



The Evolution of Forensic Genomics: Regulating Massively Parallel Sequencing

Marcus Smith · Seumas Miller

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Abstract Forensic genomics now enables law enforcement agencies to undertake rapid and detailed analysis of suspect samples using a technique known as massively parallel sequencing (MPS), including information such as physical traits, biological ancestry, and medical conditions. This article discusses the implications of MPS and provides ethical analysis, drawing on the concept of joint rights applicable to genomic data, and the concept of collective moral responsibility (understood as joint moral responsibility) that are applicable to law enforcement investigations that utilize genomic data. The widespread and unconstrained use of this technology without appropriate legal protections of individual moral rights and associated accountability mechanisms, could potentially not only involve violations of individual moral rights but also lead to an unacceptable shift in the balance of power between governments and the citizenry. We argue that in light of the rights of victims and the security benefits for society, there is a collective moral responsibility for individuals to submit their DNA to law enforcement and for MPS to be used where other, less invasive techniques are not effective. However, this application should be limited

by legislation, including that any data obtained should be directly relevant to the investigation and should be destroyed at the conclusion of the investigation.

Keywords Forensic genomics · Massively parallel sequencing · DNA evidence · Biogeographical ancestry · Joint rights · Collective responsibility

Introduction

Forensic genomics has evolved over the past decade, enabling law enforcement agencies to undertake more efficient and detailed analysis of suspect samples, using a technique known as massively parallel sequencing (MPS), to include information such as physical traits, biological ancestry, and medical conditions. This article discusses the implications of MPS and provides ethical analysis, drawing on the concept of joint rights applicable to genomic data, and the concept of collective (joint) moral responsibility that are applicable to law enforcement investigations that utilize genomic data. At the society-wide level and based on the achievements of DNA identification over the past thirty years, using MPS in law enforcement investigations is likely to make an important contribution to a society's overall security. However, given the significance of genomic data, the widespread and unconstrained use of this technology without appropriate legal protections of individual moral rights and associated accountability

M. Smith (✉) · S. Miller
Charles Sturt University, 10 Brisbane Avenue,
Canberra ACT 2600, Australia
e-mail: marcussmith@csu.edu.au

S. Miller
e-mail: semiller@csu.edu.au

mechanisms, could potentially not only involve violations of individual moral rights but also lead to an unacceptable shift in the balance of power between governments and the citizenry. While the ethics of the use of DNA evidence and genomic analysis in law enforcement have previously been discussed to a limited extent (see e.g. Miller and Smith 2022, 2021), to our knowledge, this is the first discussion of MPS in the ethics literature.

This first part of the article describes the technology, applications, and issues associated with the use of this technique in law enforcement investigations. The second part discusses the associated ethical and regulatory issues, with a focus on collective (joint) moral responsibility and joint rights. We argue that in light of the rights of victims and the security benefits for society, there is a collective moral responsibility for individuals to submit their DNA to law enforcement and for MPS to be used where other, less invasive techniques are not effective. However, it is important that this application be limited by legislation, including that any data obtained should be directly relevant to the investigation, and should be destroyed at the conclusion of the investigation.

Technology, Applications, and Issues

The application of DNA in forensic investigations has continued to evolve since its first use in the mid-1980s, in conjunction with advancements in genomics and associated science and technology over the past forty years (Butler and Willis 2020; Smith 2016). The initial technique, based on analysis of short tandem repeat markers of repetitive regions of the genome and their frequency in the population, facilitated the identification of suspects by comparing the DNA of two or more people, without revealing further information about the individuals concerned beyond their identity, and the development of large forensic DNA databases, significantly enhancing criminal investigations (Santos, Machado, and Silva 2013).

Genomic technologies are developing rapidly. In the past twenty years, since the mapping of the human genome in 2003, medical research and the field of genomics has provided for a more wholistic analysis of human DNA, and of the genes associated with human traits and diseases, using single-nucleotide polymorphisms, eliciting a vast and increasingly

detailed amount of information, the significance of which continues to be further researched and understood (Scudder, et al. 2019).

