Forensic applications of molecular genetics: ethics and law to inform public policy issues

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Abstract

ABSTRACT

Molecular analysis of DNA variation has usurped the place of all earlier technologies in forensic identification of victims and suspects alike. Although the field of ethics has made attempts to cope with the plethora of available genetic information, especially in clinical application, there has been little scrutiny of emerging ethical issues in the forensic domain. Legal scholarship highlights some aspects of the emerging issues, with particular relevance to the challenges faced in court and those regarding individual liberties.

The overall objective of this thesis was to evaluate the scientific validity, ethical acceptability and legal accountability of the forensic applications of molecular genetics. In particular, contemporary science has allowed us to access information far beyond what was originally anticipated, such that trace DNA can be obtained trivially from any individual. As a consequence, the scope and composition of existing DNA banks far exceeds the legislative mandate. Chapter 1 reviews the current legal standards for evidence and assesses the level of exactitude necessary for forensic DNA testing to meet evidentiary standards. An evaluation of current practices in DNA banking revealed adequate informed consent practices; the need for a re-examination of access to public health samples with attention to local population interests and the necessity for developing standardized guidelines for banking practices and uniform quality assessment measures (Chapter 2). Comparing current forensic and genomic markers revealed similar concordance and discordance rates with a slight performance advantage towards the forensic markers. The results indicate that multiple runs are necessary to ensure reliability (Chapter 3). A significant ethical issue arises from the forensic practice of surreptitious DNA sampling. This lack of transparency violates autonomy, threatens the legitimacy of the State's interest and may exacerbate existing issues of discrimination, universality, privacy,

Abstract

confidentiality, consent and coercion (Chapter 4). Public consultation is required to develop guidelines for socially agreed upon methods of DNA sampling, the utilization of forensic samples in research, resource allocation and the extent to which a universal DNA bank is appropriate. Canada should take the lead in developing protocols for ethical use of DNA in forensic testing (Chapter 5).

RESUMÉE

L'analyse moléculaire des variations de l'ADN a supplanté toutes les technologies médicolégales antérieures d'identification des victimes et des suspects. Bien que le champ de l'éthique ait tenté de gérer la pléthore d'information génétique disponible, particulièrement dans les applications cliniques, il y a eu peu d'examen des enjeux éthiques émergeants dans le domaine médicolégal. La recherche juridique met en évidence certains aspects des enjeux émergeant avec une pertinence particulière pour les défis auxquels les tribunaux sont confrontés ainsi que les défis à l'égard des libertés individuelles.

L'objectif général de cette thèse était d'évaluer la validité scientifique, l'acceptabilité éthique et la responsabilité légale dans les applications médicolégales de la génétique moléculaire. En particulier, la science contemporaine nous a permis d'accéder à des informations qui vont au-delà de ce qui était anticipé à l'origine si bien que des traces d'ADN peuvent être obtenues trivialement de tout individu. En conséquence, l'étendue et la composition des banques existantes d'ADN excèdent de loin le mandat législatif. Le premier chapitre revoit les standards légaux d'évidence et évalue le niveau d'exactitude nécessaire afin que les tests d'ADN médicolégaux rencontrent les standards d'évidence. Une évaluation des pratiques actuelles dans la mise en banque d'ADN a révélé des pratiques de consentement éclairé adéquate, le besoin de réexaminer l'accès aux échantillons de santé publique en portant l'attention aux intérêts des populations locales et la nécessité de développer des lignes directrices standardisées pour les pratiques de mise en banque et de mesures uniformes de l'évaluation de la qualité (chapitre 2). La comparaison des marqueurs médicolégaux actuels aux marqueurs génomiques a révélé des taux de concordance et de discordance similaires avec un léger avantage en faveur des performances des marqueurs médicolégaux. Les résultats indiquent que de multiples essais sont requis pour assurer la fiabilité (Chapitre 3). Un enjeu éthique substantiel émerge de l'échantillonnage subreptice de l'ADN. Ce manque de transparence viole l'autonomie et menace la légitimité de l'intérêt des États et pourrait exacerber les enjeux existants de discrimination, d'universalité, de vie privée, de confidentialité, de consentement et de coercition (Chapitre 4). La consultation publique est requise afin de développer des lignes directrices pour des méthodes socialement acceptées pour l'échantillonnage d'ADN, l'utilisation d'échantillons médicolégaux pour fins de recherche, l'allocation des ressources et le caractère approprié d'une banque universelle d'ADN. Le Canada devrait prendre les devants dans le développement de protocoles pour l'usage éthique de l'ADN dans les tests médicolégaux (Chapitre 5).

