http://doi.org/10.53587/25792865-2024.11-41

THE LATEST PARADIGM SHIFT IN FORENSIC GENETICS - MASSIVELY PARALLEL SEQUENCING AND LARGE VOLUMES OF DATA

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Over the last several decades forensic genetics has evolved to provide extremely sensitive and highly resolving results for human identification in criminal cases and humanitarian efforts. The advent of DNA databases increased the power of forensic DNA typing even further by rapidly developing investigative leads. Even with the current methods, markers and databases, hundreds of thousands to millions of criminal, civil and humanitarian cases have not been resolved even though biological evidence is available. To address this deficiency the forensic genetics community is poised for a paradigm shift fuelled by the advent of massively parallel sequencing and a reinvigoration of genetic markers known as single nucleotide polymorphisms. Two examples – forensic genetic genealogy for human identification and virome analyses for geolocation – are discussed to illustrate the power and future of forensic genetics or more appropriately termed forensic genomics.

Keywords*: forensic genetics, human identification, massively parallel sequencing, single nucleotide polymorphisms, forensic genetic genealogy, virome, geolocation*

History and State-of-the Art

The use of DNA derived from crime scene biological evidence and unidentified human remains is a forensic science success story. With roots grounded in basic and applied science and years of collaboration among scientists worldwide, capabilities and applications have been developed and implemented providing a wide range of analyses and remarkable sensitivity of

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detection. Four decades ago, the discipline of forensic DNA typing was born. It was ushered in by the seminal work of Alec Jeffreys who used a restriction fragment length polymorphism (RFLP) and multilocus probe methodology to link the perpetrator of two separate cases in which two teenage girls in Leicester, England were raped and murdered [1,2]. These two cases hallmarked the first time DNA was used for source attribution in a criminal investigation. This pioneering work demonstrated that DNA from crime scene evidence could be analyzed, the accessible genetic variation offered high discrimination power, DNA was a stable target, and DNA could be detected in a variety of body tissues, such as blood, semen, and saliva which are commonly found in sexual assault cases. DNA markers, originally variable number of tandem repeats (VNTRs) or minisatellites, far exceeded the value of protein markers, such as the ABO blood group, phosphoglucomutase-1, and group-specific component, that were commonly used up until that time [3].

Although groundbreaking and a fundamental leap forward in the analysis of biological evidence for source attribution, the RFLP method was relatively short lived. The method required substantial amounts of template DNA (10-50 ng) and relatively intact DNA (fragments of several thousand to >10,000 bases long) to obtain a result [4]. Neither of these requirements for successful RFLP-VNTR analyses could be met routinely for the wide range of biological evidence found at crime scenes. Additionally, the methodology was laborious and time consuming, taking up to several weeks to generate a profile of several VNTR loci. By the early to mid-1990s the RFLP methodology was replaced by polymerase chain reaction (PCR)-based procedures culminating with worldwide adoption of short tandem repeat (STR), or microsatellite, loci as the primary markers for human identification [5,6]. PCR-based methods enable analysis of trace quantities of DNA (<1 ng of template), amounts that are more commonly found in forensic evidence. STRs had several features that made them the markers of choice for human identification. They were relatively small-sized markers (amplicons between ~100-400 base pairs in length) and thus were amenable to amplification by the PCR. Moreover, the shorter fragments permitted analysis of somewhat degraded DNA. Like VNTRs, STRs also were highly polymorphic with heterozygosities >0.70 and when multiplexed provided exceedingly high powers of discrimination, such that a full single source DNA profile (of 13-~20 markers) could reduce the population of potential contributors of biological evidence to a few, if not one individual.

The forensic DNA field continued to mature based on autosomal STRs as the primary markers for analyses and data exchange. Core STRs were defined to enable exchange of DNA results among laboratories (including international agreements [7,8]), commercial kits were developed to facilitate core STR marker typing, capillary electrophoresis (CE) for separation and detection of STR amplicons provided a semi-automated technology greatly reducing labor and democratizing the technology, validation studies were performed, substantial peer-reviewed publications were generated to support reliability and guidance on how to effectively use and interpret the genetic data (especially mixture evidence), sound population genetics

and statistical analyses were established to convey the weight of evidence, and national DNA databases were implemented to solve future crime and identify missing persons and human remains [9]. Today, in laboratories worldwide, human identification applications employ commercial kits containing reagents to target and amplify between 20-30 autosomal STR markers for analysis of evidence derived from crime scenes, mass disasters, parentage testing, and missing persons and human trafficking cases.