The cost and time required to conduct this analysis has continued to decrease dramatically, and it has become increasingly available, to researchers, clinicians, government, the private sector, and direct to the general public, by companies that now hold the genomic data of tens of millions of people. It is important to note that these developments have taken place alongside the rapid increase in the availability of data about individuals more generally, for example, smartphone metadata, online health records, biometric security, and surveillance technologies such as CCTV incorporating biometric facial templates, and a lagging but increasing awareness of, and regulatory focus on, data protection (Smith, Mann, and Urbas 2018; Miller and Smith 2021). A related development is the access and use of genomic data created by direct-to-consumer genomics companies in law enforcement investigations which has been widely discussed in the literature and is illustrated by the “Golden State Killer” case in 2018 (Wickenheiser 2019; Smith and Miller 2021).

Contemporary genomic analysis includes the capacity to determine physical traits (phenotyping), ancestry, familial relationships, ethnicity, inherited diseases, and more detailed analysis of mitochondrial DNA; and to do so more quickly and efficiently than previous techniques (UKNDNAD 2017). It also allows many sections of DNA to be sequenced in parallel, significantly reducing the time required to analyse a sample. The contemporary approach is collectively referred to by different nomenclature, including Next Generation Sequencing, Whole Genome Sequencing, and Massively Parallel Sequencing (Ryan, et al. 2021). MPS is currently the most commonly used in a forensic context and will be the term referred to in this article. MPS involves the analysis of single-nucleotide polymorphisms to provide a range of information beyond identity, which was the case with the traditional DNA profiling based on short tandem repeat sites. The technique is now being used by law enforcement agencies around the world, including the Australian Federal Police (AFP), to assess the physical appearance of suspects. The AFP are “looking to widen the prediction capabilities” and continue to expand the extent of its use in the future (AFP 2021).

Forensically, MPS is faster than traditional profiling, and in addition to identification, provides for biogeographical ancestry, externally visible characteristics, and mitochondrial DNA analysis for intelligence purposes in criminal investigations in cases when a direct match to a known offender on a DNA database is not obtained. However, it can include further analysis to determine medical conditions, such as inherited diseases and other physical or psychological predispositions to the extent provided for by contemporary medical science and diagnostic capabilities (Schneider, Prainsack, and Kayser 2019). In a criminal investigation, there are wide range of externally visible characteristics that can be determined using MPS technology for the purpose of narrowing down a list of suspects. Ancestry, eye and hair colour were the first to be used in a forensic context, and in many cases will be the most pertinent; however scientific advancements mean that many more can now be derived, such as nose width, skin tone, and height (MacLean 2014).

In a criminal investigation, biogeographical ancestry can provide an indication of the likely physical appearance of an unknown suspect, particularly in association with other externally visible characteristics. This can corroborate witness statements or narrow the focus of an investigation in their absence and would have greater value in populations that are more homogenous, for example, where a European suspect is identified in an investigation in Asia (Phillips 2015). While from one perspective this may be considered a form of racial profiling, it can actually help to overcome biased or inaccurate eyewitness testimony or misidentification, which is a leading cause of wrongful conviction (70 percent) (Innocence Project 2023). MPS also enables a detailed analysis of mitochondrial DNA, which has an important application for degraded biological samples due to being present in greater amounts than nuclear DNA (Amorim, Fernandes, and Taveira 2019).¹

Information about a suspect's medical status could also potentially be obtained through MPS. Genetic testing can reveal whether a person has an inherited

disease, whether the individual is a carrier of a gene associated with these conditions that could be passed on to an offspring, or whether they are likely to develop a certain disease, such as breast cancer; with the application of genetics in medical diagnosis and treatment increasing (Claussnitzer 2020). These are far less likely to be relevant to an investigation than the identity or physical appearance of a suspect. So there are ownership and privacy issues arising from information derivable from MPS and used in law enforcement and health contexts. Given the close relationship between individual ownership/privacy rights and autonomy, information derivable from MPS in these contexts also has profound implications for individual autonomy.

