

2018 - Peer Reviewed Journal Article

Citation:

Scudder, N., McNevin, D., Kelty, S. F., Walsh, S. J., & Robertson, J. (2018). Massively parallel sequencing and the emergence of forensic genomics: Defining the policy and legal issues for law enforcement. *Science and Justice*, 58(2), 153-158. DOI: 10.1016/j.scijus.2017.10.001

Copyright:

©2018. This manuscript version is made available under the CC-BY-NC-ND 4.0 license <http://creativecommons.org/licenses/by-nc-nd/4.0/>

Version:

This is an Accepted Manuscript of an article published in *Science & Justice* available online at <http://dx.doi.org/10.1016/j.scijus.2017.10.001>.

Changes resulting from the publishing process may not be reflected in this document.

Massively parallel sequencing and the emergence of forensic genomics: defining the policy and legal issues for law enforcement

Abstract

Use of DNA in forensic science will be significantly influenced by new technology in coming years. Massively parallel sequencing and forensic genomics will hasten the broadening of forensic DNA analysis beyond short tandem repeats for identity towards a wider array of genetic markers, in applications as diverse as predictive phenotyping, ancestry assignment, and full mitochondrial genome analysis. With these new applications come a range of legal and policy implications, as forensic science touches on areas as diverse as ‘big data’, privacy and protected health information. Although these applications have the potential to make a more immediate and decisive forensic intelligence contribution to criminal investigations, they raise policy issues that will require detailed consideration if this potential is to be realised. The purpose of this paper is to identify the scope of the issues that will confront forensic and user communities.

Keywords

Forensic DNA, predictive phenotyping, forensic genomics, privacy

1. Introduction

Forensic science has benefited greatly from advances in technology [1-3]. From the development of alternate light sources for detecting biological material at crime scenes to increased digitisation and databasing, the world of forensic science has not stood still. However, forensic laboratories are now facing a major technology and policy shift, the likes of which it has arguably not yet had to grapple [4]. The increasing use of forensic genomics, both through more cost-effective analysis of single-nucleotide polymorphisms (SNP) and the widespread adoption of massively parallel sequencing (MPS) will not only alter the technological platform of contemporary laboratories, but will pose new legal and policy challenges as well.

Early adoption of DNA analysis for forensic science, more than thirty years ago, came with assertions concerning so-called ‘junk DNA’ [5]. The argument for policy-makers was that forensic DNA profiling, while derived from and subject to the underpinning laws of genetics, purposefully selected as markers repetitive elements of DNA called satellites or tandem repeats for their variance and support for statistical modelling which were not associated with genes known to make us who we are as individuals [6, 7]. The argument that any short tandem repeats (STRs) are, in fact, ‘junk DNA’ cannot now be reasonably sustained [8, 9]. Nonetheless, forensic laboratories collected information about a relatively small number of markers, and distilled that data into profiles in a database, returning to population genetics only for the purposes of expressing the results in a valid statistical form in terms of their frequency of occurrence [10].

New advances in our understanding of functional genomics have consigned the ‘junk DNA’ argument firmly to the history books. More sophisticated, yet cost-effective capabilities,

now give forensic scientists the ability to investigate a wider array of genetic markers, for predictive phenotyping, ancestry assignment, and full mitochondrial genome analysis [11, 12].

In doing so, laboratories will open themselves to concepts such as ‘big data’, health records discrimination, and a granularity and accessibility of raw genetic data perhaps best described as being akin to home viewers moving from analogue video tape to digital media. Like that move, forensic labs must remain focused on providing fit for purpose and cost-effective DNA services in support of the criminal justice system and new methods must support and not undermine public confidence in those now well-established outcomes.

2. New technology - from traditional DNA profiling to predictive phenotyping

Our ability to analyse multiple genetic markers simultaneously, at greater speed and lower cost, together with more readily available population databases, makes it feasible to draw a range of genetic inferences. Drawing on tools used in medical research fields to identify genes associated with hereditary disease and applying similar techniques to forensic samples of unknown origin presents many opportunities [13, 14].

2.1. Externally visible characteristics

In forensic science, the focus of predictive phenotyping is principally on genes that may influence our externally visible characteristics (EVCs), with eye and hair colour being the focus of much of the early research [15]. Claes et al [16] note that ‘the ultimate goal of evaluating evidentiary DNA is to assign a biological origin to the sample with a high degree of statistical certainty’ and that ‘to help an investigation out of an impasse...a DNA based prediction of externally visible characteristics... or ancestry from the evidentiary sample can be considered’.

The accuracy of these methods, and the types of externally visible characteristics that can be targeted, is increasing [17]. Commentators have outlined a variety of research currently under way into new methods, including prediction of ‘male baldness, hair morphology, and body height’ [18]. One commentator even hypothesised that methods could extend to ‘probability-weighted physical description of...gender, race or ethnicity, skin pigmentation, eye color, natural hair color, hair texture, nose width, dimpling in chin and cheek, earlobe attachment, adult height, patterned baldness, chronological age, natural dominant hand, lip height, freckling, and in some cases, even surname’ [19].