For most criminal cases STR profiles from evidence and persons of interest are compared directly in a one-to-one manner to assess if a particular individual can be included or excluded as a potential contributor. Identification of human remains can be compared in a similar manner as crime scene evidence if an antemortem sample is available. However, chain of custody issues have a degree of uncertainty regarding the source of antemortem samples, particularly for personal items, such as toothbrushes, hairbrushes, and razors. Therefore, most associations by DNA testing of human remains cases involve indirect (or kinship) comparisons of DNA profiles from the remains with a DNA profile from typically a putative first-degree family member(s). The indirect comparison is a viable means for identification of human remains because family members share more DNA in common than do unrelated individuals. The majority of human identification testing in criminal cases is done by direct comparison of STR profiles; it is relatively infrequent that indirect comparisons by STR typing have been used in criminal casework. In contrast, most humanitarian efforts to identify human remains and civil human identification testing (predominately paternity testing) primarily perform kinship testing relying on STR markers.

There are some analyses in which additional genetic marker systems are employed. These systems are Y chromosome STRs (Y-STRs) and mitochondrial DNA. Both systems are considered lineage markers, paternal and maternal, respectively, and, barring mutation, are inherited intact from one generation to the next [10-14]. Thus, they can extend kinship analyses beyond first-degree relatives, as long as an appropriate lineage reference sample is obtained for comparison. The Y-STRs also are informative, for example, in cases in which mixtures contain large amounts of female DNA and small amounts of male DNA as may be encountered in sexual assault cases. The male DNA component could be swamped or masked by the greater contribution of the female DNA component and not be detected or only partially detected by autosomal STR typing. However, since females do not carry a Y chromosome, detection of Y-STRs is not impacted by the presence of female DNA. The number of copies of mitochondrial DNA in a cell (approximately 500-1000 copies) is far greater than nuclear DNA (two copies). Thus, mitochondrial DNA testing may provide results in cases in which samples contain little or no detectable nuclear DNA. Mitochondrial DNA is one of the most successfully typed markers in unidentified human remains cases or in criminal cases with samples that contain low quantities of nuclear DNA, such as degraded bones and hair shafts. In some complex kinship cases, such as incest, X chromosome STRs (X-STRs) can be quite informative [15]. Although informative, X-STRs are not extensively used by crime laboratories.

The value of autosomal STR typing to assist in developing investigative leads for both criminal cases and unidentified human remains cases was bolstered by the development of DNA databases. Some criminals are repeat offenders. Upon arrest or conviction, a DNA sample can be taken, and a subsequent DNA profile is generated, which then is uploaded into a database. If any of these individuals commit a future crime and biological evidence is left at the crime scene, the resultant DNA profile can be searched in a database containing reference profiles of convicted and/or arrested individuals and possibly identify potential sources of crime scene evidence rapidly and effectively. Databases exploit direct comparisons of STR profiles but in a one-to-many manner. Similarly, unknown persons can be associated in a one-to many pedigrees manner via database searching to assist in the identification process. The use of STR markers and DNA databases is a criminal justice success story. For example, as of February 2024, the FBI reported that CODIS generated 698,183 hits [16].

Overall, it would seem that PCR-based STR typing and mitochondrial DNA testing by Sanger sequencing both based on CE would be the pinnacle of forensic genetics for source attribution. Yet, there are hundreds of thousands, if not millions, of cases worldwide in which these CE-based methods are insufficient. For samples in which too little DNA is available and/or are highly degraded STRs are inadequate markers. Although STR are amenable to PCR because they reside in short amplicons of sizes ranging from ~100 to 400 base pairs in length, there are a good number of DNA samples that are so degraded that only partial profiles or no STR profiles are generated. The recoverable fragments of DNA are too short to enable amplification of STRs by PCR. In kinship cases the limited number of STRs typically only support first-degree associations, although some first-degree relationships are not detected because of the limited number of STRs typically employed. Lastly, although DNA database searching has been a realized success story, about half the forensic profiles in, for example, CODIS have yet to be associated with a potential source. Thus, STR typing is not assisting in solving those cases and identifying unknown persons which in turn means that victims, families, and communities are not provided resolution, and those perpetrators are free to commit more crimes rendering more people as victims. Furthermore, those unknown persons that have yet to be identified have not been given the respect and dignity they deserve, and their families have not gained resolution.

Massively Parallel Sequencing and SNPs – the latest paradigm shift

Two advances that have occurred over the last 20 years are now sufficiently well-developed and robust for forensic applications providing an increased level of sensitivity of detection and analysis of highly degraded DNA. These two advances are massively parallel sequencing (MPS) and access to large volumes of genetic data per sample. The genetic data are in the form of another class of markers that, in actuality, were employed early on with PCR-based analyses, i.e., single nucleotide polymorphisms (SNPs). A SNP varies at a position in the genome, and each individual harbors several million SNPs (about 1 in every 1000 base

pairs) [17]. Other than sequencing the hypervariable regions of the mitochondrial genome and employing a technique known as minisequencing (also called SNaPshot) [18], the early SNP-based methods (such as HLA-DQA1 and Polymarker [19,20]) were replaced by the STR systems because on a per marker basis STRs are more informative and at the time were considered quantitatively and resolution-wise more effective for interpreting mixtures. Beyond identity testing, SNP testing can provide genetic information about externally visible phenotypes, such as eye and hair color, and biogeographical ancestry [21-23]. But these SNP-based applications have been relegated to secondary roles in developing investigative leads primarily due to their lower discrimination power, context-specific application, and low throughput analyses on a CE platform.