The right to privacy is closely related to the more fundamental moral value of autonomy (as is the right to ownership). So much so that we usually refer in this article to privacy/autonomy rights. Roughly speaking, the notion of privacy delimits an informational and observational “space”: the sphere of privacy. However, the right to autonomy consists of a right to decide what to think and do and, of relevance here, the right to control the sphere of privacy and, therefore, to decide *who to exclude and who not to exclude* from it (and the extent of that exclusion) and to control the use to which this data is put by those granted access. (Ownership rights also consist in part in exclusion rights.) So the right to privacy (and the right to ownership) consists in large part of the right of an individual to control—a right of autonomy held against organizations and other individuals—access to, and uses of, personal data including, presumably, genomic data, as well as rights against observation and monitoring of the sphere of privacy. Naturally, as is the case with most if not all moral rights, the right to privacy/autonomy is not absolute; it can be overridden. For instance, it can be overridden by the right to life (Kleinig, et al. 2011).

Since our concern in this article is with MPS in law enforcement investigations, we set aside the ethical and regulatory implications of the use of MPS in health contexts other than, of course, those relevant to law enforcement investigations. We begin by elaborating some of the main ethical and regulatory implications of the use of MPS in law enforcement investigations. These are multiple and somewhat diverse. Accordingly, in the space available to us here we cannot hope to analyse them in detail. Instead, in the

¹ Nuclear DNA is inherited from all ancestors whereas mitochondrial DNA is only inherited from a single (maternal) lineage.

section following this one, we provide ethical analyses of two key ethical or moral (we use these terms interchangeably) concepts that are needed to resolve many of these issues but which have, thus far, received scant attention in the academic literature, namely, joint moral rights and collective moral responsibility (understood as joint moral responsibility).

There are other potential areas of future development that are relevant to MPS that we flag as part of this discussion and bear in mind; however, they have not advanced to a stage where they can be explored in detail:

- As a form of big data, information obtained from genomic analysis has potential for integration with other forms of personal data, such as metadata, facial image templates and financial information to create an increasingly detailed picture of individual lives.
- Law enforcement are already accessing and examining linkages between genomic data from commercial and health databases, a separate regulatory issue from conducting their own MPS analysis of a suspect sample (Smith and Miller 2021).
- As genomic datasets become larger, and the technology advances, artificial intelligence and machine learning techniques are likely to be applied in this area. There are a range of potential issues this could lead to, including bias towards minorities in data sets, the “black box” problem: error and associated legal responsibility, transparency of decisions, and access to, and sharing of, datasets further exacerbating privacy/autonomy issues (Dias and Torkamani 2019; Smith and Heath Jeffery 2020; Miller 2021b).
- Finally, studies over the past thirty years have described a link between the gene for the monoamine oxidase A (MAO-A) enzyme and aggressive and antisocial behaviour, a controversial research field (Smith and Urbas 2012). Though many consider it unlikely, there is the prospect of population screening on this basis, but such an approach would not be supported by current scientific evidence (Tiihonen 2015; Vassos, et al. 2014).

A central issue raised by MPS from an ethical, legal, and regulatory perspective is that the vast majority of legislation regulating DNA profiling

was developed in relation to the initial technique of short tandem repeat matching and does not address the issues and applications described above in relation to single-nucleotide polymorphisms and MPS. However, around the world, a number of jurisdictions (including, in Europe, such as in Germany and the Netherlands) have developed laws relating to externally visible characteristics such as eye and hair colour and prohibited testing for medical/psychological traits (Schneider, et al. 2019).² Despite this, MPS is now widely used around the world in a forensic context—the technology has become widely available and cost effective. Although it is primarily used for “intelligence” purposes and not disclosed publicly or presented as evidence at trial, there are ongoing transparency and rule of law issues in relation to its application to law enforcement.

