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HOW INCONTOVERTIBLE IS THE EVIDENCE OF A DNA PROFILE MATCH PRESENTED BY AN EXPERT IN A CRIMINAL TRIAL?

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ABSTRACT

Miscarriage of justice and the increased expectations the public have of DNA (Deoxyribonucleic Acid) evidence have both contributed to the recent questioning of the accuracy of DNA evidence when presented at court. While forensic DNA profiling is generally accepted as a scientifically valid method, human error and misinterpretations of what a DNA profile match means can be detrimental to the accuracy of that DNA evidence. This literature review provides a topline view of the scientific process of DNA profiling and addresses some of the advances made to assist forensic scientists when DNA is degraded such as mini short random repeat (STR) typing and single nucleotide polymorphisms (SNPs).

To further assess potential errors, the challenges in Forensic DNA profiling as a broad field beyond criminal investigations are considered. Common and uncommon problem sources such as contamination, handling of DNA evidence and DNA planting at crime scenes are discussed. When the science is correct the misinterpretation of the statistical analysis of a DNA profile match can undo all the good work at a crime scene and laboratory. This is identified in the review of the literature on interpretation issues such as calculating probability and a phenomenon known as the CSI (Crime Scene Investigation) effect.

The findings of this review show that a DNA profile match cannot be absolutely incontrovertible based on genetic science as it stands. How incontrovertible the presentation of a DNA profile match as evidence given by an expert in a criminal trial can vary. The degree by which it varies can only increase as technology and scientific methodology advance.
1 INTRODUCTION

DNA (Deoxyribonucleic Acid) evidence should be highly discriminating when assisting the judgment of a person’s innocence or guilt. However if it is not treated responsibly in practice, the consequences could be dire. Such consequences include wrongful conviction, wrongful acquittal and miscarriage of justice (Griffith & Roth, 2006). The effects of questionable forensic DNA profiling are felt in the courtroom and ripple into the public arena, therefore the importance of sound scientific understanding and practice of forensic scientists to decrease the degree of dispute around DNA evidence is paramount.

As is natural in the field of forensic science, the key stakeholders in this controversy belong to either science or law. Law will always strive for incontrovertible evidence and it is the responsibility of science to manage expectations of the degree by which DNA evidence is incontrovertible. Walsh et al (2009) discussed how the forensic science community should be guiding the legal community in utilising DNA profiling to enhance intelligence. At a broader level, this is imperative to maintain the credibility of the field of forensic science given the recent questioning of whether it is a true science as discussed by Crispino et al (2011) and Roux (2011). Penacino et al (2003) recognise that “since its beginnings, DNA was surrounded by an aura of infallibility” but in questioning this infallibility conclude that from a scientific perspective, “no amount of care can eliminate the possibility of error”. As such, this review was approached with the hypothesis that DNA evidence is not incontrovertible. However if the errors that jeopardise the accuracy of DNA evidence can be identified and minimised or eliminated by those who yield the power of DNA evidence, the degree by which DNA evidence is questioned should decrease.

2 THE SCIENCE OF DNA PROFILING

2.1 THE FUNDAMENTALS OF FORENSIC DNA PROFILING

The purpose of forensic DNA profiling is to investigate whether there is a match between two samples of DNA. One sample will come from the crime scene and the second sample will be from a suspect or a victim. DNA samples can be recovered from a crime scene from almost any biological material. Biological material can be recovered from bedding (Petricevic et al., 2005) or from shoe insoles (Bright and Petricevic 2004) for example.
2.1.1 STANDARD TECHNIQUES
CrimTrac (2011) detailed the DNA profiling process. DNA can be extracted from biological material using isolation techniques. DNA quantitation by spectrophotometry methods then ascertains how much DNA material is present in the sample. This allows the forensic scientist to decide which DNA profiling technique to use to optimise the use of the evidence.

Carey and Mitnik (2002) described how the DNA molecule features sections where nucleotides are repeated in tandem. The number of repeats varies from person to person. It is estimated there is a variance of one nucleotide per 300-1000 base pairs (Scott 2010, p254-255). When Cary and Mitnik conducted their review, the most commonly used methodology for profiling was short tandem repeat (STR) typing.