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CONTRIBUTIONS TO ORIGINAL KNOWLEDGE

This thesis draws from the disciplines of genetics, ethics, and law to inform public debate and policy development in the application of DNA in forensic identification. The multidisciplinary and translational nature of the approach taken in this thesis is both novel and original.

Specific contributions to knowledge include:

Palmour N (2003) A survey of DNA banking world-wide, In: Populations and Genetics: Legal and Socio-Ethical Perspectives. Linden, NL: Martinus Nijhoff Publishers.

This evaluation of banking practices in DNA showed that practices regarding informed consent were adequate, but that accesss to public health samples required reexamination, with particular attention to local population interests. This analysis also revealed a necessity for developing standardized guidelines for banking practices and uniform quality assessment measures.

Palmour N, Hudson TJ (unpublished) SNP'ing away at STR dominance in forensic profiling: A time for change.

Insofar as we know, this was the first pairwise comparison of the accuracy and performance of genomic vs forensic markers, using contemporary automated methods of genotyping, and is still the only study evaluating the ability of these markers to discriminate related individuals one from another. Comparing current forensic and genomic markers revealed similar concordance and discordance rates with a slight performance advantage towards the forensic markers. The results indicate that multiple runs are necessary to ensure reliability.

Palmour N (2004) Suspect and subject: Does committing a crime constitute consent for research? Amer Soc Bioethics Research-GE3LS, Winter Symposium, Vancouver BC (poster presentation) and Palmour N (in preparation).

Contributions to knowledge

There has been minimal prior attention to the derivation of the prerogative for DNA sampling from the standpoint of a liberal political framework or a social contract perspective. Chapter 4 of this thesis (to be submitted for publication in a modified form) uses these principles to conduct an extensive analysis of the "function creep" of current DNA banking practices, and calls particular attention to troubling infringements of autonomy and the social contract. In Chapter 5, we identify surreptitious DNA sampling as the greatest threat to the social contract and call for public consultation to address its expanding use and the contraction of personal liberties.

GLOSSARY OF TERMS

1. Adenine: One of the four bases in DNA; abbreviated A.

2. Allele: One of two or more alternative forms of a gene. In DNA identification, the definition is extended to any DNA region used for analysis.

3. Amelogenin: A system that generates different sized fragments for the X and Y chromosomes, and hence is used for sex identification.

4. Amplification: Producing multiple copies of a chosen DNA region, usually by PCR.

5. Autosome: Any chromosome other than the X and Y.

6. Chromosome: A physical structure in the cell nucleus consisting of a tightly coiled thread of DNA with associated proteins and RNA. The genes are arranged in linear order along the DNA.

7. Cytosine: One of the four bases in DNA; abbreviated C.

8. Denaturation: Separation of double stranded DNA into single strands.

9. Deoxyribonucleic acid (DNA): The genetic material; a double helix composed of two complementary chains of paired bases (nucleotides).

10. Deletion Loss of a segment of DNA, which may be as small as a single base, or large enough to encompass one or more genes.

11. Electrophoresis: A technique in which different molecules are separated by their rate of movement, usually through a gel, in an electric field.

12. Gene: The basic unit of heredity; a functional sequence of DNA in a chromosome.

13. Genome: The total (haploid) genetic makeup of an organism. In the human this comprises about 3 billion base pairs.

14. Genotype: The genetic makeup of an individual, as distinguished from its manifestation (phenotype); usually designated by allele symbols, e.g., A 1A2 designates the genotype of an individual with alleles A land A2. The word is used to designate any number of loci.

15. Guanine: One of the four bases in DNA; abbreviated G.

16. Hardy- Weinberg proportions: The state, for a genetic locus in a population, in which the alleles making up the genotype are in random proportions. Glossary of Terms

17. Heterozygosity: The proportion of a population that is heterozygous for a particular locus.

18. Heterozygote: A fertilized egg (zygote) with two different alleles at a designated locus; by extension, the individual that develops from such a zygote (cf. homozygote).

19. Homologous: Corresponding; used to describe the relationship between two members of a chromosome or gene pair.

20. Homozygote: A fertilized egg (zygote) with two identical alleles at a designated locus; by extension, the individual that develops from such a zygote (cf. heterozygote).

21. Linkage: Inheritance together of two or more genes on the same chromosome.

22. Locus (pi. loci): The physical location of a gene (or DNA region of interest) on a chromosome.

23. Marker: An easily detected gene or chromosome region used for identification.

24. Mitochondria1 DNA: DNA in the mitochondria. Mitochondrion (pl. mitochondria): A particle present in multiple copies per cell and transmitted from the mother to all her children.

25. Mitosis: The process of cell division in which the chromosomes are precisely distributed so that the parent and each daughter cell have the same chromosome content.

