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DNA and the Courts – An Immiscible Solution?

by

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This paper addresses some of the critical issues that have confused the issues of DNA evidence in recent court cases. This paper is not about DNA per se, although the paper contains a brief history of the introduction of DNA into forensic science.

Background

From a scientific viewpoint, the development of DNA evidence served the basic forensic needs in the following way:

1. DNA provided the discrimination power that the courts required but ABO blood grouping could not provide.
2. The courts required a statistical outcome regardless of the practicalities of the situation.
3. Science delivered!
4. The courts now continually challenge the very foundations of DNA technology, the results obtained and the statistical meaning and validity of the evidence.

As a consequence, the following questions must be asked:

Where are we now? How long will the scientists be able to give to the Courts? Perhaps more importantly, will science continue to want to provide DNA evidence?

The application of science for forensic purposes is fundamentally one of reconstruction and identification, with a view to answering the following: who was involved?, what did happen?, when did it occur?, where did it occur?, and how did it happen? It is not necessarily concerned with why (motive) the crime occurred but scientific information can also assist here. The task of a forensic scientist is to analyse evidence to assist the police and the courts to establish facts, provide opinions and, when necessary, to advise on the capabilities and limitations of the techniques used.

The analysis of deoxyribonucleic acid (DNA) has become established in forensic science in a relatively short period. Furthermore, the development of polymerase chain reaction (PCR)

technology has effectively revolutionized molecular biology and had a profound impact on DNA profiling.

Until recently, forensic scientists were applying ABO blood grouping to biological fluids. The classical ABO blood group system was used for many years but its use was limited to excluding an individual with very little inclusion value. By the early 1970's, the development of more specific genetic typing techniques, utilising other polymorphic protein and enzyme markers, such as haptoglobins (Hp), group specific protein (Gc), phosphoglucomutase (PGM), etc, allowed the forensic scientists to use genetic markers to more specifically identify the origin of an evidentiary stain.

The significance of the genetic markers is dependent on the fact that all genetic markers occur with a given frequency in any population. For example, ABO blood group A occurs in approximately 35 - 40% of the caucasian population. Clearly, the point of genetic typing is to reduce the frequency of the typing to the smallest number possible, that is, individualise the donor and, generally, to express the significance of genetic concordance. To achieve this the individual frequencies of the different markers can be cross-multiplied. However, this calculation is only acceptable if the genetic markers pass certain criteria, the most important being that the genetic markers must show no dependence between the loci or within the locus where they occur.

By design, forensic DNA typing is much more powerful than the traditional typing of biological material. However, while irrefutable results could be obtained by analysis of a complete DNA strand, the entire DNA code is too large to completely analyse and only a small portion of the human genome is targeted and included in the biological examination. This selection is achieved through the use of DNA typing technology and is known as PCR.

PCR Technology

The labeling or isolating of specific regions of DNA is commonly referred to as PCR and is achieved using specific primer sequences. Once a specific region is isolated/tagged, a process is applied that amplifies or copies these small regions of DNA millions of times. At the amplification stage the specifically designed primers attach to the alleles or DNA fragments.

Once the primers attach to the DNA fragment, they remain attached throughout the entire processes, including the detection stage.

There has been much discussion [refer R v Karger (SA, Supreme Court, 2000/01) and R v Kami (NSW, Supreme Court, 2001)] regarding the validation of DNA typing systems and the perceived requirement by Defence scientists that the primer sequences of the system must be made known to them. Accreditation requirements are for a two stage validation, i.e. validation at the developmental stage (manufacture) and validation by a laboratory before using the new technology. Primer sequences are not required for a laboratory to validate the system. Manufacturers validations are generally published and can be readily accessed. However, the Profiler Plus™ system is an exception. The Profiler Plus™ system (see below) uses unspecified primer sequences but, recently, has made the sequences available if required by the courts under a special agreement.

It should be noted that Profiler Plus™ has been constantly and regularly verified through internal case audits, proficiency testing and re-examination of cases from other jurisdictions without knowing or resorting to the primer sequences.

Profiler Plus™

Profiler Plus™, marketed by Applied Biosystems, is one of the more preferred genotyping kits that are commercially available for forensic application. The advantages gained through its use are that it is a multiplex system, typing ten (10) short tandem repeat (STR) loci (including a sex determinant) in a single analysis and it has a discriminating power in the order of 10^8. Profiler Plus™ is a fully verified, reputable DNA system that has gained acceptance by the scientific community and, contrary to allegations made in R v Karger (SA, Supreme Court, 2000/01), it is recognised by laboratories in 28 countries.