Predictive phenotyping is already in use in forensic science, albeit presently only in a tiny fraction of criminal investigations [20-22]. The technology can also be applied, in conjunction with anthropology, to assist in the identification of decomposed human remains [23, 24].

However, as Murphy [21] observed, ‘the vast majority of crimes are between people who know one another’. In those instances, traditional profiling using DNA fragment length analysis via capillary electrophoresis (CE) will likely remain the principle DNA analysis tool for the time being. This distinction is particularly relevant as DNA genotyping focusing solely on STRs is moving in a different direction: towards field-portable and ‘real-time’ devices [25]. These instruments are designed to provide faster analysis of a smaller number of genetic markers suitable for initial screening against DNA databases, thereby assisting investigators in making timely operational decisions. A significant step in this process is the signing into law in the United States in August 2017 of the *Rapid DNA Act of 2017*, which requires the Federal Bureau of Investigations to develop standards for automated DNA analysis instruments and to allow for such instruments to be connected to their national DNA database.

There is little doubt that these so-called ‘rapid DNA’ devices will make their way into the field first. But MPS technology could well follow in future years [26]. If it does, such

devices may ultimately provide investigators with near-immediate information about the likely appearance of a suspect, even without establishing identity using databases.

In the laboratory, however, the continued use of CE for DNA would only be logical while these instruments provide faster, cheaper or higher quality DNA results. Schuster [4] provides an overview of the development of MPS to date, and the challenges it has already overcome in a field dominated by CE. Should these advances continue, a time may come when every sample reaching the laboratory would be subjected to some form of MPS. It may not then be cost-effective to use different sequencing kits to target different genetic markers for different samples to provide 'new' evidence. The same level of genetic analysis could be undertaken for those samples from cases where there are no suspects, and for samples from crime scenes where there are already one or more suspects.

2.2. *Biogeographical ancestry (BGA)*

A particularly useful phenotype for investigative purposes (and one of the easiest to predict) is biogeographical ancestry (BGA). MPS will bring into widespread and cost-effective use the capability to make certain predictions about the BGA of the donor of biological material [6, 27]. The identification of ancestry informative markers (AIMs), particularly when used in conjunction with predictions about EVCs, can assist investigators to narrow a pool of suspects [20].

The usefulness of ancestry information would be dependent on population demographics. The technique clearly has increased effectiveness in populations with diverse biological ancestry. In locations with relatively homogenous populations, only a prediction rare in that population would likely be of any real assistance to investigators, for example, investigators in Asia may find results particularly probative if this method suggested a suspect

may be a red haired Northern European. As such, the adoption of the technique, if made public, could be criticised for reinforcing racial prejudice: a view that foreigners or ethnic minorities are more likely to be responsible for crime [28]. There is also potential for the technology to be applied in a skewed manner, more likely used in such cases and again facilitating a bias against minority groups.

Equally, however, it could be argued that an objective indicator of the BGA of a potential offender may help to eliminate bias in eyewitness testimony where ethnic minorities can be unfairly targeted. Of the 349 people exonerated by the Innocence Project in the United States using post-conviction DNA analysis (including 20 who served time on death row, at the time of writing), over 70% of these wrongful convictions were associated with eyewitness misidentification and over 60% of the exonerees were African American [29]. Eyewitness testimony is well known to be highly susceptible to false memories and bias [30-32]. Prediction of BGA from genotype offers the potential to at least corroborate or challenge eyewitness testimony.

2.3. Mitochondrial DNA analysis

Mitochondrial DNA analysis is not new, with a study of its forensic application by Wilson et al [33] being just one example in the literature. However, forensic genomics and adoption of MPS will further revolutionise this capability and allow for full analysis of the mitochondrial genome [34, 35]. Mitochondrial DNA analysis using MPS is already assisting the United States Department of Defense DNA Registry in identification of skeletonised human remains from conflicts as far back as the Second World War [36]. Only a few years ago, DNA from these remains may not have yielded a useable result. Now previously unidentified soldiers are being returned to their families [37]. However, laboratories engaged in this process are increasingly

becoming aware of the privacy implications of full mitochondrial genome sequencing, including the potential to reveal predictive health information about individuals or family members, and taking steps to safeguard genetic information and ensure soldiers' family members are giving their full and informed consent [38].

While forensic laboratories are in no way focused on studying genes linked to predisposition to disease, it is an inescapable fact that the genetic data is there in abundance. This raises the possibility that, under some circumstances, laboratories will need to develop policies as to how to deal with the inadvertent discovery of predictive medical information which may not have been detected using CE capabilities.

3. Emerging considerations

The benefits of predictive phenotyping rely heavily on an integrated approach to forensic analysis. The need to understand the 'context of crime', including operational imperatives, as well as the broader privacy and legal implications, will in many ways determine whether this capability can be put into effective mainstream use [39].