Ethics and Regulation

The use of MPS in law enforcement contexts can usefully be framed in terms of its potential moral goods (including moral rights protection) and its potential harms (including moral rights infringements). However, the moral good consists of both individual and collective goods, as do the harms. Thus, an individual’s right to life might be protected by the use of MPS in law enforcement investigations; for example, the right to life of a potential victim of a murderer. But such use might infringe another individual’s right to privacy/autonomy; for example, the right to privacy/autonomy of the offender.

At the collective level, the widespread use of MPS in law enforcement investigations may well over time make an important contribution to a society’s overall security (a collective moral good comprised in part in protection of the aggregate rights to life of its members). For instance, the capacity to gain useful information (intelligence/evidence—e.g. a suspect’s appearance) from a sample, that would not otherwise have been possible, would likely lead to increased detection/conviction of offenders (due to the alternatives of no direct match, unusable mixture of DNA,

² For instance, the German Code of Criminal Procedure (StPO) explicitly regulated DNA phenotyping in November 2019.

degraded sample). But such widespread use, if unconstrained by appropriate legal protections of individual moral rights and by accountability mechanisms, might not only involve violations of individual moral rights but also lead to an unacceptable shift in the balance of power between governments and the citizenry and an associated diminution in public trust.

This imbalance of power might result from the overall negative impact on aggregate individual privacy/autonomy, including from unconstrained accessing by law enforcement agencies of information relating to medical conditions and ancestry or data breaches impacting privacy/autonomy. Moreover, this imbalance of power may result in injustices such as profiling on the basis of physical, psychological, or health traits, relatives being unfairly targeted, and the potential for increased data errors. A further likely untoward consequence would be a reduction in public trust especially if MPS continues to be used without appropriate constraining legislation and accountability, including transparency.

Joint Moral Rights

While there are individual privacy/autonomy rights in relation to genomic data, as with other personal data, there are differences. Importantly, rights to genomic data appear to be jointly held rights. How so?

Each of us has a personal identity which enables us to be individuated from other persons (Perry 1985). Moreover, our personal identity is evidently in part based on our individual-specific biological identity. Further the genome of a person is constitutive of that person's individual-specific biological identity. However, that same genome is also *in part* constitutive of the individual-specific biological identities of the person's relatives. Accordingly, persons who are biologically related, such as parents and their biological offspring, have overlapping biological identities. This partially shared biological identity decreases as the degree of relatedness decreases, e.g., a brother shares his biological identity to a greater extent with his sister than with his second cousin.

Let us make the widely shared assumption that each of us have moral rights to our genomic data, given that one's genome is constitutive of one's individual-specific biological identity and, therefore, in part of one's personal identity. It would seem to follow that a brother and his sister, for instance, have

a *joint moral right* to their shared genomic data. So there are joint moral rights to genomic data. This raises the question: What are joint rights? On one influential account of joint rights two or more agents have a joint moral right to some good, G, including epistemic goods such as knowledge or data, under the following conditions (Miller 1999). Firstly, each have an individual moral right to G. Secondly, no-one else has a moral right to G. Thirdly, the individual moral right of each to G is dependent on the individual moral rights of the others to G, i.e., there is interdependence with respect to these individual rights. Thus the individual moral right of one of these persons possessed of this jointly held right to G, has an essential reference to the individual moral right of each of the other persons possessed of this right to G. Fourthly, one cannot unilaterally exercise one's right to G. Thus, in the case of joint rights to data, a person with a jointly held right to the data in question cannot *unilaterally* decide to provide access to this data to those who do not have a jointly held right to it (Miller 2021a).

