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

Nathan Scudder[a,c,*], Dennis McNevin[a], Sally F. Kelty[b], Simon J. Walsh[c], James Robertson[a]

[a] National Centre for Forensic Studies, Faculty of Education, Science, Technology and Mathematics, University of Canberra, ACT 2617, Australia
[b] Centre for Applied Psychology, Faculty of Health, University of Canberra, ACT 2617, Australia
[c] Australian Federal Police, GPO Box 401, Canberra, ACT 2601, Australia

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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.

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 modeling 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

* Corresponding author at: National Centre for Forensic Studies, University of Canberra, ACT 2617, Australia.
E-mail address: Nathan.Scudder@canberra.edu.au (N. Scudder).

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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].