3.1. Operationalising the capability

'It is a capital mistake to theorise before you have all the evidence. It biases the judgement' - Sherlock Holmes (A. C. Doyle, *A Study in Scarlet*, 1887)

Forensic evidence can significantly inform investigations, sometimes representing a turning point through guiding the prioritisation of investigative leads. Like all forms of forensic evidence, there is always a danger that DNA evidence can be misconstrued, resulting in the

diversion of scarce police or scientific resources away from other lines of enquiry [40]. The different expectations concerning forensic evidence, when applied in an intelligence context as against a prosecutorial context, was discussed in a recent report by the United States President's Council of Advisors on Science and Technology [41] and by Kayser [17].

Employing EVC or BGA prediction does not – at the fundamental level – change the likelihood that forensic evidence can be contextually misconstrued. DNA from post-blast fragments could carry the DNA of the bomb-maker, a victim or an innocent third-party. However, this is where phenotype and ancestry prediction needs to be approached with a degree of caution. If biological material was analysed using traditional methods for STRs, yielding a DNA profile but no match on a DNA database, the profile would likely remain on that database as an unidentified crime scene profile indefinitely. With no match, no further utilisation of that DNA evidence would be possible, unless and until a suspect was otherwise identified and a reference DNA sample obtained.

If the fragment did match a reference sample on the database, we could expect that lead to be followed up and the individual confirmed as a suspect or excluded (for example, the DNA might have come from a victim, have been deposited before the crime occurred, or have been deposited afterwards by an investigator or first responder at the crime scene).

With EVC and BGA prediction, however, our biological sample may yield intelligence information immediately - perhaps eye or hair colour – that could influence the prioritisation of investigative leads [17]. The weighting that can be given to that evidence, in the overall context of a forensic examination, is therefore quite important.

This is, of course, the same for all types of evidence. In *United States v. Wade* 388 U.S. 230, 288 (1967), the United States Supreme Court noted that the ‘vagaries of eyewitness identification are well-known; the annals of criminal law are rife with instances of mistaken identification’ [32]. The National Academy of Sciences [32] notes that a person’s ‘vision does

not capture a perfect, error-free “trace” of a witnessed event’ but is rather influenced by a range of factors. A study by Brigham, Maass, Snyder and Spaulding [30] used eyewitness behaviour in controlled circumstances to show that, in cases where an eyewitness attempted to make an identification of an individual from six photographs, accuracy rates were surprisingly low. Wixted et al [42] noted the importance of considering the correlation between confidence in identification and the accuracy of that identification.

An eyewitness identification, supported by prediction of EVCs from recovered genetic material at the crime scene, significantly alters the confidence around both the eyewitness and forensic evidence [20]. In this way, investigators can benefit from both the ability of an eyewitness to give context (such as their interpretation of a suspect’s actions, demeanour or behaviour) and the ability of predictive phenotyping to add a statistical framework around assertions of their physical appearance.

The value of EVC or BGA prediction will, of course, vary depending on the facts of the case. There are many instances where a forensic scientist, in triaging or prioritising their evidence plan, can make reasonably objective determinations as to the inculpatory nature of biological evidence. Fresh blood at a murder scene or semen on clothing from a sexual assault are more inculpatory than a cigarette butt of unknown origin located after a crime in a public place.

Commentators such as Brady and Engelhaupt [20] have noted the real advantage of predictive phenotyping is in narrowing down the suspect pool. Given the variation in the inculpatory nature of evidence, this could perhaps be rephrased as an ability to more effectively prioritise leads. That is, at a stage in the investigation before any suspect or group of suspects have been settled upon, it may be possible to prioritise investigative resources and ensure that individuals more likely to have been the source of genetic material at a crime scene are considered first.

However, if a suspect pool is small enough, it can be argued that identification markers more appropriately come into play. It is difficult to presently anticipate an investigative or cost advantage in undertaking phenotype or ancestry analysis for a case involving a handful of suspects, whose identities are already known to police. The larger the suspect pool, the more investigators must assess the inculpatory nature of the forensic evidence, so as to ensure that suspects are not ruled out based on a ‘red herring’ scenario [20, 43].

A corollary of this, however, is that phenotype prediction has a potential of deterrence. Already, it has been used to generate billboard images of suspected litter-bugs in Hong Kong and to produce photo-fits of dogs wanted for soiling parks in Great Britain [44, 45] where the probable inaccuracies of these images are outweighed by their deterrence value. As the public become more aware of this technology, it has the potential to deter criminals who may once have believed that their lack of inclusion on a DNA database made them immune to such detection. Criminals may also, of course, respond by increasing their forensic counter-measures, to attempt to minimise the shedding of DNA or possibly to pollute the DNA at crime scenes.