26. Multiplex: A system for analyzing several loci at once.

27. Nucleotide: A unit of DNA composed of phosphate, a sugar and a purine or pyrimidine base.

28. Phenotype: The recognizable manifestation of the genotype; it may be externally visible, as hair color, or observed by a special technique, as blood groups or enzymes.

29. Polymerase chain reaction: An in vitro process for making many copies of a chosen fragment of DNA; abbreviated PCR.

30. Polymorphism: The presence of more than one allele at a locus in a population; usually the word is used only when at least two alleles are fairly common.

31. Random match: A match in the DNA profiles of two DNA samples, where one is drawn at random from the population.

32. Random-match probability: The probability that the DNA in a random sample from the population has the same profile as the DNA in the evidence sample.

33. Restriction enzyme, restriction endonuclease: An enzyme that cuts a DNA molecule in a specified short base sequence.

34. RFLP: Restriction fragment length polymorphism: Variation in the length of a stretch of DNA.

35. Sex chromosomes: The X and Y chromosomes.

36. Short tandem repeat: A tandem repeats in which the repeat units are two, three, four, or five base pairs; abbreviated STR.

37. SNP: Single nucleotide polymorphism. A mutation in a single base at a specific location.

38. Somatic cells: Body cells; cells other than those in the cellular ancestry of egg and sperm.

39. Thymine: One of the four bases in DNA; abbreviated T.

40. Uracil: A base in RNA, corresponding to T in DNA; abbreviated U.

41. VNTR: Variable number of tandem repeats: A polymorphism based on repeating units of a DNA sequence, in which different alleles have differing numbers of the repeating unit.

42. X chromosome: A sex chromosome, present twice in female cells and once in male.

43. Y chromosome: A sex chromosome present once in males, and transmitted directly from a father to all his sons.

44. Zygote: A diploid cell produced by fusion of an egg and sperm.

Preface

PREFACE

The use of molecular genetics in the criminal justice system, specifically the identification of heritable variants in deoxyribonucleic acid (DNA) has increased exponentially, both in frequency of use and in sophistication, over the last two decades. Though the field of ethics has made attempts to cope with the plethora of available genetic information, especially in the field of clinical applications, there has been little scrutiny of emerging ethical issues in the forensic domain. Legal scholarship highlights some aspects of the emerging issues, with particular relevance to the challenges faced in court and those regarding individual liberties.

The overall objective of this thesis is to evaluate the scientific validity, ethical acceptability and legal accountability of the forensic applications of molecular genetics. In particular, contemporary science has allowed us to access information far beyond what was originally anticipated, such that trace DNA can be obtained trivially from any individual. As a consequence, the scope and composition of existing DNA banks far exceeds the legislative mandate.

Specifc Objectives

The specific objectives of this project are to:

Review current legal standards for evidence and evaluate whether present forensic DNA tests meet these standards (Chapter 1);

Evaluate the current practices in DNA banking (Chapter 2);

Compare the performance of standard forensic tools with tools standardized for genomic biology (Chapter 3);

On the basis of these data, to analyze issues of validity and reliability in the application of molecular genetics to forensic analysis of DNA evidence (Chapter 3);

Preface

Conduct an ethical analysis of issues which arise from the application of molecular genetics to forensics (Chapter 4);

Identify areas which require the elaboration of policy and legislation, and to call for public consultation regarding these topics (Chapter 5).

CHAPTER 1: BACKGROUND AND INTRODUCTION

1.1 Scientific History

Identification is the foundation of forensic science. As with any science the object's characteristics are described, it is identified, classified and then compared with similar items to help determine what makes it unique or individual among a similar class of items.



One of the oldest and most enduring forms of art and identity is the

Mesopotamian cylinder seal, originating in 3500-3000 BCE (Collon 1988). The cylinder seals, like the one pictured here¹, are small (2-6 cm) cylinder-shaped stones carved with an engraved decorative design. The seal when rolled over a wet clay tablet would leave a continuous impression of the design, reversed and in relief. They served to identify their owners and to seal a variety of business transactions just as a signature would today. The cylinder imprint, which covered an area as large as desired, was used to mark or identify clay tablets, envelopes, ceramics and bricks. The seals, intimately associated with writing, were usually small and were often worn as jewelry or as magical amulets. The seal impressions, even before the owner's name was incised on the cylinder, stood for a particular individual and met the needs required by the development of city states.