Included in the users are the Federal Bureau of Investigation (FBI), Royal Canadian Mounted Police (RCMP) and members of the European Network of Forensic Science Institutes (ENFSI).

Although the conditions of use for Profiler Plus™ have been well documented, scientific investigations have shown that the technique has a sensitivity that exceeds that stated by Applied Biosystems. It is a very robust and resilient system that can be used successfully for

lower levels of DNA than previously recorded and, on occasions, the Northern Territory has been able to enhance otherwise unreportable results for exclusion and inclusion purposes. Through necessity, other states are now studying and applying Profiler Plus™ to low level DNA.

Statistics and DNA

Because the entire DNA profile is not examined in forensic science casework, it is necessary to apply a statistical analysis to any result. In this way information can be provided concerning the "degree of uniqueness" of any DNA profile. To do this, population data must be generated and representative DNA databases have now been compiled by all jurisdictions to provide the statistical basis of opinions presented.

The requirements of the profiling systems chosen for forensic DNA analysis demand that the "degree of uniqueness" will be enormous and this is certainly the case. Most DNA profiles can only occur in one person in tens of billions and the most common expected profile, according to a statistical calculation, exceeds one in 360 million.

However, while the introduction of the science of statistics does provide a scientifically sound basis for the interpretation of DNA evidence, it also introduces an opportunity for multiple interpretations and a range of conclusions.

Now, as previously stated, statistics are used to give a weighting to the evidence for the assistance of the courts. The use of either the allele or the genotype frequency for this purpose is determined by the jurisdiction, for example, the NT calculates the relative frequency of a complete DNA profile using genotype frequency. Statistical results are then reported very conservatively, in the following way:

> "It is 99.9% probable that the true relative frequency of any DNA profile identified, at the nine loci tested, is rarer than ***one in 200 million*** in the general population (i.e. 1 in approximately 10 × the population of Australia)."

The figure of ***one in 200 million*** is based on the fact that it can be shown statistically, that the relative frequency of the most commonly expected DNA profile in the Northern Territory database is estimated to be ***one in 368 million***. Based on this figure it is predictable that this

DNA profile has not yet been seen within the Northern Territory database and probably never will be. Now, by halving (approximately) this figure and quoting ***one in 200 million***, a conservative but fair figure is provided that most people can relate to particularly as this approach represents 10 times the Australian population. In this way, effectively, the statistics have been taken out of the equation.

Other laboratories report figures 10^{10} or larger that are meaningless to most people and have, in some cases, been criticised by judges as being prejudicial.

Perhaps the most confused and misunderstood feature of the Northern Territory approach is the inclusion of the 99.9% factor known as a 'Confidence Interval (C.I.)'. This is a statistical feature necessary to incorporate the limited sample size of the database.

It can not be overemphasized that it does not indicate an error. It is a consequence of applying statistics and it simply means that there is less than 0.1% chance that the DNA profile is more common than ***one in 200 million*** in the general population.

The application of statistics is more complicated in the case of mixed DNA profiles, where the allele frequencies are used to calculate the likelihood ratio. This is the ratio of the probability that the DNA evidence came from the suspect to the probability that the DNA evidence came from a random person. This is a necessary statistical adjunct to this type of scientific result but, again, its inclusion does significantly complicate the results and associated opinions

Database Legitimacy

The use of a database is only as legitimate as the database itself and therefore database integrity must be checked periodically. The Northern Territory database and the Profiler Plus™ data have been validated and verified by Associate Professor Janet Chaseling, a statistician from Griffith University.

All the data has been checked for dependencies with the result that very little meaningful dependency has been identified. This is also the case with databases from other jurisdictions in Australia.

There have been arguments both in scientific circles and in the courts, that Hardy-Weinberg equilibrium should be implied in all cases. However, an understanding of population trends show that it is not legitimate to assume Hardy-Weinberg equilibrium in population databases, primarily because the databases include several generations within the population and the populations are not infinite in size and nor is mating random. Notwithstanding this social development, the use of the product rule to calculate the genetic concordance is still valid because, as previously stated, no significant dependencies have been found to exist in the databases. When the databases have been compared it has been found that each State and Territory has a similar distribution of alleles within the caucasian and declared aboriginal databases and therefore "local population" statistical arguments are not valid. That is, the quoted statistics are genuine, regardless of the population origin. Many of these issues are no longer critical because of the effects of the substantial size of the databases and the use of multi loci typing systems.