3.2. Privacy implications

Critics have suggested that EVC and BGA prediction represent a significant invasion of privacy or a path to more subjective genetic predictions concerning criminality and guilt [21, 46-48]. Should we be concerned, as forensic laboratories generate more and more genetic data? Or is this a logical extension of the ‘big data’ argument in other fields of medical and scientific research [13]?

It is important to distinguish up-front between the capability of the technology and its practical application. It is true that, in many cases, the same basic instrument (employing

different panels of genetic markers) could shift focus from eye colour to cancer risk. But is such a situation likely to occur? Forensic laboratories already possess DNA swabs taken from convicted criminals, suspects and victims. A nefarious scientist could already divert these samples into a medical testing environment, should they so wish. Although commentators such as Murphy [21] have speculated as to the possible application of more sensitive testing to known forensic samples, there is no evidence to date of such misuse of genetic material entrusted to forensic scientists. The counter-argument, put forward in 2001 by commentators such as Webb and Tranter [48], is that the growth of conventional forensic DNA analysis within the criminal justice system has already normalised the process. According to such a view, predictive phenotyping represents just a step towards more intrusive testing, such as attempts to genetically predict and punish future criminal behaviour.

Notwithstanding this view, the most immediately identifiable risk with any repository of genetic information comes from data re-identification and aggregation [49, 50]. When forensic DNA was first adopted, laboratory processes quickly distilled raw genetic data into genotypes. Some limited raw genetic data was generated, but seldom left the CE instrument. It was the allele designations at particular loci that had the statistical and probative relevance.

The application of forensic genomics could see more genetic data stored, indexed and databased. But the data is still quite targeted, looking at BGA and EVCs, predominantly from crime scene samples of unknown origin [15]. What privacy implications could there be? Is the risk to privacy any greater than a forensic investigator relying on an eyewitness account to narrow a pool of suspects to a particular ethnic minority with particular facial characteristics? Is the risk any greater than an investigator narrowing a pool of suspects to known asthma sufferers, which may have probative value for the investigation of a crime where a metered dose inhaler was recovered from the crime scene?

It can be argued that crime scene samples, of unknown origin, pose few privacy implications. Privacy issues might arise if a forensic laboratory proposed searching its genetic data against medical data or genealogical DNA holdings from commercial providers, so as to potentially identify the donor of a particular biological specimen [7]. However, it can be argued that these issues relate to the policy and legal implications around the release of that medical or genealogical data, known to be from specific individuals, rather than use of the genetic information held by the forensic laboratory. A robust privacy and legislative regime around the collection of genetic samples, for such purposes as medical diagnosis, ought to carefully consider donor consent around secondary use [51]. If they do not, privacy and health data protection legislation such as the United Kingdom's *Data Protection Act 1998*, Australia's *Privacy Act 1988* and the United States *Health Insurance Portability and Accountability Act of 1996* may well stand as a barrier to such a 'dragnet' approach.

Predictive phenotyping technology already exists commercially, and is easily accessible by consumers, particularly in a genealogy context. A rogue investigator or scientist could potentially already access this technology, covertly analysing samples and sidestepping a variety of legal and policy safeguards [52].

Setting this scenario aside, however, other privacy issues could also arise. Once a suspect comes to police attention and a DNA reference sample is taken, the unknown crime scene sample ceases to be unknown if there is a match. By linking an unknown crime scene sample to an individual, the laboratory may well have created a piece of 'personal data', 'personal information' or 'protected health information' (depending on the legislation in effect in the country in which the identification is made). At this point, additional safeguards may come into play.

However, once this link to a suspect is made, STRs are logically of highest evidentiary significance. Predictive EVC or BGA information ceases to be of much investigative relevance.

Once these identification markers are shown to ‘match’, the case moves to a different investigative phase and it is quite likely that the predictive genetic information, while necessary to show transparency in the forensic or investigative process, will not reach the court room.

Where EVC or BGA prediction remains relevant, new sensitivities could come into play. A prediction of a suspect’s ancestry could challenge the way that individual perceives themselves, particularly in Western culture with a reasonably narrow view of ethnicity [53]. As commercial providers are finding, genetic ancestry prediction is vexed: for every individual who unexpectedly finds out from a genealogy service that they have a component of their genetic makeup consistent with the indigenous American or the Australian Aboriginal populations, there is almost sure to be another who finds out that grandma was not, as she said, ‘half Cherokee’ [6], at least not in a purely genetic sense. The laws of random inheritance can cause a divergence between genealogy in a linear sense, and genetics [6].

Another privacy concern relates to ‘data breaches’, should they occur with respect to identified genetic data held by a forensic laboratory [54]. The genetic data held by the laboratory is, as has been discussed, limited to those markers that would assist in determining gender, or predicting externally visible characteristics or ancestry. However, overlapping markers between forensic data and external data holdings could allow that other genetic data, thought to be anonymous, to be re-identified. Once re-identified, it may be possible to make other inferences about an individual, such as prevalence to certain diseases, and to tie those inferences to a known individual.