STR typing is most commonly done using polymerase chain reaction (PCR) amplification of non-coding lengths of the DNA molecule by counting the number of short tandem repeats (STRs) that appear at a set number of loci (10) found in these sections of DNA. If the number of STRs at each locus is the same for each profile, the comparison is a match (Griffith & Roth 2006).

2.1.2 NON STANDARD TECHNIQUES
Where degraded DNA samples are the only available option, Hughes-Stamm et al (2011) have shown that mini-STRs and single nucleotide polymorphisms (SNPs) are more successful in producing complete profiles than standard DNA profiling techniques. They artificially degraded nuclear DNA extracted from whole blood. The samples were profiled using STR, mini-STRs and SNPs methods. In all cases, the mini-STR and SNP methods were more successful in providing a correct profile (figure 1).

Crimtrac (2011) also suggested profiling mitochondrial DNA (mtDNA) as an alternative but note that there are many challenges associated with mtDNA. For example, mtDNA is highly susceptible to contamination, it is time consuming, mtDNA is not as discriminating as nucleic DNA and seemingly unrelated individuals may share the same mtDNA because of a common female ancestor.
2.1.3 Techniques developed from other applications of DNA profiling

Y-chromosome profiling or Y-STR typing is a technique used in genetic relationship testing and in particular, paternity testing. Carey & Mitnik (2002) discussed the increasing trend for this application of DNA profiling to be used in forensic casework. In one sexual assault case involving 5 perpetrators, two suspects were found to be related via Y-STR typing. They also found Y-STR typing to be highly sensitive in female/male mixed samples because additional female DNA STRs fall outside the range of the loci used in the interpretation of Y-STR typing. They concluded that Y-chromosome profiling would be a “promising technique” if applied to forensic casework.

DNA profiling is important in the application of disaster victim identification (DVI). After the attacks on the World Trade Centre in 2001, Budimlija et al. (2003) profiled over 500 tissue samples using the standard PCR and STR typing techniques described above. While 75% of samples produced accurate profiles, the remaining samples were highly degraded due to the extreme conditions they were exposed to. They also contained mixed profiles due to the high incidence of co-mingling of remains. As a result, they had to call upon a team of forensic anthropologists to collaborate. Anthropologists were able to group tissue samples from the same body parts and this allowed further DNA profiling to be performed. New victims were
uncovered and matched tissue samples identified. Their challenge was overcome through the corroboration of DNA evidence with other evidence.

While the focus of this review is the application of forensic DNA profiling in criminal investigations, we have seen from the above applications of DNA profiling that the reliability of DNA evidence could be improved. Corroboration of evidence as demonstrated by Budimlija et al (2003) and the value of alternative techniques as demonstrated by Carey and Mitnik (2002) could be transferrable to criminal investigations.

3 ERRORS ENCOUNTERED IN FORENSIC DNA PROFILING

If a DNA profile from a crime scene matches the DNA profile of a suspect, “this is strong evidence that that they came form the same source but it is not conclusive” (Griffith and Roth 2006) and there is only a small chance that they come from two different people (Crimtrac, 2011). However, as the literature fervently debated, there was a lot that can affect this “strong evidence” and “small chance”.

In Lawyer Alecia Simmonds’s (2007) opinion piece in The Sydney Morning Herald, she argued that DNA is fallible because of factors such as human error, cross-contamination and contamination, the probability of a chance match and the possibility that DNA evidence has been planted. She was not alone. In Griffith and Roth’s (2006) parliamentary briefing paper, the “reliability of DNA evidence can be affected by contamination, lab error and planting.” In their quest to answer the question of whether DNA tests are infallible, Penacino et al (2003) identified potential errors made when collecting and transporting DNA evidence from the crime scene and into lab storage, contamination, scientific error and the interpretation of results. Jiang et al (2010) exposed the freak occurrence of false indirect exclusions in the process of DNA profiling as a result of the failure of the scientific equipment used. Some errors were human errors, some were scientific errors and some errors existed because of the influence of subjective thought in the interpretation of DNA evidence.