Cylinder seals provided an ingenious way to bind individuals to contracts and minimize forgeries. Similarly the Chinese in the 700's CE were the first to use fingerprints as an

¹ Cylinder seal of Ibni-sharrum, a scribe of Shar-kali-sharri (left) and impression (right), ca. 2183–2159 B.C.; Akkadian, reign of Shar-kali-sharri. Mesopotamia. Cuneiform inscription in Old Akkadian. Serpentine; H. 3.9 cm (1 1/2 in.); Diam. 2.6 cm (1 in.). Musée du Louvre, Département des Antiquités Orientales, Paris AO 22303. http://www.metmuseum.org/explore/FIRST_Cities/seals_meso.htm

identification mark on documents and pottery (New York Times 1919; Beavan 2001). Those fingerprints are still used to identify works today (Chinese Fingerprint Museum 2007).

Identification is a necessary prelude to individualization in forensic science (Kirk, 1963). As such the first recorded beginnings of what would later become forensic science or Criminalistics focused on identification. The text *Hsi Duan Yu* (the Washing Away of Wrongs) was written in China in 1248 and was the first description of the application of medical knowledge to the solution of crime and its relevance under the law (Song 1249). The principle focus of the author in one section was the determination of the cause of death (Song 1249). The text outlined methods for distinguishing natural deaths from those caused by drowning (indicated by the presence of water in the lungs) or strangulation (damage of the neck cartilage and pressure marks on the throat).

The timeline of forensic sciences is replete with contributions from individuals in a variety of disciplines mostly focusing on identification, from documents to fingerprints and even the presence of blood. A more comprehensive forensic science timeline is available at http://forensicdna.com by Norah Rudin and Keith Inman. One contribution to forensic science, that forms the basis of most of the work done in the area, was proposed by Adolph Quetelet in the 1830s.

Adolph Quetelet, the father of descriptive social statistics was a 19th Century Belgian statistician and sociologist (Britannica 1995). He pioneered the notion that no-two-are-alike, or more precisely he hypothesized that nature never creates biological duplicates (Kirk 1963). This theory, which cannot be proven mathematically (Saks 1998), was adopted by Alphonse Bertillon in

"I saw him not, I heard no sound; but traces everywhere I found" (Brotherton 1900).

This line of poetry is just one description of the many thousands of exchanges of material that occur between ourselves and our environment on a daily basis.

the late 1800s and used to develop the first system of forensic identification, known as anthropometry, or bertillonage (Kirk 1963). Eleven different physical features of each prisoner were measured and the data assembled into special files. The goal was to thwart prisoners' use of aliases on re-arrest through identification by their measurements. The system gave credence to Quetelet's hypothesis and Bertillon established the concept that people varied to such a degree on some attributes that certain measures could be useful in identifying them.

As a student of Bertillon, Dr. Edmond Locard advocated the application of scientific methods to identification and criminal investigation. His work in the 1930's formed the basis for what is widely regarded as a cornerstone of the forensic sciences (Chisum & Turvey 2000). His three paper treatise on dust laid the foundation, as it were, for his Theory of Transfer also known as The Exchange Principle (Locard, 1930a,b,c). "When two objects meet, some evidence of that meeting can later be found and verified" (Locard 1920; Horswell 2004). We constantly shed organic material such as skin cells, hair, fibers from our clothes, and saliva on drinking glasses and at the same time we pick up material from our surroundings such as fibers from carpeting and furniture, hair left from pets and others, even viruses. The Locard principle of exchange is the bedrock of a subset of forensic sciences, criminalistics, which helps to establish circumstantial evidence in crime scene detection (Rudin and Inman 2002). The term "criminalistics" was coined by Dr. Hans Gross and evolved into a distinct discipline between the early to mid-1900's.

Criminalistics or forensic science has been defined as the science of individualization (Kirk 1963) while others claim it "is that profession and scientific discipline directed to the recognition, identification, individualization, and evaluation of physical evidence by the application of the natural sciences in law-science matters" (Osterberg 1968). Regardless of the

designation identification and individualization or individuation is the goal. Identifying the item involves grouping that item into a class of similar items. Individuation comes about by looking for those traits that make this item unique from those within its class. Once the distinctiveness of the item is established it can then be matched with its source. "While normal science is concerned with establishing regularities, forensic science is concerned with exploiting irregularities among objects within classes" (Saks 1998).

The discovery of the human ABO blood types by Karl Landsteiner in 1900, for which he was awarded the 1930 Nobel prize, was one of the first ways in which differences among the blood of individuals could be exploited (Rudin and Inman 2002). The technique was quickly adapted by Max Richter to type blood stains. This allowed the exclusion of a large percentage of the population as potential donors at a crime scene. Genetic typing of biological fluids became a potent technique in forensic science. There are four common blood types: type A, B, AB, and O each with known frequency within populations (Sudbery 2002). The goal of typing biological fluids is to reduce the frequency to the smallest possible percentage of the population. As a tool, combined with other blood typing systems, it can be used to both include and exclude individuals.