Very recently, Edge et al. went further by demonstrating that 90–98% of forensic STR records can be connected to corresponding SNP records and vice versa using 13 STR loci in one data set and 643,563 genome-wide SNPs in the other science [55]. The STR and SNP profiles had no shared markers but the associations were able to be made because some of the markers are in linkage disequilibrium (because they are located close to each other on

chromosomes). This clearly demonstrates the ability to associate phenotypic markers (SNPs) with identity markers (STRs).

There are other potential privacy risks, and even enhancements, of this new technology, which require further analysis. Commentators such as Gloudemans and Shamaprasad [14] have cautioned against what they term community DNA ‘dragnets’ [56]. Such community-wide DNA sampling programs are rare, and generally quite expensive, but not unheard of. However, it is possible that the use of predictive phenotyping on crime scene samples could allow investigators to limit or target such a strategy – at least in the first instance. Just as some previous community-wide sampling programs have been limited, quite logically, to one biological gender, it might be possible to limit the collection to individuals with specific EVCs. Such an approach could reduce the privacy intrusion and represent a cost-saving.

Predicting EVC and BGA could also narrow lines of enquiry and limit privacy intrusions, without the need for a DNA ‘dragnet’. If a physical characteristic allowed police to limit a suspect pool, it may in theory limit the need to access telephone records, social media or other personal information which may presently be analysed more extensively.

An important limitation, however, is the changeable nature of characteristics such as hair and eye colour, which would make such targeting relatively easy for an offender to circumvent [57].

3.3. Legislative implications

There are few legislative models in existence for the forensic use of EVC and BGA. The Netherlands passed legislation in 2003 permitting the use of phenotyping but restricting its use to traits the donor would be aware of. For practical purposes, this restricts use to EVCs visible from birth [58]. Certain countries, including Germany, restrict DNA to non-coding markers

although such a distinction between coding and non-coding – while establishing a legal intent – is not scientifically as clear [59]. More recently, there has been debate in Germany about proposals to permit prediction of EVC, with some commentators suggesting a cautious approach [60].

Aside from specific regulation of the forensic use of EVC and BGA, the capability does intersect with legislative requirements concerning the management of health records. These requirements are not new to medical research and diagnostic laboratories. However, forensic laboratories have seldom had to grapple with this issue in the past. Laboratories may find that forensic genomics puts their results on the fringe of legislation concerning health records. The state of New South Wales in Australia, for example, exempts law enforcement agencies from these requirements (*Health Records and Information Privacy Act 2002 (NSW)*, s 17). However, this will require consideration on a jurisdiction by jurisdiction, even laboratory by laboratory, basis.

EVC and BGA prediction, and the possibility of mitochondrial DNA analysis, were not in the minds of legislators when most jurisdictions' DNA legislation was being drafted. In fact, in the Australian context, the Australian Law Reform Commission made only a brief reference to such advances in technology under 'function creep' in their 2003 report into genetic privacy [61]. Importantly, the legislation is predominantly focused on the processes for obtaining reference DNA samples, either voluntarily or coercively, and the resulting DNA databasing processes. The contribution made by EVC and BGA prediction is at an intelligence phase of an investigation, and arguably the two processes can co-exist with very different legal requirements.

It would seem illogical to add phenotype marker results to such databases, given existing STRs are sufficient for comparing profiles between databases. Mitochondrial DNA results are slightly different, given the potential for enhanced familial matching. Again,

however, database searching capabilities would work best with a relatively small number of pre-defined markers, rather than a bulk upload of an entire mitochondrial genome.

A question may arise, however, if it becomes financially expedient for a laboratory to use MPS or similar technologies on a wider range of genetic samples. A laboratory might ultimately find it less efficient to use current DNA technology when MPS offers cost or productivity benefits.

Current technology, and DNA databases, already contain one marker which is highly informative. The XX or XY genotype at the amelogenin locus is indicative of biological gender, and has long been a part of DNA testing methods and kits. Arguably, biological gender is relevant only to an unsolved crime scene sample, and is useful to investigators in the same way as EVC or BGA prediction. However, it would be extremely inefficient to process DNA samples from known individuals with different chemistry to crime scene samples, to exclude the amelogenin locus for samples of known origin.

Creating information about a donor's biological gender serves only one logical purpose: a form of quality control. A mismatch in the amelogenin result between a crime scene and reference sample could be indicative of a larger problem, but a match adds nothing to the forensic result and little to the derived statistical probabilities.

Could EVC or BGA prediction then be viewed merely as an extension of the way in which a biological gender informative marker has been used for nearly 20 years?

Or could samples be processed using MPS, with only those identification markers intended for upload to a DNA database retained and the remaining data purged? Such an approach may also attract criticism. There have been instances where reference sample profiles have been switched, sometimes not discovered until a suspect has served years in prison [62]. Purging some of the data may remove traceability and undermine quality assurance practices.