As is the case with other moral rights to data, the joint right to genomic data includes a right (a joint right in this case) to control access to this data. In the case of the joint right to control one's genomic data, the right is held jointly with one's relatives and, in the case of mitochondrial DNA, with those relatives sharing the same maternal lineage. However, this joint moral right is qualified. It is a qualified joint right by virtue of the fact that even in the case of close relatives, such as a mother and her son, the genomic data of the mother is not identical to that of the son; rather the two sets of data are overlapping. Moreover, in the case of very distant relatives, the degree of overlap might be slight; so slight as to remove the possibility that there was a joint right to the genomic data in question. Accordingly, the question arises as to the degree of overlap necessary to underpin a joint right. It follows from this that joint rights to genomic data have somewhat vague boundaries. So, as is the case with vague boundaries more generally, fixing the exact limits of joint rights to genomic data may involve making somewhat arbitrary decisions.

If the rights to genomic data are joint rights, then a person may not have an exclusive individual right to provide his or her genomic data to consumer genetic testing providers or to law enforcement. Naturally, in the case of offenders convicted of serious crimes

such as murder, there can be moral rights in play, e.g., right to life, that have greater moral weight than moral rights (whether individual or joint) to genomic data. Accordingly, the consent of an individual (let alone the consent of multiple related individuals) to access his or her genome data is not necessarily required, e.g., if the individual is a past offender and hence his or her genomic data in the form of a DNA profile is held in a law enforcement database.³ However, there are other more difficult cases, such as those in which law enforcement needs to rely on the genomic data of relatives known to be innocent, if an offender is to be identified. Specifically, if the relatives in question have a joint right to the data needed by law enforcement then a problem may arise: perhaps *all* of these relatives need to consent to the collection of the genomic data.⁴ This is because a person who provides his or her DNA to law enforcement is, in effect, providing law enforcement with the partially overlapping DNA data of his or her relatives. However, one's moral right to one's DNA data is a right to control access to that data and, in the case of overlapping DNA data, the moral right in question is a joint moral right. So a person does not have a moral right to make a unilateral decision to provide law enforcement with, for instance, his sister's DNA data without her consent, notwithstanding that his and his sister's DNA data overlap. Accordingly, a person does not have a moral right to *unilaterally* provide law enforcement with his or her own genomic data, given that there are others with overlapping DNA profiles. Rather it seems that all (or most) of those possessed of the relevant joint right have to consent.

Of course, joint moral rights can be overridden; as can most, if not all, moral rights. Consider a person who wants to know whether he is vulnerable to a hereditary disease. He has a moral right to know this, and yet his acquiring this knowledge might entail providing his genomic data to health

authorities without the consent of any of his relatives—relatives who have a jointly held right to this genomic data. Arguably, his right to know whether or not he is vulnerable to the hereditary disease overrides the joint right to the genomic data. More germane to our concerns in this article, the joint moral right of members of a family to their genomic data may well be overridden in the context of a murder investigation. In particular, their collective moral responsibility (see next section) to assist law enforcement to bring a murderer to justice may well override their joint moral right to their genomic data. Of course, there is another moral right in play here, namely, the moral right not to self-incriminate. The member of the group who committed the murder has a moral right not to self-incriminate. Arguably, this moral right trumps any obligation to assist law enforcement by providing a sample of one's own DNA. Let us now elaborate on the notion of collective moral responsibility in play here.

Collective Moral Responsibility

Central to collective responsibility is the responsibility arising from joint actions and joint omissions. A joint action can be understood as follows: two or more individuals perform a joint action if each of them intentionally performs his or her individual action but does so with the (true) belief that in so doing each will do their part and they will jointly realize an end which each of them has and which each has interdependently with the others i.e. a collective end (Miller 1999). On this view of collective responsibility as joint responsibility, collective responsibility is ascribed to individuals; moreover, if the joint action in question is morally significant, e.g. by virtue of the collective end being a collective good or a collective harm, then the individuals are collectively *morally* responsible for it. Each member of the group is individually responsible for his or her own contributory action, and (at least in the case of most small-scale joint action) each is also individually (fully or partially) responsible for the aimed at outcome, i.e. the realized collective end, of the joint action. However, each is individually responsible for the realized collective end, *jointly with the others*; hence the conception is relational in character. As already mentioned, if the collective end of the joint action

³ On the other hand, there is the potential collateral damage to the relatives of criminals, given partially overlapping DNA profiles.