The forensic genetics field now is poised for a paradigm shift that rivals the impacts that PCR, STRs and CE previously had on the discipline. At the beginning of the 21st century, the first draft human genome was reported [24]. This feat was a monumental task that involved over 40 institutions, took over a decade to accomplish, and cost several billion US dollars. However, the human genome project provided much more than a draft map of a single human genome; it established infrastructure that fostered innovation. Shortly after the first reported genome, MPS was introduced. Instead of sequencing one target at a time as is done by low throughput Sanger sequencing, the methodology used for determining the sequence of the first human genome, MPS enables sequencing of thousands to millions of targets simultaneously [25-28]. This high throughput feature was unparalleled substantially reducing the cost and time to sequence DNA such that sequencing multiple forensic targets or an entire human genome now can be accomplished within the operational laboratory and for some cases could be more cost effective than traditional technologies. As an example, the production of genome wide analyses on ancient DNA (aDNA) samples has increased substantially inversely correlated with a reduction in cost per gigabase sequenced (Figure 1).

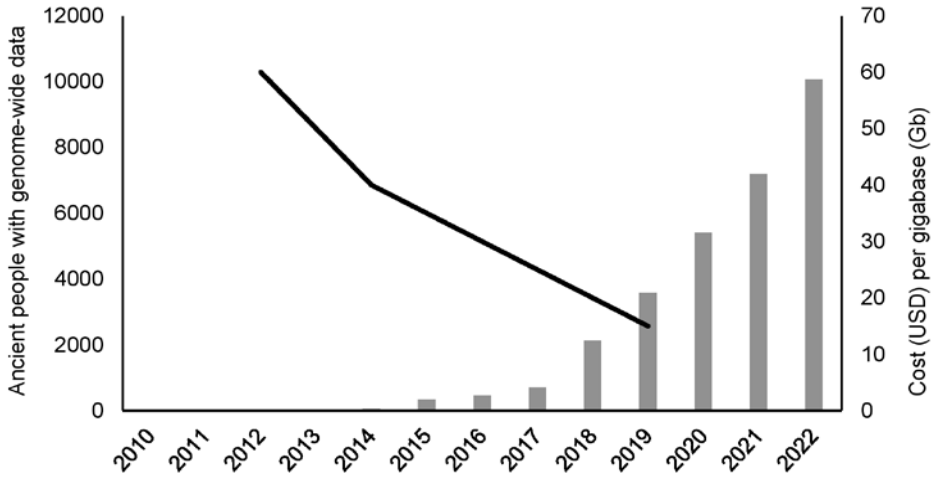

Figure 1. Ancient human genome-wide data and reduction in sequencing cost per recent years. Number of ancient human genome-wide samples was obtained from [29] and cost of sequencing from [30].

aDNA genomes can be considered some of the most difficult human DNA samples due

to high degradation/damage and low quantities, which parallels the needs and is indicative of potentials of success for forensic DNA analyses. But more importantly because of the ability to scan the entire human genome, dense SNP testing in a quantitative manner is now feasible. Any portion of the human genome or the entire genome can be sequenced, providing unprecedented levels of genetic information that can be used for source attribution. With MPS a few thousand to over a million SNPs can be analyzed simultaneously, far exceeding anything imaginable by CE. This throughput and genetic data access have renewed interest in SNPs. Their combined exceedingly high power of discrimination, shorter fragment size compared with STRs, powerful kinship associations, potential for interpretation of mixtures, enhanced bioancestry assessments, and geolocation, to name a few applications make SNPs a versatile marker set that can be informative in a wide variety of forensic casework. Moreover, cases that have yet to be solved with the assistance of STR typing modalities may be reconsidered with the advanced capabilities that MPS and SNPs afford.

Several MPS platforms are available for forensic analyses with the MiSeq and NovaSeq from Illumina (San Diego, CA, USA) and the S5 from ThermoFisher (South San Francisco, CA, USA) being the primary platforms. Their chemistries are different and validation studies are necessary to understand the nuances of each chemistry. The differences are not necessarily limitations impacting the use of various chemistries; it is, however, essential to know through validation studies the limitations of output data for proper interpretation.

The cost has dropped significantly over the past decade especially due to Illumina's chemistry and the NovaSeq system, and it may be cheaper to perform WGS than current DNA typing procedures for some cases and already cost effective compared with targeted SNP testing (screening a few thousand to around 95,000 SNPs [31-33]). Recently, the AVITI from Element Biosciences (San Diego, CA, USA) has come to the market offering high quality, high throughput at more affordable prices and, for example, Ultima Genomics has suggested that its novel platform may reduce the cost of sequencing a human genome another order of magnitude [34], although this achievement has yet to be realized. The continued improvements in technology and reductions in cost will democratize MPS such that its use will not be relegated to large genomics centers or private industry but be a routine tool of the forensic laboratory, much like what was experienced with CE more than 25 years ago.