Profiling Errors and Reporting Incorrect Profiles

As is the case with any system that requires human involvement and process, there is the potential for error. However, with appropriate laboratory protocols and procedures these can be eliminated. The most common sources of potential errors can be listed as follows:

- *Sampling errors*
  Sampling from the wrong place or inadequate sampling. Scientists make many decisions when sampling, based on police information and other possible explanations.
- *Contamination errors*
  This is always an issue. The use of aseptic techniques and protocols that demand that only one item/sample is examined at any one time contribute to the removal of this risk.
- *Analytical errors*
  Problems with the typing kit, the Genescan instrument, stochastic effects and scientific error.

- *Translation errors*

  The transcription of the DNA profiles from the spreadsheet, to the database and to the case files.

When considering errors it is of paramount importance to realise that alleles and profiles CAN NOT BE RANDOMLY PRODUCED during the profiling process. However, alleles can "drop out" or "disappear" resulting in a partial profile that will require a revised "degree of uniqueness".

Logic determines that, even if it was possible to randomly "manufacture" a profile, the odds of producing a profile that matches the defendant, complainant or any other individual is equal to the odds of randomly producing any other profile, that is ***billions to one***!

The DNA of any individual is only obtained if it was actually present or was due to contamination. If, for some obscure reason, an unidentified error occurred, then the logic of this discussion most certainly would ensure that an "unknown individual's" profile would appear in the results to the obvious advantage of the defendant whether he be guilty or not.

Defence Evidence

The presentation of scientific evidence and, in particular, DNA evidence has been an issue since its introduction. Most DNA evidence is presented by scientists employed in forensic science laboratories in Police Departments or other Government Departments. This evidence is generally provided through the Prosecution because it was originally produced when assisting police investigations. This is the case whether the laboratory is in a Police Department or not. Evidence for the Defence is provided by forensic science laboratories engaged to review the evidence and to advise on it. These laboratories can be other Government forensic science laboratories or laboratories in the private sector. Significantly, for defence experts employed in these latter laboratories, it is fair to say that most of them are not users of Genescan and/or Profiler Plus^{TM} technology.

To the chagrin of the courts or, perhaps, to their relief and delight, scientists will disagree from time to time. It is hoped however, that recent judgements in *R v Karger* (SA, Supreme

Court, 2000/01) and *R v Kami* (NSW, Supreme Court, 2001) will assist in providing some common sense to the system.

However, it must be said that the tolerance that the justice system shows to court appearances by unqualified "experts", and the demeaning treatment afforded to genuine experts is now a great source of frustration to forensic scientists. It would seem that the introduction of time consuming and expensive international accreditation procedures and standards has meant nothing and they have merely provided just another opportunity for "forensic mischief making" by scientific and legal opportunists.

The Latcha Judgement

The so-called "Latcha Rules" are a series of guidelines proposed by three Northern Territory Judges in the criminal appeal of Ricky Latcha against his conviction. These guidelines which, effectively, serve as rules for the presentation of DNA evidence, impact on laboratories and scientists.

Although these guidelines were written in response to biological evidence they are, in some instances, general enough to apply to any scientific evidence.

Furthermore, the Northern Territory forensic science laboratory has no issue with the majority of the guidelines as they are more focussed on the Prosecution obligations to the Defence and the courts.

However, several of the guidelines do refer specifically to DNA and they require some comment. These are as follows:

> (2) "Provided that the expert has the necessary data, it may then be appropriate for it to be indicated how many people with the matching characteristic are likely to be found in Australia, or in a more limited relevant sub-group, for instance, the sexually active males in the Darwin area, depending on the circumstances of the case."

The frequency of the amelogenin typing (a sex determinant) has never been used in the statistical calculation of the frequency of any DNA profile. The statistical database consists of individual DNA profiles and does not include any individual characteristics i.e. sexually

active males in the Darwin area because this information is not relevant for DNA match criteria.

(9) "Experts called to give statistical evidence should be led by the Crown as to any assumptions made in their calculations which, even though widely accepted, are not supported by empirical research. Including:

(a) Hardy-Weinberg equilibrium

(b) where the offender is of a racial group or sub-group for which there is no valid database and a general database has been used which does not take that fact into account, that fact."

The points made in (2) and (9) of the guidelines have been previously addressed in this paper, (refer to section 'Database Legitimacy'). The use of highly discriminating, multi loci systems, like Profiler Plus^{TM}, that produce odds in the order of 10^8 for "degree of uniqueness" and the fact that population studies have shown that there are very little differences between the sub-groups removes the requirement for the use of specific 'local' databases.

Summary

DNA has finally become established in the courts as an invaluable means of identification. The science is firmly established in many scientific disciplines including animal husbandry, medical research, anthropology, horticulture. It is disappointing although perhaps understandable, that, despite international quality assurance and quality control procedures, the basic science is still vigorously challenged in many court hearings.

One really must ponder whether science will ever meet the expectations of the law and whether the law would accommodate it anyway.

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