Forensic DNA Analysis in Criminal Investigations

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DNA analysis is a most powerful tool for human identification and has clear forensic applications in identity testing (crime scene and mass disaster investigations) and parentage determination. The development of forensic DNA technology in other countries and its potential to improve the Philippine criminal justice system are briefly discussed. The utility of forensic DNA testing in criminal investigations was highlighted using an actual criminal case wherein DNA evidence played a clear role in the resolution of the case.

Keywords: Criminal investigations, DNA profiles, forensic DNA analysis

Owing to the efficacy of forensic DNA analysis in human identification, the application of modern DNA technology has played a crucial role in identity testing (crime scene investigations, mass disaster and parentage testing). The method of fingerprinting using an individual’s DNA isolated from both living and deceased persons as long as biological samples are not exposed to adverse environmental conditions and/or microbial contaminants that can degrade them.

The method of fingerprinting using an individual’s DNA-based methods of identification

DNA, or deoxyribonucleic acid, is the fundamental building block of a person’s entire genetic makeup. DNA is present in all human cells and is the same in every cell (Figure 1). It is composed of sugar, phosphate and nitrogen bases namely Adenine (A), Guanine (G), Cytosine (C) and Thymine (T). The order of the nitrogen bases determines the so-called ‘DNA sequence’.

Several DNA molecules make up a gene. Humans have 22 pairs of body chromosomes (autosomes) and 1 pair of sex chromosomes per body cell.

The genetic make-up of each individual is unique (except for identical twins) and may be used to identify a person. DNA is also very stable and can be

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Figure 1. Diagram of DNA strand, gene and chromosome. Several DNA molecules make up a gene, and genes are located in chromosomes (nuclear DNA) or mitochondrion (mitochondrial DNA).
DNA (DNA fingerprinting) was invented by Alec Jeffreys in 1984 at the University of Leicester while studying the human myoglobin gene. The technique developed, called Restriction Fragment Length Polymorphism (RFLP), utilizes a special class of enzymes (restriction enzymes) to cut human DNA into smaller fragments that can be visualized as distinct banding patterns (Figure 2).

Since then, other techniques have been developed. These include the use of the reverse dot blot methods for the characterization of the human leukocyte antigen DQα (HLA-DQα) and other Polymarkers (Figure 3), sequencing of mitochondrial DNA (Figure 4), and the amplification of non-coding regions of the human chromosome with variable number of tandem repeats (VNTR) or short tandem repeats (STR) via the Polymerase Chain Reaction or PCR (Figure 5). STR markers are short DNA regions characterized by repeated sequences. Individuals may possess different numbers of repeats per copy of the STR marker (known as allele) without affecting their overall metabolism.

PCR-based methods that target highly polymorphic STR markers are currently the most prevalent procedures worldwide. This technology is preferred by many laboratories due to the extensive genetic variability of STR markers, the high success rate in generating DNA profiles with small quantities of DNA, the availability of standard kits and protocols, and the relative ease of DNA analysis. The UK, US, Australia and New Zealand use 6-20 STR markers for routine casework analysis. In the Philippines, the DNA laboratories of the National...
Figure 4. DNA sequencing results. Sequencing refers to the determination of the order of nitrogen bases A, C, G and T of DNA molecules in a prescribed region.

Bureau of Investigations (NBI), the Philippine National Police (PNP), St Luke’s Medical Center (SLMC) and the Natural Sciences Research Institute (NSRI), University of the Philippines routinely use 9-15 STR markers, albeit laboratories differ in the actual markers used. Some common STR markers used are listed in Table 1.

**DNA-based methods of identification**

The use of RFLP testing in human DNA identification was pioneered by Alec Jeffreys and was first used in the investigation of the rape/murder of two British schoolgirls in November 1986. In the initial investigations, semen samples isolated from the victims’ bodies did not match the suspect’s DNA. To find individuals with the same DNA pattern as that of the semen sample, police investigators requested 17 – 34 year old males living in the area to voluntarily submit blood samples. Over 4000 reference samples were processed and compared with the DNA pattern of the semen samples. Eventually, the DNA profile from the semen samples matched the DNA profile of a man named Collin Pitchfork. Pitchfork later confessed to the crime and was subsequently convicted for the rape and murder of the two schoolgirls. In this instance, DNA