3.4. Policy implications

As forensic laboratories move to implement phenotype prediction and/or ancestry assignment, mitochondrial DNA analysis or any number of other applications to which forensic genomics may ultimately be put, there will be a need to grapple with relevant policy considerations.

How should the technology be applied? What are the most appropriate standards around information access, storage and security? How much genetic data needs to be retained, and for how long?

In addition to policy around laboratory practices, there will be a requirement to critically examine training requirements, both for forensic scientists involved in delivering results to investigators and to investigators themselves. Ensuring a high awareness of how MPS and its applications differ from previous DNA technology and uses will clearly be important.

3.5. Intelligence uses

Predictive DNA technology can obviously offer far more than a few investigative leads and some genetic ‘wanted’ posters, predominantly for cold case investigations. There is significant potential to assist in the detection and disruption of crime, and to feed into intelligence at its broadest. Like other forensic information, it can be mined, combined with other criminal intelligence, and analysed over time. Older samples can be revisited based on new scientific breakthroughs [63]. The identification of a suspect, by whatever means, can create a feedback loop to further enhance the technology and the way law enforcement are trained to use it. How accurate were the predictions? What factors may influence the ability of police to successfully use the information derived?

Walsh describes an ‘integrated model’, where forensic intelligence provides both real-time analysis of case information, but also a meaningful contribution of ‘non-comparative forensics’, defined as tools that can ‘suggest intelligence links by virtue of their outcomes [rather than] direct comparison’ [64]. Predictive phenotyping capabilities are such a tool, and their inclusion within a larger context of crime analysis could save investigative resources, reduce recidivism and have a deterrent effect.

4. Conclusion

The introduction of MPS, making prediction of EVC and BGA more cost effective, has occurred without a great deal of scrutiny by other actors in the criminal justice system. However, this is expected, as the contribution of these processes is in the forensic intelligence rather than the prosecution phase of an investigation [64].

Basic genetics hasn’t changed, and nor has the way the vast majority of the DNA results that find their way into courtroom will be presented.

However, the more widespread use of this technology will require forensic scientists and investigators to work even more closely together to ensure the context and limitations of predicting EVC and BGA are understood by all. The potential for such DNA evidence, viewed in isolation, to divert an investigation at a critical juncture is real. The potential for predictions to reinforce existing prejudices clearly exists. As does the concern of those who are drawn into the criminal justice system as to what personal information may be derived, directly or indirectly, which may reveal information about their health status.

Each jurisdiction will need to ensure appropriate consideration is given to policy, privacy and legal considerations within the broader criminal justice system. These aspects are the intended subject of future papers.

Bibliography

- [1] S.J. Walsh, Recent advances in forensic genetics, *Expert Review of Molecular Diagnostics*, 4 (2004) 31-40.
- [2] C. Lennard, M. Stoilovic, Application of forensic light sources at the crime scene, in: *The Practice of Crime Scene Investigation*, CRC Press, Boca Raton, FL, 2004.
- [3] P.D. Martin, H. Schmitter, P.M. Schneider, A brief history of the formation of DNA databases in forensic science within Europe, *Forensic Science International*, 119 (2001) 225-231.
- [4] S.C. Schuster, Next-generation sequencing transforms today's biology, *Nature*, 200 (2007) 16-18.
- [5] J.K. Wagner, Out with the "Junk DNA" Phrase, *Journal of Forensic Sciences*, 58 (2013) 292-294.
- [6] T. Frudakis, *Molecular Photofitting: Predicting Ancestry and Phenotype Using DNA*, Academic Press, Boston MA, 2010.
- [7] D. Gusella, No Cilia Left Behind: Analyzing the Privacy Rights in Routinely Shed DNA Found at Crime Scenes, *BCL Rev.*, 54 (2013) 789.
- [8] M. Gymrek, T. Willems, A. Guilmatre, H. Zeng, B. Markus, S. Georgiev, M.J. Daly, A.L. Price, J.K. Pritchard, A.J. Sharp, Abundant contribution of short tandem repeats to gene expression variation in humans, *Nature Genetics*, (2015) 22-29.
- [9] A.N. Kitchen, Genetic privacy and latent crime scene DNA of non-suspects: how the law can protect an individual's right to genetic privacy while respecting the government's important interest in combating crime, *Crim Law Bull*, 52 (2015).
- [10] Federal Bureau of Investigations, Combined DNA Index System (CODIS). <https://www.fbi.gov/services/laboratory/biometric-analysis/codis> (accessed 08.03.17).
- [11] P. Shrivastava, T. Jain, V.B. Trivedi, DNA fingerprinting: a substantial and imperative aid to forensic investigation, *European Journal of Forensic Sciences*, 3 (2016) 1.
- [12] M. Zieger, S. Utz, About DNA databasing and investigative genetic analysis of externally visible characteristics: a public survey, *Forensic Science International: Genetics*, 17 (2015) 163-172.
- [13] C.G. Chute, M. Ullman-Cullere, G.M. Wood, S.M. Lin, M. He, J. Pathak, Some experiences and opportunities for big data in translational research, *Genetics in Medicine*, 15 (2013) 802-809.
- [14] M. Gloudemans, N. Shamaprasad, Current Issues in Forensic DNA Applications. <http://www.pged.org/wp-content/uploads/2015/04/Current-Issues-in-Forensic-DNA-Applications.pdf>, 2015 (accessed 17.05.16).
- [15] G. Emmerich, The Reality of DNA Phenotyping. <http://www.promegaconnections.com/the-reality-of-dna-phenotyping/>, 2016 (accessed 29.10.16).