Sequencing is not a novel methodology to support characterization of biological evidence as mitochondrial DNA has been sequenced by Sanger sequencing for more than 30 years [35]. However, because of its low throughput Sanger sequencing of the mitochondrial genome was limited primarily to the non-coding region (hypervariable regions HV1 and HV2), consumed several aliquots of a DNA extract, was relatively costly (at ~$3000 US dollars per test by some commercial providers) and was quite laborious. Moreover, 75% of mitochondrial genome variation resides in the coding region, the region that was relatively untapped by the forensic community [36]. With the high throughput of MPS, the whole mitochondrial genome can be sequenced, capturing the full genetic variation available, while consuming less sample

and reducing labor and cost. Additionally, MPS is a more quantitative methodology, compared with Sanger sequencing and CE, so mitochondrial DNA analyses can extend beyond single source samples, such as hairs, bones, and teeth, to the wider range of forensic samples, including mixtures. Some forensic laboratories already have embraced the power and robust nature of MPS and its lower cost compared with Sanger sequencing and validated and/or implemented whole mitochondrial genome sequencing for casework analyses [37-40]. Given the choice today of MPS or Sanger sequencing for analysis of mitochondrial DNA from biological evidence, MPS is the viable choice going forward.

MPS Workflow

The typical workflow for generating sequence data is in part similar to current workflows. The foundations already established for DNA extraction are the same. They enable recovery of DNA of sufficient purity for downstream analyses. Extraction procedures that recover short DNA fragments may be worth pursuing as MPS is well suited for analysis of degraded samples. In this regard, procedures used in the aDNA arena are useful [41,42]. Indeed, the Neanderthal genome would not have been deciphered without MPS and short fragment analyses – a feat that was recognized with this past year's Nobel Prize being awarded to Svante Pääbo [43].

Similar issues regarding co-purifying materials also need to be considered. Inhibitors and biological contaminants (e.g., microbial nucleic acids in soil) can impact sensitivity, reliability and typing success. For example, initially microarrays were used to scan the genome for SNPs from forensic evidence [44]. They generated a large amount of data at a relatively low cost. However, analytical success is hampered by the presence of microbial DNA. MPS is less sensitive to the presence of microbial DNA as any sequence data generated that are not human in origin can be filtered out bioinformatically. Furthermore, there are capture methods, that can enrich extracts for DNA of interest, so the analysis of specific DNA markers is not as compromised by competition for sequencing reagents [32].

Methods that assess the quality and quantity of the DNA, typically done by quantitative PCR, convey what downstream analyses are feasible. Kits such as Quantifiler™ Trio DNA Quantification Kit (ThermoFisher Scientific), PowerQuant® System (Promega Corporation), and Investigator Quantiplex Pro Kit (QIAGEN) are examples of suitable kits for PCR-based STR-analyses, as well as whole-genome sequencing (WGS) [45-47]. However, for DNA that is highly degraded, these methods are not indicative of the quantity of template suitable for MPS. Alternate quality and quantity metrics are required, such as using the iSEQ (Illumina), a lower throughput sequencer to assess the DNA before proceeding to WGS.

Either for targeted marker amplification by PCR or WGS, the next step in the workflow is library preparation. Simply, library preparation modifies the DNA by adding onto the ends of template fragments short pieces of known DNA to facilitate sequencing. Simply stated - library preparation is nothing more than getting the sample ready for sequencing. For MPS

generally adapters, short sequences of DNA, are added to each fragment so the DNA can be tethered to a support for subsequent cloning and sequencing. Additional sequences can be added so primers can bind to initiate sequencing-by-synthesis reactions (one of the possible strategies for sequencing) and short unique sequence tags (i.e., barcodes) can be added to every fragment of a sample. With these unique signature tags, multiple samples can be pooled (or multiplexed) further increasing throughput. The genetic data per sample are parsed bioinformatically after sequencing via the unique signature tags (known as demultiplexing).