3.1 THE INFLUENCE OF HUMAN ERROR IN SCIENTIFIC PRACTICE

3.1.1 CONTAMINATION
An example of an Australian miscarriage of justice in relation to DNA evidence was the Jama case. This case was discussed by both Rayment (2010) and Scott (2010). The case involved the conviction of Mr Farah Abdulkadir Jama, of rape based on what was found to be a
contaminated DNA sample. The contamination occurred at the hospital at which the victim was swabbed. A sample was taken from a different woman who had engaged in sexual activity with Mr. Jama. This earlier sample was taken for a completely unrelated reason at the same hospital, by the same doctor in the same room on the previous day. Unfortunately, Jama’s biological material had stuck around and contaminated the sample taken from the alleged rape victim.

In her article in New Scientist, Geddes (2012) referred to two Australian studies (Goraya et al, 2011) and one German study (Schwark et al, 2011). They all have shown how easy it can be for DNA contamination or DNA transfer to occur.

3.1.2 Handling of DNA evidence

Penacino, et al (2003) made the recommendation that maintaining a chain of custody could help track any errors that occur between the collection of DNA evidence at the crime scene and what happens to DNA samples at the lab. In the process they described, samples should be properly sealed to prevent transfer and contamination. They also discussed the importance of preventing contamination while working with a DNA sample and suggested that “working in different area(s) pre and post amplification” is good lab procedure to minimise contamination risk. They also discussed the benefits of proficiency testing and quality controls.

3.1.3 Planting of evidence

As mentioned, the planting of DNA evidence is of concern when considering the degree by which DNA evidence can be incontrovertible. In Graham Philips’s 2004 report called “DNA Doubt” on Catalyst, a science program on the ABC (Australian Broadcasting Corporation), Biotechnologist Dr David Berryman and former DNA Detective Robin Napper proved that this is a problem. They carried out an experiment whereby an innocent person was successfully framed by planting DNA evidence. However Director of the National Institute of Forensic Science Dr. Tony Raymond was a little dubious of the reality of this scenario and was of the opinion that most criminals are not smart enough to plant DNA evidence.

3.1.4 Misbehaving equipment

On one hand, some believed that the majority of errors that jeopardise the degree by which DNA evidence is incontrovertible were due to human involvement. Hurth et al (2010) suggested that we need to eliminate human intervention in support of their automated instrument for human STR identification. In the interest of cost and time efficiencies, others were excited about the development of hand PCR amplification devices. These devices would allow crime scene investigators to run a DNA profile while on the scene (Liu et al, 2008).
On the other hand, is it really safe to leave so much up to a machine? The case report by Jiang et al (2010) identified an error that occurred as a result of an Identifier kit failing to amplify one allele to see the existence of another. This resulted in a false negative on a maternity test. In addition to allele drop-out, Penacino et al (2003) also referred to problems such as spurious signals and mutations, which might not otherwise be picked up if DNA profiling is completely automated.

3.1.5 Education, expertise & training

In Jennifer Joan Raymond’s 2010 thesis, she recognised the need for a more holistic approach when it comes to DNA evidence and that this would be useful in the application to volume crimes such as theft. As a result of her study she identified a lack of training and proficiency testing in trace DNA cases.

Not only was training and expertise important in increasing the accuracy of DNA evidence, it seemed common sense to Scott (2010) to declare that DNA testing laboratories should also be subject to strict accreditation audits. He also pointed out that the use of industry standards should also be commonplace.

In light of potential human errors found in the literature, it should also be highlighted that the forensic science community will always be trying to improve the accuracy, efficiency and credibility of DNA profiling. A demonstration of this was the work of Griffiths et al (2011) and their work in increasing the accuracy of PCR amplification.

3.2 Inference and interpretation by experts

DNA evidence could be considered ‘circumstantial evidence’ because an inference is required to connect it to fact (CSIRO, 2012). Where there is uncertainty, there is subjective thought and this can vary the degree by which DNA evidence is incontrovertible, regardless of how good the science is.