<table>
<thead>
<tr>
<th>STR Marker</th>
<th>Definition of Marker</th>
<th>Chromosomal Location</th>
<th>Range of STR alleles in Philippine population</th>
</tr>
</thead>
<tbody>
<tr>
<td>HUMHILDA</td>
<td>Human urothelial 1</td>
<td>12</td>
<td>14 to 23</td>
</tr>
<tr>
<td>HUMFES1/FPS</td>
<td>Human 1q54.3</td>
<td>15</td>
<td>7 to 15</td>
</tr>
<tr>
<td>HUMF13401</td>
<td>Human 1q31.1</td>
<td>6</td>
<td>3.2 to 16</td>
</tr>
<tr>
<td>HUMTHD1</td>
<td>Human 9q33.1</td>
<td>11</td>
<td>6 to 10</td>
</tr>
<tr>
<td>HUMCSF1RD</td>
<td>Human myeloid 1q23</td>
<td>5</td>
<td>7 to 15</td>
</tr>
<tr>
<td>D8S1171</td>
<td>Not identified</td>
<td>8</td>
<td>2 to 13</td>
</tr>
<tr>
<td>HUMMDP23</td>
<td>Human MDR2 1q41</td>
<td>6</td>
<td>6 to 10</td>
</tr>
<tr>
<td>HUMHGA</td>
<td>Human alpha-fibrinogen 1</td>
<td>4</td>
<td>17 to 44.2</td>
</tr>
<tr>
<td>HUMNPOC</td>
<td>Human Thyroid pseudo-</td>
<td>2</td>
<td>6 to 12</td>
</tr>
</tbody>
</table>
evidence played an important role in excluding the first suspect and in identifying a second suspect who turned out to be the real perpetrator of the crime.

The initial success of DNA fingerprinting in resolving the rape/murder case of the two British schoolgirls in 1986 helped to promote the use of forensic DNA technology by law enforcement agencies. Since then, use of forensic DNA analysis to assist in criminal investigations has become routine in many countries such as the United Kingdom, United States, Canada, Germany, Japan, Australia, and New Zealand.

The use of forensic DNA analysis in criminal investigations depends on the availability and the proper processing of biological samples from crime scenes. Properly collected samples contain biological material that may lead to criminal identification. In the process of collecting and processing samples, great attention must be paid to issues of proper handling, contamination, and storage/preservation because of the minute amounts of DNA being handled. Reactions used in forensic DNA analysis are specific for humans, although in some instances, may also have positive reactions for DNA of non-human primates, e.g. chimpanzee, orangutan and gorilla. Due to the general specificity of the reactions used in forensic DNA analysis for human DNA, the common sources of false positive reactions are contaminations from human handlers, e.g. curious bystanders at the crime scene, investigators, medico-legal examiners, forensic analysts, evidence custodian and lawyers. Notably, false positive reactions do not occur when common non-human sources of contamination, e.g. dog, cat, bacteria, dirt, soil, grass or other plants, and water are mixed with human biological samples. For this reason, meticulous documentation of the chain of custody of samples listing the identities and extent of handling of all personnel, starting from the crime scene to the court, is required if physical evidence is to be made admissible. To date, DNA has been successfully isolated from diverse types of biological samples such as blood and bloodstains, saliva, semen and seminal stains, vaginal smears/swabs, tissues and organs, bones and hair with follicles in varying states of degradation.

Analysis of DNA evidence

Once samples are processed, possible sources of DNA profile/s are evaluated. Sources may be a) the victim; b) human handlers such as crime scene investigators, medico-legal officers, forensic analysts and lawyers; c) the perpetrator of the crime.

Suspect is excluded as source of evidence

The presence of two or more mismatches between the evidentiary stain and suspect’s reference sample necessarily excludes him as the source of the evidentiary sample. Notably, mismatches do not necessarily equate with innocence, but merely show that the suspect is not of the source of the evidentiary sample. Other evidence collected from the crime scene may still contain the suspect’s DNA or that the suspect did not leave sufficient DNA, if he is indeed the real perpetrator of the crime. Alternatively, the suspect may not have left sufficient DNA at the crime scene and other physical evidence (e.g. ballistics, shoeprint evidence) and information, (e.g. eyewitness testimony) must be used to further the case.

Nonetheless, the exclusion of a suspect as possible source of non-victim DNA that is not that of any known human handler is crucial in criminal investigations since this indicates the presence of another individual at the crime scene who remains unaccounted for.

Suspect as possible source of evidence

If a suspect’s reference sample is consistent with the DNA profile of the evidentiary sample, then the suspect remains as a candidate source of the sample. Since only a selected set of STR markers are analyzed, there remains a probability that another individual has the same DNA profile. If the alleles comprising the DNA profile are rare, then this profile may be attributed to only a few persons in a given population, and the likelihood of the suspect being the source of evidence is higher. Hence it is essential that the significance of matching profiles must be estimated using established statistical principles. In addition, match probability estimates and/or likelihood ratios must accompany all DNA reports submitted to
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Forensic DNA technology in the Philippines

Empirical examination of archived trial court records in the Supreme Court of affirmed Death Row inmates revealed the unavailability of physical evidence in many cases. For example, of the 58 Death Row cases (52 involving rape, two for robbery possession and one for multiple murder) reviewed by the UP DNA Analysis Laboratory in the first quarter of 2002, samples for only two cases are still available for DNA testing. Apparently many victims especially minors, do not immediately report the rape out of fear and shame. This was further compounded by the absence of guidelines for the proper collection and storage of collected evidence in local health and police units. Attempts to locate samples were unsuccessful because many samples were not stored or could not be located.