After sequencing, data analysis and interpretation of results are performed. Because of the massive amount of data that is generated by MPS, bioinformatics (i.e., software tools) is an essential and critical part of the process. Manual interpretation is no longer feasible. So forensic genetics analysts must move from analog to digital data analyses. The flow of data processing generally proceeds from quality assessments of raw data (trimming the sequencing reads, filtering the low-quality data) to comparative analysis (reference-based alignment) or *de novo* assembly. During data analysis, several quality metrics such as quality scores for base calling, alignment, depth of sequencing, strand bias and variant calling are monitored. Base calling is the identification of the specific nucleotide present at each position in a single read while a read is the DNA sequence of a single molecule (or clone of a single molecule). The more reads obtained, the greater is the confidence in a base call (achieved by call consensus). A quality threshold of base scoring is typically set with a Q score. A threshold of Q20 sets the minimum base call accuracy at 99% which permits an incorrect base call per read at 1 in 100; a Q30 score sets accuracy at 99.9% allowing incorrect base calls per read at 1 in 1,000. These low frequency errors are overcome by generating a number of reads for a DNA region(s) per analysis, i.e., the sequencing depth. There are no quality standards set regarding a Q score threshold, but a Q20 score seems to be generally used. However, a Q score less than 20 may still provide reliable data when numbers of reads are analyzed, and the consensus sequence with a majority base is called.

Human Identification and Extended Kinship Associations

With such large volumes of SNPs being assayed, the lower diversity per marker of SNPs is overcome readily and now affords far greater discrimination power than a typical commercial STR kit. Coupled with the fact that large dense SNP data can be generated without consuming more of the DNA than is required for STR typing, MPS becomes quite appealing as a human identification tool. Moreover, studies indicate that MPS offers greater sensitivity of detection and robustness than STR-CE methods [48-52]. The enhanced features offered by MPS enable source attribution of crime scene biological evidence to move from primarily direct comparisons with a person of interest and/or a donor if he/she resides in a national DNA database to exploiting kinship associations to develop investigative leads. If a donor of forensic evidence is not in a database, STR typing is rather limited in generating investigative leads. However, relatives of the donor of the evidence increases the pool of reference samples

substantially. By accessing relatives upwards of 7th degree and beyond, the pool of reference samples to assist in source attribution leads increases significantly and the limitation of STRs and current databases of requiring the donor to be in the database is overcome [30]. This capability also expands the pool of family members many degrees distant that can assist in identifying human remains and resolving problematic parentage cases, readily overcoming traditionally vexing, uninformative cases by STR typing of half sibs and second-degree relatives. The lineage markers (Y-STRs and mitochondrial DNA) that were used, sometimes as critical information, in kinship analyses to expand associations beyond first-degree kinship analyses are not necessarily required to make associations, which reduces the need to specifically access paternal and/or maternal relative reference samples. In some cases, these lineage markers are uninformative, for example, a father-daughter relationship cannot be determined by use of Y-STRs or mitochondrial DNA. But with dense SNP panels, such an association becomes a trivial genetic challenge. Thus, it is reasonable to expect that more leads will be obtained by MPS-SNP analyses.

The combination of MPS-SNP typing on forensic evidence has contributed to a new subdiscipline of forensic genomics known as forensic genetic genealogy (FGG). The resultant dense SNP profiles are searched against a database(s) of dense SNP profiles from volunteers who have consented to use their profiles to support law enforcement investigations. These databases are comprised of dense SNP profiles from volunteers who, in a citizen science manner, have consented to allow law enforcement access to their genetic data to be used to indicate kinship relationships that may lead to the source of crime scene evidence or identification of human remains. The volunteers who contribute their DNA to a database are not persons of interest nor are they victims or suspects; they serve as sources for developing investigative leads. The searches identify potential near and distant relatives of the donor of forensic evidence. Subsequently the associated relatives form focal points for performing genealogical research to home in on candidates of the source of forensic evidence [30,53]. The differences in FGG databases and current government managed STR national DNA databases are the former are managed by private entities, the reference profiles in the database are from volunteers obtained by informed consent, and the genetic data do contain private genetic data, so privacy, security, transparency, good governance, and proportionality assessments are essential [54]. Once associations are made, genealogists access public records to develop pedigrees to find a most likely common ancestor(s) of the evidence and the relative(s) identified by dense SNP searching. As recently as December 2023 318 criminal cases and 461 unidentified human remains cases have been assisted with dense SNP analyses that were uninformative by STR typing [55] which is an underestimate of cases assisted.

The resolution of the Golden State Killer case by FGG is the most known example of the value of this enhancing technology and brought the use of MPS and dense SNP analyses to the forefront of forensic genomics [56]. A high-density SNP profile generated from a crime scene sample was uploaded to the public database GEDMatch [57]. There were several potential

distant relatives' profiles (~third cousins) that associated with the crime scene sample. After these potential relatives were identified by the genetic data analyses, searching of genealogical records anchored on these potential distant relatives was undertaken. Together with other information (such as age, location, etc.), investigators were able to narrow their search to a particular person. Then, the autosomal STR profile(s) derived from the evidence and from the person of interest were generated to confirm the association.

The United States has been the country employing the majority of FGG cases; however, several other countries have made use of it and are establishing guidelines for use [58,59]. Overall, the combination of dense SNP profiles with genealogical data from public databases or commercial genetic genealogy services, investigators are establishing familial relationships that turn into investigative leads in cases where traditional methods have been unsuccessful. That is the concept of identification by kinship association, which was relegated primarily to humanitarian efforts, now is an approach for determining who may be the source of evidence found at crime scenes and has become far more powerful for effective comparisons of reference samples with human remains samples.