Another early contribution to the use of genetic polymorphisms to identify individual variability was the work done by Harris and Hopkinson. In 1976 they typed common blood protein polymorphisms (14 enzymes) and performed population analyses to determine the prevalence of these variants in the UK population. The probability of expressing the single most common phenotype at all 14 loci was 6/1000 and the probability that two individuals would have exactly the same genotypes was 3/1000 (Harris and Hopkinson 1976).

The fundamental approach to conventional blood grouping is the same as that used in DNA typing in crime scene reconstruction. The object is distinguishing between individuals and identifying those individuals who are unknown but were present at the commission of a crime. In forensic science this is accomplished in a variety of ways all employing Locard's principle of transfer to examine fibers, hairs, fingerprints, discarded items, skin cells, blood and other body fluids. More specifically the focus is on those minute bits of biological matter that will allow us to identify an individual through DNA. This is the arena of forensic DNA profiling.

1.1.1 Forensic DNA Profiling

Deoxyribonucleic acid, or DNA, is contained in all cells of the body except the red blood cell and, within a given individual, has the same base pair sequence regardless of whether it is extracted from semen, hair bulbs, tissue or white blood cells. This fact coupled with the uniqueness of each person's DNA is the basis for DNA profiling. The concept of hereditary substances being passed down from parent to offspring was advanced by the Moravian Monk Gregor Mendel in 1865. His experiments and observations with garden peas are considered to be the beginnings of genetics. The term gene, to encompass this hereditary material was coined by W. Johannsen a Danish plant scientist (Kirby 7). Genes are contained within DNA and it is this DNA which acts as a messenger carrying all of the necessary information for offspring. The elucidation of the structure of DNA and the ability of nucleotides to pair with each other by Watson and Crick in 1953 is the basis for all genetic manipulations of recombinant DNA and DNA typing (Witkowski 1990).

The foundation for the concept of identity profiling using recombinant DNA was established by Wyman and White in 1980. They observed "a polymorphic DNA locus characterized by a number of variable length restriction fragments called restriction fragment

length polymorphism's (RFLP'S)" (Kirby 1992) essentially using "several regions of DNA that vary from person to person to identify the source of a tissue sample" (Drlica 1997).

"Fingerprinting" using DNA is contemporary, dating from 1985, and started with the publication of a paper by Alex Jefferys et al. The paper entitled "Hypervariable Minisatellite Regions in Human DNA" outlined the discovery of a region, in the human myoglobin gene, consisting of a 33-base pair (bp) sequence repeated four times (a tandem repeat or "minisatellite") within an intervening sequence. Such regions are hypervariable because the number of tandem repeats is variable both within a locus and between loci. They also discovered that each repeat unit contains a smaller 16-bp core in common with other minisatellites (Jeffreys et al. 1992). "When DNA is isolated, cleaved with a specific enzyme, and hybridized under low stringency conditions with a probe consisting of the core repeat, a complex ladder of DNA fragments is detected. The profile generated from the variable number tandem repeat method (VNTR) appears to be unique to each individual. Different core repeats were later isolated and used to produce a number of different probes useful for fingerprinting" (Kirby 1992). This technique was quickly embraced in the United Kingdom and was later adopted in the US for general use by crime laboratories (Fridell 2001). The terminology was later changed, opting for the more descriptive term of DNA profiling or DNA typing. DNA typing would have remained a small-scale analytical method if it weren't for the work of Kary Mullis and the development of polymerase chain reaction (Mullins 1990; Sambrook and Russell 2001).

1.1.1.1 Polymerase Chain Reaction

The use of DNA evidence in criminal cases was limited for years because of the need for large samples in order to garner enough information about the perpetrator of a crime. Even with the technology available, all of the bodily evidence collected at crime scenes was limited to the

circumstantial range, because there was often not enough sample present to be analyzed. Hair was matched for color or texture and blood samples were matched for type. The probability of someone other than the accused having these traits was easily arguable. The problems of limited sample size were substantially reduced with the discovery of thermostable DNA polymerase, specifically *Thermus aquaticus*, and development of polymerase chain reaction (Saiki et al. 1985; Mullis et al. 1986; Mullis and Falonna 1987; Lieu et al. 1976; Kaledin et al. 1980). This breakthrough allowed the use of molecular genetics to be expanded to a routine forensic tool and applied to crime scene DNA (Rudin and Inman 2002).