3.2.1 Contextual bias

Roux (2011) described an example of contextual bias. He was working on a case in which the suspect was suspected of firing a gun. To test this hypothesis, the suspect’s hands were swabbed and tested for gunshot residue (GSR). The test result was positive. It was unknown at that point that the suspect had recently removed the number plates from his car, which would cover his hands with residue of the same elemental components as GSR. Until this was known, it was presumed that the suspect had fired the gun, when he had not. Contextual bias could similarly be applied to DNA evidence.
3.2.2 Calculating probability
The case of Forbes v The Queen was described by Scott (2010). It involved an unlawful sexual assault where a young girl was accosted in a dark street by a man and forced upon him. DNA was found on the inside and outside of the girl’s underwear. The defence had claimed that it was not absolute that it was the defendant’s DNA that was found on the victim. Scott explains that the forensic expert said it is impossible to prove the uniqueness of the defendant’s profile, as this would require the DNA of every person on the planet to be profiled. Secondly, the STR technique profiled only 10 loci on the DNA molecule and not the whole DNA molecule. The length and complexity of the human genome makes this impractical. The forensic expert was correct in justifying the analysis and interpretation of the DNA profile results, however it is evidence that could decrease the degree by which DNA evidence is incontrovertible.

In 1997, Chambers et al discussed the difficulties of absolute identification alongside quality of population data, genetic differentiation within a population and how this affects the calculation of probabilities. They herald the use of a likelihood ratio to express the probabilities of contact.

Thompson and Schumann named the Prosecutor fallacy and the Defence attorney fallacy in 1987. The Prosecutor fallacy occurs when the probability of the results (e.g. a DNA match) given the hypothesis (e.g. Mr. X was present) is mistaken for the probability of the hypothesis, given the results. The defence attorney fallacy occurs when no weight is given to the evidence as a result of statistical analysis. These dangers often present themselves in court.

Nance and Morris (2005) discussed how expert testimony could affect the willingness of a jury to convict based on the presentation of a random match probability (RMP). They carried out research that concluded that jurors tend to undervalue evidence that is explained using an RMP and that the degree by which they underestimate is determined by whether or not the RMP calculations are presented to them.

3.3 The “CSI effect”
In recent years, a phenomenon called the CSI (Crime Scene Investigation) effect has been touted about the media. The research of Brewer and Ley (2009) indicated that portrayals of DNA evidence in entertainment and media could predict the views that the public has about DNA evidence. In Laura Huey’s 2010 study, she noted that no empirical studies have been done to measure the CSI effect. In her research it was found that the public expectation of forensic science is high and unrealistic as a result of these shows. This high expectation, when
applied to DNA evidence in court, can be detrimental to the interpretation of DNA evidence by jury, who perceive DNA evidence to have a high degree of incontrovertibility.

Further to this, Haesler (2011) discussed the probabilistic interpretation of other Australian cases. In doing so, he prescribes direction to both the legal system and scientific experts alike when presenting statistical analysis of DNA profile results to the court.

After reviewing all the factors that could invalidate DNA evidence, it was worth noting that in comparison of individualisation evidence (latent fingerprinting, incriminating images, DNA profiling and incriminating voice recordings) in the eyes of the Australian legal system, DNA profiling is the only evidence that is scientifically validated (Edmond, 2011). So of all the individualisation evidence that a forensic scientist can bring to court, DNA evidence could be considered the most incontrovertible.

4 CONCLUSION

The incontrovertibility of DNA evidence has been shown to be a debate for both science and law. The onus is on science however to optimise and further develop scientific methodology to collect, process, analyse and interpret DNA evidence. Action on these responsibilities is evident in the literature, not only in the use of Forensic DNA in criminal investigations, but in other applications of Forensic DNA profiling.

Where human error was of detriment to the degree by which DNA evidence is incontrovertible, it was recommended by the literature that universal processes and the strict enforcement of standards and procedures should be implemented. Where misinterpretations of DNA evidence are of issue, the presentation of DNA evidence to criminal courts by forensic scientists will need to be free from contextual bias, and will be greatly improved through the adoption of holistic training for forensic scientists. Checklists against prosecutor’s fallacy and defence attourney’s fallacy will also minimise the risk of a misinterpretation of DNA evidence. These considerations will all increase the degree by which a DNA profile match presented by an expert in a criminal trial is incontrovertible.

Until absolute human DNA individualisation can be proven by science, a DNA profile match presented by an expert in court cannot by incontrovertible, however as techniques and methodology improve, the degree by which a DNA profile match presented by an expert in a criminal trial is incontrovertible, is increased.
5 REFERENCES


Huey L. 2010. 'I've seen this on CSI': Criminal investigators' perceptions about the management of public expectations in the field. *Crime, Media, Culture* 6 49-68