Human Microbiome for Developing Investigative Leads and Identification of Humans and Human Remains

Humans are born with approximately 20,500 genes, but shortly after birth carry over 1,000,000 genes [60-62]. This huge increase in the number of genes is due to the acquisition of microorganisms as part of the normal human development and life. Often when one performs WGS on human samples, the focus is on human DNA. Nucleic acids from the microbiome also are extracted and sequenced; however, these molecules are not assayed during human DNA focused procedures because they are filtered out bioinformatically. Yet, they harbor substantial genetic information of forensic value, such as human identification by the individualizing microbiome, determination of postmortem interval by the succession of microbes after death over time, body fluid sourcing, and tracking of sexually transmitted diseases in child molestation, rape cases and questioned malpractice cases in healthcare provider outbreaks [63].

MPS also has been adopted in forensic applications involving microbe detection, predominately for tracing food borne pathogens and sourcing of human viruses (Figure 2).

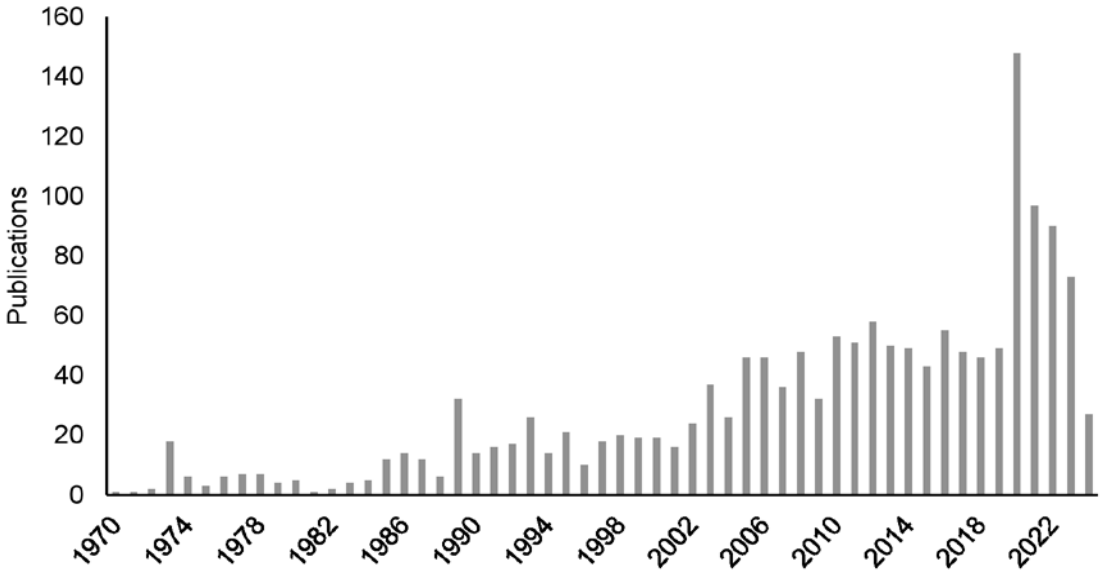

Figure 2. Number of publications on virus genome data generated over recent years. Publications of viruses in forensic science have increased with advances and reduction in cost of massively parallel sequencing (MPS). Virus related publication numbers obtained from Scopus with search terms (title-abstract-keywords (virus) OR title-abstract-keywords (viral) AND title-abstract-keywords (forensic)) on 24 June 2024.

Already a decade ago, the US Food and Drug Administration converted to MPS as its primary genetic tool for pathogen detection [64]. Moreover, several efforts have described characterization of human microbiomes for identifying human body fluids [65-67]. Additionally, human skin bacterial communities were analyzed for human identification [68,69]. For example, Fierier et al [68] were able to recover skin-associated bacteria from surfaces (up to two weeks after deposition) and use their genetic profiles to distinguish who touched the items. By scanning entire microbial genomes ranging from a few thousand to a few million base pairs in size exquisite sensitivity, specificity, and resolution are achieved for public health and safety applications.

Forensic virology has been represented primarily by analyses of human immunodeficiency virus (HIV), hepatitis C virus, and SARS-CoV-1/2 coronaviruses in cause-of-death, criminal, and epidemiologic investigations [70-72] and today are facilitated by MPS. However, the human virome can be used potentially for characterizing unidentified cadavers, even in cases of skeletonized remains. aDNA studies, for example, have accessed human remains to identify the ancient hepatitis B virus (HBV) strains [73]. Those studies demonstrated that viral genomes can persist for centuries in human bone. Toppinen et al [74] were the first to extend use of persistent viruses' genomes for assisting in identification of human remains. They showed that parvovirus B19 (B19V) DNA sequences were as prevalent in World War II human remains (~45%) as in modern day cadavers [75,76]. Moreover, based on specific viral genotype (gt 3) detected in two of the ~70-year-old femurs (alleged casualties from two unidentified soldiers) pointed that there was greater support that the remains were soldiers of the Soviet Red Army. The particular virus type observed had never before been reported in

Northern Europe, and when combined with the maternal and paternal lineage DNA marker systems the determination of the origin of the remains was well-supported.