Polymerase chain reaction, or PCR, is "a test tube reaction in which a specific region of DNA is amplified many times by repeated synthesis" using DNA polymerase and specific primers to define the ends of the amplified regions" (Drlica 1997). PCR mimics the normal cellular replication of DNA. What makes PCR possible is *Thermus aquaticus* or *Taq* polymerase which maintains its activity after exposure to heat. With current methodology, a given segment of DNA can be magnified thousands or even millions of times if necessary to permit identification of existing alleles. However, the quality of the starting sample remains important; if the DNA which has been collected is either contaminated (with other DNA samples) or degraded, the value of the end product will be significantly reduced. The primers selected for forensic applications are all in non-coding regions of DNA (Butler 2001; Kaye 2001). Primers are synthetic polynucleotide "tags" that facilitate the synthesis of specific sections of DNA, and essentially provide bookends which tell the Taq polymerase where to start reading and where to stop (Strachan and Read 1999). Chemically, the primers are the sequence complements to the initial (forward primer) and terminal (reverse primer) ends of the DNA which is to be amplified. While multiple primers can be amplified at the same time, many labs have found that the best

fidelity is obtained if each region is amplified separately, and then multiplexed for gel analysis (Butler 2001).

PCR basically consists of three phases: denaturation, annealing and primer extension, repeated over and over. The principle through which PCR operates is simple: when a sample of DNA is heated the hydrogen bonds holding the two strands together are broken (denatured) and the two strands uncouple. The synthetic primers, which are present in excess, move in to bind to their complementary regions (annealing). From the primers a faithful copy (primer extension) of the targeted region of DNA is created with the aid of thermostable (Taq) DNA polymerase and DNA precursors; the four deoxynucleoside triphosphates (Strachan and Read 1999). The first step of replication lays down a small fragment on each parent strand, while during the second round, new complementary DNA is produced to both the parent strand and the fragment. After multiple rounds of DNA synthesis, there remain only a few parent strands of DNA and many hundreds of copies of the fragment that is under investigation. The small fragments of DNA are then separated either by gel or capillary electrophoresis. For gel electrophoresis the final mixture is then applied, with controls, to a polyacrylamide gel matrix, which may be either in the form of a sheet or a column. The gel then has an electric charge passed through it and the fragments of DNA separate based on their molecular weight. "Because the PCR products are produced in such large quantity and most is loaded onto the gel, the target fragments can be readily detected simply by staining the gel with silver or with ethidium bromide" (Easteal, McLeod and Reed 1991). The end result is a picture of several lanes with bands in each lane except for the negative control, which should be empty (see figure 1). The bands of several samples can then be compared. The fragments may be segregated by gel electrophoresis as described above or by capillary electrophoresis.

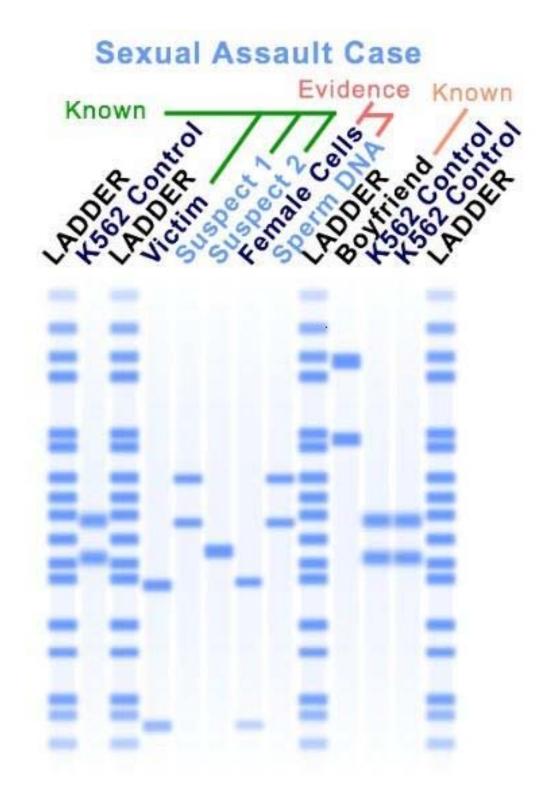


Figure 1: Gel electrophoresis results for a single marker. Jane Wang. The Science Creative Quarterly, <u>www.scq.ubc.ca</u>.

The capillary electrophoresis method requires the primers to be fluorescently tagged before PCR so that as they become incorporated into the amplified DNA fragments, the fragments will be fluorescent (Spencer 2004). The capillary electrophoresis method uses fine-bore glass capillaries filled with a gel material. The DNA sample is placed at the top of the tube, an electric current passes through the gel and the DNA fragments separate by size as they migrate down the capillary toward the positive electrode. A laser detector at the bottom of the tube detects florescent DNA as it passes the end of the capillary. The data are then collected and analyzed by software that calculates the DNA fragment sizes as well as the relative amount of DNA in each band. Data are visualized as graphs showing the position and size of the DNA fragments, which can be used as forensic evidence.

