Policy required for entry of DNA profiles onto the National Forensic DNA Database of South Africa

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DATES:
Received: 10 Dec. 2013
Revised: 26 Feb. 2014
Accepted: 03 Mar. 2014

KEYWORDS:
DNA profiling; chromosomal abnormalities; medical confidentiality; forensic science

HOW TO CITE:
Heathfield LJ. Policy required for entry of DNA profiles onto the National Forensic DNA Database of South Africa. S Afr J Sci. 2014;110(7/8), Art. #2013-0374, 3 pages. http://dx.doi.org/10.1590/sajs.2014/20130374

The recent *Criminal Law (Forensic Procedures) Amendment Act (2013)* provides a definition for forensic DNA profiles and, in so doing, states that medical information about an individual may not be revealed through a forensic DNA profile. Yet chromosomal abnormalities can exhibit as tri-allelic patterns on DNA profiles and such information can expose medical conditions such as Down syndrome. This short report highlights this concern and suggests a policy be created for the entering of such DNA profiles onto the National Forensic DNA database of South Africa.

In South Africa, the use of DNA within a forensic context has largely been accepted as the gold standard of evidence for human identification. The value of DNA in forensic science lies both in its objectivity as well as in its highly discriminatory nature, and it currently serves a pivotal role in the courtroom.[1,2] A recent case from the Supreme Court of Appeal, *S v SMM* 2013 (2) SACR 292 (SCA), provides a classic example: the offender who was sentenced to life for the rape of his 13-year-old niece appealed against both his conviction and sentence, but the appeal against conviction was dismissed immediately, as the DNA testimony was sufficient evidence of penetration.[3]

The passing of the *Criminal Law (Forensic Procedures) Amendment Act (2013)* signified the satisfying culmination of a persevering journey by the forensic community in South Africa. The 'DNA Act' amends several other Acts in terms of collecting and retaining bodily samples for forensic identification and intelligence purposes; it also provides regulations with regard to the formation and maintenance of the National Forensic DNA Database of South Africa (NFDD).[4] This very necessary section of legislation serves to fill the gap in terms of allowing current forensic methodologies to be used to their full potential, especially in an era in which the need for applied innovation and technology is so crucial.

DNA profiling is a method used in forensic science to aid identification of potential suspects as well as to link scenes of crime together. It relies on the amplification and subsequent sizing of specific repetitive markers within DNA to generate a DNA profile – essentially a set of numbers which, when statistically analysed, can be highly discriminatory between individuals.[5] Various commercial kits are available. South Africa currently employs the AmpFlSTR Profiler Plus™ PCR kit which analyses 10 markers, including Amelogenin, the sex determining marker, although we are in the process of upgrading to the AmpFlSTR Identifiler Plus™ PCR kit, which analyses 16 markers including Amelogenin.[2,6] Within the forensic biology unit at the South African Police Service, DNA profiling is carried out according to standard operating procedures and quality checks are performed, especially during the interpretation and reporting phases. Once a profile has been generated, interpreted and reported, it is committed to the NFDD through software called STRlab™.

Clause 15E (l) in chapter 5B of the DNA Act, defines a forensic DNA profile as

> *…the results obtained from forensic DNA analysis on bodily samples taken from a person or a crime scene, providing a unique string of alpha numeric characters to provide identity reference: Provided that it does not contain any information on the health or medical condition or any information on the predisposition or physical information of that person other than the sex of that person.*[4]

Clause 15G (5) of the same chapter specifies that the indices on the NFDD

> *…shall not contain the following information derived from a bodily sample which was taken from a person:*
>
> *(a) The appearance of the person, other than indicating the sex;*
>
> *(b) medical information of the person;*
>
> *(c) historical information relating to the person; and*
>
> *(d) behavioural information of the person.*[4]

Therefore, a DNA profile and the NFDD may not contain details regarding the individual's appearance (excepting sex) nor any medical, historical or behavioural information. However, many of the markers initially identified for DNA profiling came from the Cooperative Human Linkage Center (CHLC) (http://www.chlc.org) which forms the basis of genome scans used for genetic linkage studies.[7]

Numerous association studies have been carried out, with one resulting in the exclusion of the HumARA marker in DNA profiling kits because of its direct association with spinal and bulbar muscular dystrophy.[8] Yet other studies have given rise to a set of complex, and at times contradictory, results[9,10], specifically with regard to the TH01 marker[11–15]. Kimpton et al.[16], working with the European DNA Profiling Group, prematurely acknowledged that