Indeed, human organs are rich in viral genomes and an array of viral sequences could improve original geolocation attribution for developing leads to identify human remains in which soft tissues are available, especially with those individuals who perish during the massive migrations of today. Using a custom in-solution hybrid capture method Toppinen et al [75] also enriched for 38 viral targets in femur bone extracts and were able to detect 12 different viruses including B19V, HBV, torque teno virus, JC polyomavirus (JCPyV), Merkel cell polyomavirus, human papillomavirus 31, herpes simplex virus 1 (HSV-1), varicella-zoster, Epstein-Barr, cytomegalovirus, and human herpesvirus 6B. More recently, Pyöriä et al [76] identified 17 different DNA viruses (primarily herpes-, parvo-, papilloma- and anelloviruses) present in soft tissues of recently deceased individuals, on average 6.7 per individual. Of these viruses, the global distribution of JCPyV has been studied by Forni et al [77]. They compared via phylogenetics over 1000 JCPyV strains and concluded that their geographic distribution roughly corresponded to major human migratory routes (Figure 3).

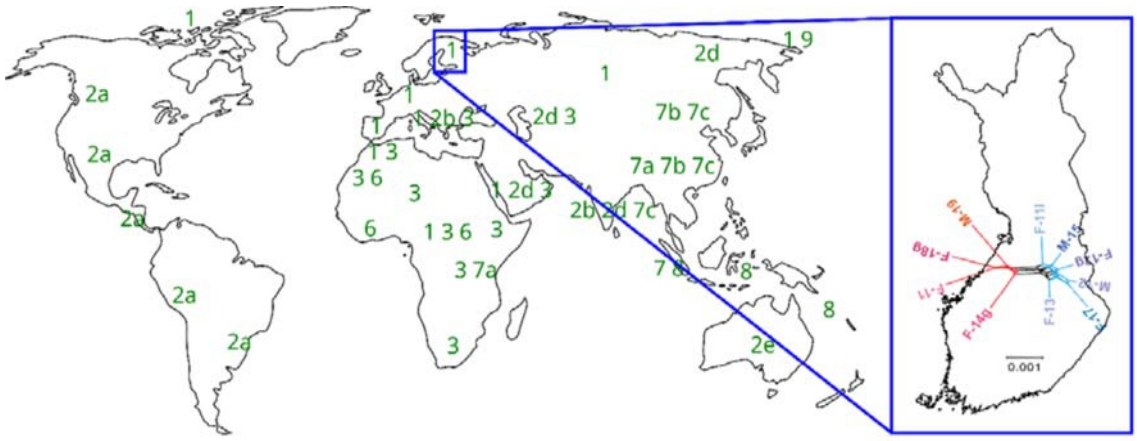

Figure 3. Distribution of JC polyomavirus (JCPyV) genotypes wordwide, and east-west division of herpes simplex 1 virus (HSV-1) strains in Finland. Forni et al [77] compared via phylogenetic analyses over 1000 JCPyV strains and concluded their global geographical distribution. Bowen et al [78] analyzed HSV-1 strains in Finland and showed particular eastern (blue strains) and western (red) virus types similar to distribution of Finnish human genomic data.

Furthermore, HSV-1 strains have been shown to fall, within Finland, into two distinct phylogenetic groups, potentially reflecting historical waves of human (and viral) migration into Finland [78]. The analysis of persistent viral DNAs in bone is encouraging and demonstrates that there is substantial untapped genetic information in genomic DNA from bone extracts for estimation of provenance and migration origins. More data are needed on the prevalence and strains from various geographic locations to increase accuracy of geolocation.

Conclusion

Two examples – forensic genetic genealogy for human identification and the virome for geolocation – are described here to illustrate the power and capabilities that MPS provides for forensic science. By employing MPS, either with targeted sequencing or WGS, the forensic community can revisit the utility and power of SNPs. By CE, testing was limited to a relatively low number of SNPs, and the methodologies were not quantitative. In contrast, with MPS thousands to upwards of several million SNPs can be assayed per sample affording tremendous power of discrimination and more versatile approaches for developing investigative leads via DNA. There are many applications that can be considered, such as direct comparison for identity testing, kinship for identification of human remains, and determinations of externally visible characteristics, bioancestry, geolocation, estimation of time of death, exploiting microbiome for human identification and estimation of time of death, to name a few. A novel avenue with MPS is investigation of the human tissue virome and its utilization in developing leads to identify human remains which is particularly important considering the ever-increasing mobility of people. It is evident that today MPS is an attractive, if not necessary tool, for the forensic scientist.