- [16] P. Claes, D.K. Liberton, K. Daniels, K.M. Rosana, E.E. Quillen, L.N. Pearson, B. McEvoy, M. Bauchet, A.A. Zaidi, W. Yao, Modeling 3D facial shape from DNA, *PLoS Genet*, 10 (2014) e1004224.
- [17] M. Kayser, Forensic DNA Phenotyping: Predicting human appearance from crime scene material for investigative purposes, *New Trends in Forensic Science Genetics*, 18 (2015) 33-48.
- [18] M. Kayser, Forensic DNA phenotyping: DNA testing for externally visible characteristics, Academic Press, London, 2013.
- [19] C.E. MacLean, Creating a wanted poster from a drop of blood: using DNA phenotyping to generate an artist's rendering of an offender based only on DNA shed at the crime scene, *Hamline L. Rev*, 36 (2014) 1.
- [20] H. Brady, E. Engelhaupt, Can you rule out suspects using faces drawn from DNA?, *National Geographic*, Vol 230 No 1 (2016).
- [21] E. Murphy, Legal and ethical issues in forensic DNA phenotyping, New York University Public Law and Legal Theory Working Papers. Paper 415, (2013).
- [22] B. Pike, How just a drop of DNA can create a mugshot, in: *Daily Telegraph*, Sydney, 10 October 2015.
- [23] C. Wood, The future of forensic identification? <http://www.govtech.com/public-safety/The-Future-of-Forensic-Identification.html>, 2013 (accessed 15.09.16), Government Technology, (2013).
- [24] J. Ward, To investigate specialist DNA techniques for the identification of compromised human remains, NSW Forensic & Analytical Science Service, (2016).
- [25] P. Liu, S.H. Yeung, K.A. Crenshaw, C.A. Crouse, J.R. Scherer, R.A. Mathies, Real-time forensic DNA analysis at a crime scene using a portable microchip analyzer, *Forensic Science International: Genetics*, 2 (2008) 301-309.
- [26] T.C. Glenn, Field guide to next-generation DNA sequencers, *Molecular Ecology Resources*, 11 (2011) 759-769.
- [27] C. Phillips, Forensic genetic analysis of bio-geographical ancestry, *Forensic Science International: Genetics*, 18 (2015) 49-65.
- [28] L. Kirchner, Will 'DNA phenotyping' lead to racial profiling by police? <http://www.psmag.com/politics-and-law/will-dna-phenotyping-lead-to-racial-profiling-by-police>, 2015 (accessed 05.04.17).
- [29] Innocence Project, DNA Exonerations in the United States, <https://www.innocenceproject.org/dna-exonerations-in-the-united-states/>, 2017 (accessed 25.04.17).
- [30] J.C. Brigham, A. Maass, L.D. Snyder, K. Spaulding, Accuracy of eyewitness identification in a field setting, *J Pers Soc Psychol*, 42 (1982) 673.
- [31] J.T. Wixted, L. Mickes, J.C. Dunn, S.E. Clark, W. Wells, Estimating the reliability of eyewitness identifications from police lineups, *Proceedings of the National Academy of Sciences*, 113 (2016) 304-309.
- [32] National Academy of Sciences, Identifying the Culprit: Assessing Eyewitness Identification, National Academies Press, Washington DC, 2014.
- [33] M.R. Wilson, J.A. DiZinno, D. Polanskey, J. Replogle, B. Budowle, Validation of mitochondrial DNA sequencing for forensic casework analysis, *Int J Legal Med*, 108 (1995) 68-74.
- [34] J.L. King, B.L. LaRue, N.M. Novroski, M. Stoljarova, S.B. Seo, X. Zeng, D.H. Warshauer, C.P. Davis, W. Parson, A. Sajantila, High-quality and high-throughput massively parallel sequencing of the human mitochondrial genome using the Illumina MiSeq, *Forensic Science International: Genetics*, 12 (2014) 128-135.