> *it is likely that many or possibly most* [forensic markers] *will eventually be shown to be useful in following a genetic disease or other genetic trait within a family and therefore*

> *this possibility must be recognised at the outset of the use of such systems.*[16]

Although forensic markers could prove to be linked with genetic disease, such associations are not blatantly obvious upon DNA profile interpretation. What is far more striking is the notion of chromosomal abnormalities. For example, D21S11 on chromosome 21 is an eligible marker which can be used to test for trisomy-21 (Down syndrome)[17,18] and the D21S11 marker is included in the DNA profiling kit used in South Africa. Similarly, D18S51, which is also a forensic marker, can be used to test for trisomy-18 (Edward syndrome).[19] Down and Edward syndromes could also occur via mechanisms other than a third chromosome (trisomy), such as copy number variation or duplication of a part of a chromosome, which could yield varying levels of pathogenicity. DNA profiling, however, is capable of revealing tri-allelic patterns at any of its markers[20–22] and thus could expose potential chromosomal abnormalities during the interpretation of results. Although further testing would be required to confirm Down or Edward syndromes – which is not performed in routine DNA profiling – a tri-allelic pattern at the D21S11 or D18S51 marker would certainly be suggestive of these syndromes. Not only can these DNA profiles reveal potentially medically sensitive information, they can simply be committed to the NFDD via STRlab™.

Although these markers are included in several DNA profiling kits globally, it is not necessarily statutory in other countries that DNA profiles may not contain medically sensitive information, as is the case in South Africa. In the United Kingdom, for example, the D21S11 marker is analysed and Down syndrome may be revealed; however, ethical guidelines exist to regulate the process of entering medically sensitive information onto the database to protect data of this nature.[23,24] Alongside the European Network of Forensic Science Institutes, the National DNA Database Strategy Board and Working Group stipulate guidelines specific to the United Kingdom for submitting DNA profiles onto its National DNA Database. Linked to this, an ethics group was formed in 2007 to independently offer advice on database operations.[25] For example, when the confirmed results of a rare allele or tri-allelic pattern are submitted to the database, the locus is given a failed designation (F, F), ensuring that a potential medical condition is kept confidential. Although this method favours the exclusion of a marker's information which could possibly increase statistical probabilities, it nevertheless guarantees medical confidentiality, even if the tri-allelic pattern was a non-pathogenic mutation or anomaly. Surely a similar policy or set of guidelines should be implemented in South Africa to protect medical confidentiality? If not, does our current DNA profiling system meet the requirements set out by the law?

The very definition of a forensic DNA profile provided in the DNA Act (clause 15E (l)) is refuted by the fact that chromosomal abnormalities can be detected using various DNA profiling kits and thus medical conditions can potentially be exposed. Furthermore, clause 15G (5) is also contested as there is no formal policy regulating the data input to the NFDD. Changing the kit used for DNA profiling is not necessarily the answer to create consistency between DNA profiling methods and the law, because using different markers will not only be a costly and timely expense, but also decrease the potential or statistical significance of matches with individuals who have been profiled on the current system.[26] Furthermore, using different markers could limit the potential of cross-country comparisons which could have a significant impact on loss of evidence as well as costs. Similarly, discounting the clause relating to medically sensitive information not being stored on a database would be in opposition to human rights. Rather, the definition of a DNA profile should be altered and a policy be implemented to regulate the process of entering data on the NFDD. These are concerns which the National Forensic Oversight and Ethics Board (clauses 15V and 15Z) could possibly address during their oversight of the ethical, legal and social implications of the NFDD. In addition, access to the NFDD should also be defined.

While the passing of the DNA Act is a necessary step to fight crime in South Africa, legislation surrounding it should enable the database to operate at its full potential, and as Morris succinctly stated in a recent editorial in this journal, 'not everyone is happy'[27]. We need to acknowledge and accept that forensic DNA markers may be associated with medical conditions, especially chromosomal abnormalities, but this association does not need to detract from the optimal utilisation of the NFDD. A solution in the form of amending the definition of a forensic DNA profile as well as introducing a policy for entry of DNA profiles onto the NFDD would suitably satisfy human rights activists and the forensic community alike. In a nation where crime and recidivism rates are extremely high, the forensic DNA process needs to be a foolproof system, commensurate with international quality standards and compliant with South African law.

Acknowledgement

The author acknowledges Rosalind Elphick for her input.

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