1.1.1.2 Genetic Markers

Tandemly repeated sequences are present in satellite DNA, but shorter repetitive sequences, such as, minisatellites and microsatellites are ideal for forensic purposes. VNTR or minisatellite loci are composed of sequence motifs ranging from around 15-50 bp in length, reiterated tandemly for a total length of 500 bp to 20 kilo-bases (kb) (Carracedo and Sanchez-Diz 2005). Microsatellites or short tandem repeats (STR's) are much shorter ranging from 2 to 6 bp for a total length between 50 and 500 bp. VNTRs and STRs differ in their distribution in the human genome and probably in their biological function (Carracedo and Sanchez-Diz 2005). Minisatellites are common in the subtelomeric regions, while STRs are widely distributed throughout the human genome and occur with a frequency of one locus every 6-10 kb (Nakamura et al., 1987). Initially the VNTR method was used for forensic identification purposes but the introduction of PCR, well-characterized and validated STR markers (requiring less initial sample), and the automation and miniaturization of equipment rendered it obsolete.

The short tandem repeat technique has essentially replaced the VNTR method and is used by researchers in a number of fields for a variety of reasons, including disease diagnostics, gene mapping, evolutionary biology and human identification (Gusamo and Alves 2005). The short segments of the STR markers are composed of repeated motifs such as CA and GATA that vary in both the number of motifs and length, and usually have many alleles (Butler 2001; Rudin and Inman 2002). This short length allows the STR's to be easily amplified using PCR. The STR loci chosen for forensic use are fairly well distributed in any given population. Currently there are 16 STR loci that are routinely used in forensic DNA profiling, 13 of which are part of the CODIS DNA profile database and used by the Royal Canadian Mounted Police (Spencer 2004). The Combined DNA Index System or CODIS is a national DNA database established by the Federal Bureau of Investigation in 1998 (Rodriguez 1998).

There are a few gender variations in STR markers that have been exploited by the forensic community, such as the amelogenin locus (Thangaraj, Reddy, Singh 2002). This gene for tooth pulp displays a length variation between genders. The female form of the gene contains a small deletion (6bp) in nonessential DNA and thus yields a shorter PCR product. Therefore, upon analysis the feminine form, with two X chromosomes, will present as a single band while the masculine form, with both an X and a Y chromosome, will present as two bands, one the same size as the female and one slightly larger. The gender length variation, however, is not uniform across all populations (Kashyap et al. 2006; Michael and Brauner 2004; Vauhkonen et al. 2004).

The sex chromosomes can also provide useful gender-specific information about the composition of forensic samples. Both X and Y-STR markers are just as amenable to typing in small or degraded samples of DNA as autosomal STR markers. Y-STRs are male specific

markers, located on the nonrecombining portion of the Y chromosome. The Y chromosome is transmitted faithfully from father to son (unless a mutation occurs) and can therefore be used for a variety of forensic purposes (Gusamo and Alves 2005).

Most of the genetic typing systems make use of loci found in nuclear DNA, as the majority of genetic material in the human genome resides in the nucleus of each cell. There is however additional genetic material contained in other small subcellular compartments called organelles that can benefit forensic identification. The mitochondrion in human cells contains an autonomous circle of DNA and is where some of the processes of cellular respiration take place. The mitochondrial DNA (mtDNA) codes for some proteins that control respiration function (Rudin and Inman 2002). The D-loop, a subsection of the mtDNA, has an extremely high mutation frequency of about 10 times that of nuclear DNA. This high mutation rate makes the region attractive for use in individual identification because mtDNA sequences are highly variable between unrelated individuals. The genetic material from mitochondria is inherited only from the egg cell of the mother, exhibits maternal inheritance and thus mothers pass on their mtDNA to all of their progeny regardless of gender (Rudin and Inman 2002). Mitochondrial DNA is often used in forensic casework analysis when the evidence material contains scarce amounts of DNA. The high copy number of the mitochondrial genomes per cell allows mtDNA analysis to be employed for human identification, overcoming the limitations of minimal or degraded DNA and providing, often crucial, forensic evidence (Allen and Andreasson 2005).

Single Nucleotide Polymorphisms (SNP) are single base differences in the DNA sequence. They are usually diallelic, highly abundant and are estimated to occur at 1 in every 1000 bases in the human genome (Sorbino and Carracedo 2005). They are considered to be the most common class of human polymorphism. The majority of SNPs are located in noncoding

regions of the genome and appear to have no dectable impact on the phenotype of individuals. It has been suggested (proven) that SNPs can be used as markers to identify genes that predispose individuals to complex disorders by using linkage disequilibrium and are currently of keen interest to researchers (International HapMap Consortium et al. 2007; McCarroll and Altshuler 2007; Sorbino and Carracedo 2005; Altshuler 2000). SNPs are also receiving attention from the forensic community, as they may be the smallest genetic marker available for identity testing as STRs require a minimum fragment length range of 100-360 base pairs of template DNA versus 57 to 146 base pairs for SNPs (Dixon et al. 2005). The single base change detection, even for multiple loci necessitates minimal sample size and makes SNPs more amenable to typing under degraded conditions or poor sample quality. Further, the diallelic nature of SNPs and current technological innovations makes them amenable to automated, high-throughput genotyping with no electrophoresis required. The development of SNP marker sets for forensic applications are underway (Budowle and van Daal 2008; Pakstis et al. 2007; Onofri et al. 2006).