Theoretically, the chance of isolating biological material is much higher in sexual assault cases compared to other crimes due to the direct physical contact; hence transfer of biological material, between assailant and victim. In fact, the best biological samples that may be used as DNA evidence are those obtained from the victim and her clothing worn at the time of the assault. Sperm DNA is generally stable up to 72 hours in the female reproductive organs, provided that the victim does not bathe or wash during this time period. In the US, DNA testing is mostly used in rape and homicide prosecutions.

The unrealized potential of using forensic DNA technology in accelerating criminal investigations in the Philippines is great. Medical records at the National Bureau of Investigations (NBI) and the Philippine National Police (PNP) from August to November 2000 contained 780 sexual assault cases (366 cases in NBI and 414 cases in PNP). Within this period, the PNP Crime Laboratory examined 174 victims (42%) <72 hours after the commission of the crime compared to 106 NBI cases (29%). In theory, the proper collection and subsequent laboratory analysis of samples, in particular vaginal swabs, should facilitate the identification of the real offenders in these cases. Clearly, if DNA analysis was conducted on all these samples, then the identification of real suspects or exclusion of individuals would have undoubtedly accelerated the criminal investigations of these cases.

Figure 6. Microscopic preparation of sperm cells (Eosin stain). Sperm cells have a characteristic cellular morphology: head with a long tail. This morphology is easily distinguished by visual inspection under a microscope, but is vulnerable to cellular disruption. The tail region is mainly used for motility whereas sperm DNA is located at the head region. Sperm cells are may be disrupted thus characteristic cell morphology is destroyed, releasing sperm DNA. In this instance, sperm cells are not detected but sperm DNA may have been transferred to the victim, which may be amplified at prescribed STR markers to generate a DNA profile of the perpetrator.

DNA evidence in Philippine courts: A case report

To demonstrate the role of DNA evidence in criminal courts and in resolving a disputed paternity case, the case of People of the Philippines vs. Victoriano Paras (Criminal case nos. 85974-85978 Regional Trial Court, Branch 163 Pasig City) is discussed. This case was selected because it involved a man charged with sexual assault resulting in the victim’s pregnancy and birth of a child. DNA evidence was obtained by conducting a simple paternity test on the DNA of the suspect, the victim and the child.

The accused was charged with five counts of rape committed on various dates, namely: December 31, 1989, the first, second and fourth weeks of January 1990, and first week of February 1990 leading to the birth of a child on November 8, 1990. The case was filed on March 31, 1991. However inconsistencies were detected in the testimony and subsequent cross examination of the victim. In addition, the defense presented evidence that showed the accused was not in Pasig during the period covered by the charges. The defense also argued that the child was born 10 months after the last incident of the supposed rapes.

Hence, to determine whether the accused was indeed the father of Joanna Ocra, the Court ordered the UP-NSRI DNA Analysis Laboratory to conduct DNA tests on the child, the victim and the accused. Two weeks later, on the basis of mismatching DNA profiles at four out of five STR markers tested, the
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accused was excluded from being the father of the child.

The results of the laboratory examination, the inconsistencies of the victim’s testimony as well as other evidences presented by the defense in Court on the whereabouts of the accused during the stated time and dates of the incidences of rape cast a very serious doubt in the mind of the court as to the guilt of the accused on the five incidences of rape filed against him (Judge Aurelio C. Trampe, 5 May 1999). The accused was subsequently acquitted and released.

Nonetheless, the accused was imprisoned and his petition for bail was denied whilst the case was being tried in court. Thus, prior to acquittal, the accused had already been incarcerated for six years. In contrast, DNA analysis that provided key evidence in this case was conducted within two weeks. Consideration of these facts highlights the need to incorporate forensic DNA testing in routine criminal investigations to decrease the possibility of erroneous convictions as well as to accelerate the progress of pending cases that clog Philippine courts.

Conclusion

The strength of DNA evidence to resolve crimes is well established in the US, Germany, Australia, New Zealand, Japan, the UK and other countries. However, the routine use of DNA evidence in criminal cases has yet to be adopted in the Philippines. The development of DNA testing and forensic science overseas has provided us a remarkable opportunity to improve our criminal justice system. Further, the availability of forensic DNA technology in the Philippines necessitates the amendment of current rules of evidence to incorporate scientific advances which enables the judiciary to better appreciate the value of physical evidence in criminal courts.

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References

Asplen CH. From crime scene to courtroom: integrating DNA technology into the criminal justice system. Judicature 1999;83(3):144-149.


De Ungria MCA, Frani AM, Magno MMF, Tabbada KA, Calacal GC, Delfin FC, Halos SC. Evaluating DNA tests of motherless cases using a Philippine genetic database. Transfusion 2002;42:954-957.


