process. Target testing serves to deny or confirm the genetic relationships of the people within the *theoretical* family tree and try to get as close as possible to the genetic profile found in the forensic sample, thereby further narrowing down the ‘search pool’ [15]. Target testing may also be performed when there are issues with large number of relatives. Testing some critical living relatives may help knock out entire branches of the family tree, thereby assisting the family tree building process. The entire iFGG process ends with the production of a suspect investigational lead.

5.2.5. Confirmatory test

Although it is not part of iFGG, confirmatory testing is discussed since it is a necessary step in the whole criminal investigation process (Step 3, Fig. 1). Once a suspect is identified via iFGG and relevant police work, confirmatory testing must be done using STR profile direct matching. The procedure is similar to the initial steps of preliminary investigation, only that the biological sample is now collected from a suspect identified from iFGG. Confirmatory matching requires a direct match between the suspect’s and forensic sample’s STR profile data, failure of which means exclusion of the suspect from further investigation [17].

It is possible to end up with more than one lead, as in the double murder case in Linköping, Sweden. Using iFGG, two brothers matched all the criteria within the family tree built for that purpose, and upon confirmatory testing, one of them matched that of the forensic sample. The latter confessed and was later convicted for the double murder [3]. However, a direct match does not necessarily mean a guilty verdict. Law enforcement must present additional evidence outside STR profile match data, failure of which the case should not proceed to court.

As a final note, we emphasize that in contemporary law enforcement practice, iFGG data are currently not presented as evidence in criminal courts given their limited role of producing suspect investigational leads. It is the results of confirmatory matching using STR profile match data that are used for prosecution [83]. The purpose of iFGG is only to

generate suspect investigational leads and not to confirm the perpetrator's identity [17]. In spite of all the various steps involved in the process, iFGG is not meant to and cannot provide 100 percent certainty on the identity of the perpetrator [83].

6. Conclusion

We present investigative forensic genetic genealogy (iFGG) as the most appropriate nomenclature to refer to the law enforcement method of generating suspect investigational leads from forensic sample DNA using genetic genealogy databases. It is a proper subset of forensic genetic genealogy, that is, FGG as applied by law enforcement in criminal investigations; hence, we name it as 'investigative FGG' or iFGG. It is used by law enforcement only after other DNA-based methods, such as STR profile matching—and in some jurisdictions, familial DNA searching, Y-STR, mtDNA and X-chromosome searching, forensic DNA phenotyping for external visible characteristics and biogeographical ancestry, and mass DNA testing—failed to produce a suspect. Hence, it is currently the method of last resort in solving criminal cases.

As to the steps involved in iFGG (Fig. 1), it only begins with the generation of an SNP profile and ends with the production of suspect investigational leads (Step 2). It does not cover crime scene sample collection (Step 1) as it is not currently the first method of choice in a criminal investigation. It also does not cover the confirmatory step of matching the suspect's DNA sample with that of the forensic sample (Step 3), the results of which may be presented in criminal courts. The results of iFGG as a method are not currently introduced in court as evidence. We proffer this nomenclature and clarified the steps surrounding it for future studies evaluating iFGG's legal implications on the right to genetic privacy.

CRediT authorship contribution statement

Oliver M. Tuazon: Writing – review & editing, Writing – original draft, Visualization, Resources, Methodology, Conceptualization. **Ray A. Wickenheiser:** Writing – review & editing, Resources, Methodology. **Ricky Ansell:** Writing – review & editing, Resources, Methodology. **Christi J. Guerrini:** Writing – review & editing, Resources, Methodology. **Gerrit-Jan Zwenne:** Writing – review & editing, Supervision, Resources, Methodology. **Bart Custers:** Writing – review & editing, Supervision, Resources, Methodology.

Declaration of competing interest

The following authors declare the following financial interests/personal relationships which may be considered as potential competing interests: **CJG and RAW** are members of the Investigative Genetic Genealogy Working Group of the Scientific Working Group on DNA Analysis Methods (SWGDM); **RAW** is a member of the National Technology Validation Implementation Collaborative (NTVIC) Steering Committee and also the Chair of its Forensic Investigative Genetic Genealogy Policy and Procedures Committee; **CJG** is a member of the NTVIC Forensic Investigative Genetic Genealogy Policy and Procedures Committee. The other authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Oliver M. Tuazon, MS, JD, LLM [o.m.tuazon@law.leidenuniv.nl], is a lawyer and holds master's degrees in Microbiology and in Forensics, Criminology and Law; he is currently a PhD candidate at the Center for Law and Digital Technologies (eLaw) of Leiden University, where he works on the intersection between science and law.

Ray A. Wickenheiser, DPS, MBA [RAY.WICKENHEISER@troopers.ny.gov], is the Director of the New York State Crime Laboratory System, former President of the American Society of Crime Laboratory Directors, and Chairman of the Policy and Procedure Committee of the National Technology Validation and Implementation Collaborative (NTVIC), USA.

Ricky Ansell [ricky.ansell@polisen.se], Senior Forensic Advisor and Adjunct Associate Professor at the Biology Unit of the Swedish National Forensic Centre, Linköping, Sweden, and the Department of Physics, Chemistry and Biology (IFM), Linköping University, Linköping, Sweden.

Christi J. Guerrini, JD, MPH [guerrini@bcm.edu], is an Assistant Professor in the Center for Medical Ethics and Health Policy at Baylor College of Medicine.

Gerrit-Jan Zwenne, LLM, PhD [g.j.zwenne@law.leidenuniv.nl], is a full professor of privacy and data protection law at Leiden University and the Open University; he consults for the Dutch government as a lawyer and partner at the Hague-based Pels Rijken law firm where he handles privacy and data protection, health care, artificial intelligence and cybersecurity cases, among others.

Bart Custers, MS, LLM, PhD [b.h.m.custers@law.leidenuniv.nl], is a full professor of Law and Data Science and head of the Center for Law and Digital Technologies (eLaw) of Leiden University.