- [35] L. Chaitanya, I.Z. Pajnič, S. Walsh, J. Balažic, T. Zupanc, M. Kayser, Bringing colour back after 70 years: Predicting eye and hair colour from skeletal remains of World War II victims using the HIrisPlex system, *Forensic Science International: Genetics*, 26 (2017) 48-57.
- [36] M. Bittle, DNA lab plays vital role for military families, in: *Delaware State News*, 12 Dec 2015.
- [37] M. Parker, Korean War soldier from Pinckneyville to be buried 66 years after being reported missing, in: *The Southern*, 23 Nov 2016.
- [38] United States Department of Defense, DOD Instruction 5154.30: Armed Forces Medical Examiner System (AFMES) Operations, (2015).
- [39] S.J. Walsh, O. Ribaux, J.S. Buckleton, A. Ross, C. Roux, DNA profiling and criminal justice: a contribution to a changing debate, *Australian Journal of Forensic Sciences*, 36 (2004) 34-43.
- [40] R. Julian, S. Kelty, J. Robertson, Get it right the first time: critical issues at the crime scene, *Current Issues Crim. Just.*, 24 (2012) 25.
- [41] President's Council of Advisors on Science and Technology, Report to the President - Forensic Science in Criminal Courts: Ensuring Scientific Validity, (2016).
- [42] J.T. Wixted, L. Mickes, S.E. Clark, S.D. Gronlund, H.L. Roediger III, Initial eyewitness confidence reliably predicts eyewitness identification accuracy, *American Psychologist*, 70 (2015) 515.
- [43] P. Gill, *Misleading DNA Evidence: Reasons for Miscarriages of Justice*, Elsevier Science, London, 2014.
- [44] E. Chan, E. Chow, Litterbugs are shamed on electronic billboards after their faces are recreated using DNA taken from cigarette butts – and even a condom – discarded on the streets of Hong Kong, in: *Daily Mail*, 21 May 2015.
- [45] B. Webster, Pugshots to shame messy culprits, in: *The Times*, 13 July 2016.
- [46] E. Kretowicz, Emerging DNA technology will impinge on privacy: Civil Liberties Australia, in: *The Canberra Times*, 17 Nov 2013.
- [47] S. Rushton, *Familial Searching and Predictive DNA Testing for Forensic Purposes: A Review of Laws and Practices*, Victoria Law Foundation Legal Police Internship Program, (2010).
- [48] E. Webb, K. Tranter, Genes R Us ethics and truth in DNA, *Alternative Law Journal*, 26 (2001) 168-173.
- [49] B. Malin, L. Sweeney, Re-identification of DNA through an automated linkage process, in: *Proceedings of the AMIA Symposium*, American Medical Informatics Association, 2001, pp. 423-427.
- [50] M. Humbert, K. Huguenin, J. Hugonot, E. Ayday, J.-P. Hubaux, De-anonymizing genomic databases using phenotypic traits, *Proceedings on Privacy Enhancing Technologies*, 2015 (2015) 99-114.
- [51] Royal College of Pathologists of Australia, *Massively Parallel Sequencing Implementation Guidelines*, (2015).
- [52] P. Aldhous, M. Reilly, How my genome was hacked, *New Scientist*, 201 (2009) 6-9.
- [53] P. Morrison, Patt Morrison asks: Rachel Dolezal on racial fluidity and her changing identity, in: *Los Angeles Times*, 8 Mar 2017.
- [54] A. Cambon-Thomsen, The social and ethical issues of post-genomic human biobanks, *Nature Reviews Genetics*, 5 (2004) 866-873.
- [55] M.D. Edge, B.F.B. Algee-Hewitt, T.J. Pemberton, J.Z. Li, N.A. Rosenberg, Linkage disequilibrium matches forensic genetic records to disjoint genomic marker sets, *Proceedings of the National Academy of Sciences*, 114(22):5671-6, 30 May 2017.

- [56] R. Scammell, DNA evidence and family secrets snare Italian child murderer, *The Guardian*, (2016).
- [57] C. Fridman, M.M.S.G. Cardena, F. de Araújo Lima, F. de Toledo Gonçalves, Is it possible to use forensic DNA phenotyping in Brazilian population?, *Forensic Science International: Genetics Supplement Series*, 5 (2015) e378–e380.
- [58] B.J. Koops, M. Schellekens, Forensic DNA phenotyping: regulatory issues, *Colum Sci & Tech L Rev*, 9 (2008) 158-202.
- [59] M. Kayser, Forensic DNA Phenotyping: Predicting human appearance from crime scene material for investigative purposes, *New Trends in Forensic Science Genetics* 18 (2015) 33-48.
- [60] Macmillan Publishers, DNA justice: Germany is considering proposals to extend the use of DNA evidence in criminal cases, *Nature* 543 (2017) 589-590.
- [61] Australian Law Reform Commission, *Essentially Yours--The Protection of Human Genetic Information in Australia*, Volume 1 and Volume 2. Report 96, 2003.
- [62] E. Murphy, *Inside the Cell: The Dark Side of Forensic DNA*, Nation Books, New York NY, 2015.
- [63] K. Dacey, Police hope composite image can help close cold case, in: *WBALTV*, 3 Feb 2016.
- [64] S.J. Walsh, *Evaluating the role and impact of forensic DNA profiling on key areas of the criminal justice system*, 2009.