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ԴԱՏԱԿԱՆ ԳԵՆԵՏԻԿԱՅԻ ՎԵՐՋԻՆ ՀԱՐԱՑՈՒՅՑԻ ՓՈՓՈԽՈՒԹՅՈՒՆԸ, ԶԱՆԳՎԱԾԱՅԻՆ ԶՈՒԳԱՀԵՌ ՍԵՔՎԵՆԱՎՈՐՈՒՄ ԵՎ ՏՎՅԱԼՆԵՐԻ ՄԵԾ ԾԱՎԱԼՆԵՐ

Տոպինեն Մ., Սադժանտիլա Ա., Բուդոուլ Բ.

Հոդվածում նշվում է, որ դատական գենետիկան զարգացել է վերջին մի քանի տասնամյակների ընթացքում՝ ապահովելով չափազանց զգայուն և բարձր ճշգրտության արդյունքներ, որոնք օգտագործվում են քրեական, քաղաքացիական և հումանիտար գործերում անձի նույնականացման նպատակով: Ուշադրություն է դարձվում այն փաստի վրա, որ ԴՆԹ փվյալների բազաների ի հայտ գալը առավելապես ընդլայնել է արագ զարգացող հետաքննչական վարկածների բացահայտման գործընթացում ԴՆԹ դրոշմավորման ներուժը: Ընդգծվում է, որ նույնիսկ արդի մեթոդների, մարկերների և փվյալների բազաների կիրառման պարագայում հարյուր հազարավոր քրեական և քաղաքացիական գործեր չեն բացահայտվել՝ չնայած առկա են եղել անհրաժեշտ կենսաբանական ապացույցներ: Հոդվածում հիմնավորվում է այն միտքը, որ նման բացթողումը լրացնելու նպատակով, դատագենետիկների միջազգային մասնագիտական հանրությունը պատրաստ է հարացույցի փոփոխության, որը պայմանավորված է զանգվածային զուգահեռ սեքվենավորմամբ և միանուկլեոտիդային բազմություն անվանումով հայտնի գենետիկական մարկերների կիրառության վերսկսմամբ: Դատական գենետիկայի կամ առավել ճշգրիտ՝ դատական գենոմիկայի ներուժն ու հեռանկարները ներկայացնելու նպատակով հոդվածում քննարկվում են հետևյալ երկու օրինակները՝ դատագենետիկական ծագումնաբանության հետազոտությունների անցկացում՝ անձի նույնականացման նպատակով և վիրոմային վերլուծության իրականացում՝ տարածքաշրջանային դիրքի որոշման նպատակով:

Բանալի բառեր. *դատաբժշկական գենետիկա, անձի նույնականացում, զանգվածային զուգահեռ հաջորդականություն, միանուկլեոտիդային պոլիմորֆիզ, դատագենետիկական ծագումնաբանություն, վիրոմ, տարածքաշրջանային դիրք:*

НОВЕЙШАЯ СМЕНА ПАРАДИГМЫ В СУДЕБНОЙ ГЕНЕТИКЕ. МАССОВОЕ ПАРАЛЛЕЛЬНОЕ СЕКВЕНИРОВАНИЕ И БОЛЬШИЕ ОБЪЕМЫ ДАННЫХ

Топпинен М., Саджантила А., Будоул Б.

В статье отмечается, что за последние несколько десятилетий судебная генетика развивалась, имея целью обеспечить получение чрезвычайно чувствительных и высокоточных результатов, используемых для идентификации личности при расследовании уголовных дел и разрешении гражданских споров. Обращается внимание на то, что появление баз данных ДНК еще более расширило возможности судебно-медицинского типирования ДНК в быстро меняющейся криминалистической обстановке. Подчеркивается, что даже с использованием нынешних методов, маркеров и баз данных сотни тысяч уголовных и гражданских дел, с учетом их гуманитарно-правовой значимости, не были раскрыты, несмотря на то, что имелись необходимые биологические доказательства. В статье обосновывается мысль о том, что с целью устранения данного недостатка, международное профессиональное сообщество судебных экспертов-генетиков готово к смене парадигмы, вызванной появлением массового параллельного секвенирования и возобновлением генетических маркеров, известных как однонуклеотидный полиморфизм. В статье, с целью иллюстрирования текущих возможностей и будущего развития судебно-медицинской генетики (судебно-медицинской геномики), рассматриваются два практических примера: проведение судебно-генетических генеалогических исследований с целью идентификации личности и проведение анализа вирома с целью определения геолокации.

Ключевые слова: *судебная генетика, идентификация личности, массово-параллельное секвенирование, однонуклеотидные полиморфизмы, судебно-генетическая генеалогия, вирom, геолокация.*

The article was submitted: 30.04.2024
The article was accepted: 17.06.2024