⁴ This consent issue adds to other problems that can be raised in relation to direct-to-consumer genetic testing, such as the accuracy of the tests; and in cases where they relate to medical conditions, the fact that the results are not provided in a clinical setting by a healthcare professional to provide the individual with individualized advice on management.

is a collective good or a collective harm, then these individual persons are collectively morally responsible for this good or harm.

Let us now apply this concept of collective moral responsibility to access to genomic information by law enforcement agencies to investigate and prosecute crime. Certainly, there is a collective good to which, let us assume, the use of this information will make a significant contribution, namely, the investigation and prosecution of serious crimes and the prevention of harm and the preservation of the lives of those who may otherwise have been harmed if a serial killer or rapist is not brought to justice as swiftly as possible. Naturally, those whose lives would not have otherwise been preserved receive a moral good, namely, their life, that those who would not have been impacted do not receive. Moreover, crime imposes economic and social costs for society that affect individuals more broadly than those who are directly victimized by crime.

Other things being equal, there is a collective moral responsibility on the part of members of the state to submit their DNA to assist law enforcement investigations in certain circumstances. Of course, other things might not be equal. Some person or persons likely has privacy/autonomy rights to the data. Moreover, the data might be used to prosecute the person being asked to make it available in which case the moral right not to self-incriminate is in play. However, there is a collective moral responsibility of joint rights holders of DNA to provide this DNA to law enforcement, at least in the case of serious crimes. That is, their joint moral right is overridden by their collective moral responsibility. However, this collective moral responsibility applies in specific cases on a piecemeal basis; it is not a collective moral responsibility to provide their DNA data in a manner that contributes, for instance, to MPS being undertaken on an entire population of a country, justified by the potential law enforcement benefits. Moreover, it is not a collective moral responsibility to provide their DNA data on a permanent basis. Rather they have a joint moral right that the data be destroyed upon the conclusion of the specific criminal investigation and associated trial.

Conclusion

The use of MPS is an important development in modern law enforcement investigations that offers great potential to improve detection and prosecution of serious crimes but is also associated with significant ethical implications. In addition to enabling many sections of DNA to be sequenced in parallel, reducing the time required to analyse a sample, MPS integrates analysis to determine a suspect's physical traits, ancestry, familial relationships, ethnicity, inherited diseases, and more detailed analysis of mitochondrial DNA.

There are privacy/autonomy implications associated with MPS, such as analysis to determine externally visible characteristics and medical conditions. As is the case with most, if not all moral rights, the right to privacy/autonomy is not absolute; it can be overridden. For instance, it can be overridden by the right to life, which may be relevant to the investigation of serious offences. We accept that this form of analysis could be justified for serious investigations such as murder but note that it should nonetheless be regulated by legislation, given the sensitivity of this data, particularly in relation to traditional forms of DNA identification that provide only an indication of identity, rather than detailed information about an individual.

The use of MPS to undertake biogeographical ancestry screening highlights the shared nature of genomic data and the associated joint rights of a sample donor/suspect's relatives, adding a further element of complexity.

There is a collective good in allowing law enforcement to undertake this form of analysis and investigation: the investigation and prosecution of serious crimes and the prevention of harm in bringing an offender to justice as efficiently as available scientific techniques allow. We argue that there is a collective moral responsibility of joint rights holders of DNA to provide their DNA to law enforcement in relation to the investigation of serious crimes because their joint moral right is overridden by their collective moral responsibility. However, this application should be regulated by legislation, including that any data obtained should be directly relevant to the investigation, and should be destroyed at its conclusion. MPS technology should

not be used to create population wide databases accessible for law enforcement purposes.

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