However, all of the work and innovations in DNA profiling analyses would be moot if this, often crucial, evidence was not recognized in a court of law. The admissibility of scientific evidence in the courtroom has been an arduous and surprising journey in law.

1.2 Legal Background on Scientific Evidence

In this section a historical perspective will be employed for a legal analysis of the acceptability of scientific evidence followed by the use of DNA evidence within the courtroom. As such legal literature from both Canada and United States shall be selectively drawn from to identify, contrast and discuss the salient legal issues raised by the use of DNA in a forensic context. No attempt will be made to be comprehensive and canvass all of the 50 US State or 12 Canadian jurisdictions; the cases cited are illustrative of the issues and selectively referenced.

This comparative view is designed to reveal the evolution of issues and the role(s) of law at this interface of technology, ethics and law.

For evidence to be admissible in a court of law, particularly expert evidence, it must satisfy certain criteria (Faigman, Kaye, Sacks and Sanders 2000, 2002; Federal Rules of Evidence (rules 702 to 706) 2000; Canada Evidence Act 1985). Knowledge of these criteria are important in considering what types and under what conditions DNA evidence can be admitted in the courtroom. At the most fundamental level, the methods of collection and the security of samples affect not only the admissibility and weight, but also the reliability and validity of DNA data. To further add to the confusion of the acceptance of scientific evidence it was not always clear which standard to follow, statutory or case law, at the federal level thereby establishing a framework for states and provinces.

Historically, that is before 1923, expert evidence was judged by the "commercial marketplace test" (Faigman 1994; Saks 1998). Under this test the knowledge or expertise of the

proposed expert had to have a commercial value and the relevant facts or opinions had to be beyond the range of knowledge of the fact finder. A commercial market for the witness's "learning" had to exist, more precisely they had to be able make a living selling the knowledge they possessed (Faigman 1994; Saks 1998). The "marketplace" standard was problematic in areas where the utility or commercial value of the expertise was limited and by equating a commercial value commensurate with scientific validity (Faigman 1994; Saks 1998).

"Just when a scientific principle or discovery crosses the line between the experimental and demonstrable stages is difficult to define. Somewhere in this twilight zone the evidential force of the principle must be recognized, and while the courts will go a long way in admitting expert testimony deduced from a wellrecognized scientific principle or discovery, the thing from which the deduction is made must be sufficiently established to have gained general acceptance in the particular field in which it belongs" (Frye v. United States, 1923).

After 1923, the admissibility of novel scientific evidence was judged in most US states by the Frye standard. The test named after the U.S. Circuit court case, *Frye v. United States* 293 F. 1013 (D.C.Cir.1923, disallowed the use of the polygraph as evidence. The ruling gave a broad range of ways in which evidence could be evaluated, predominantly rooted in the "general acceptance in the scientific community", however, some believed it was so broad as to be basically meaningless (Baird 1992). Essentially the market test was relocated to a different marketplace, from commercial to intellectual or professional. *Frye*'s contribution, however, was three fold. The switch in marketplace allowed knowledge that was too new to be marketed commercially, or knowledge that might never have a commercial application to be considered by the court (Faigman 1994; Saks 1998; *Frye v. United States* 293 F. 1013 (D.C.Cir.1923).

Second, the court by explicitly recognizing, in a legal context, that a body of asserted knowledge has an existence separate from any individual disassociated the expert from the expertise (Faigman 1994; Saks 1998).

Lastly, and likely unintentionally, the shift of marketplace exchanged buyers for sellers as the appraisers of the offerings and their validity. The virtue of the commercial marketplace standard was that buyers were the appraisers of the expertise and its validity where as now those that were offering the expertise were also attesting to its validity (Faigman 1994; Saks 1998).

Until 1995 variations of the *Frye* standard were enacted as tests of admissibility. The three prong test used in *People v. Castro*, 144 Misc.2d 956, 545 N.Y.S.2d 985, 995 (Sup.Ct.1989) asked: Is the theory generally accepted in the scientific community? Are the techniques and experiments reliable in their results? Did the lab performing the test follow the accepted scientific techniques? The three prong test was considered to be the most rigorous in the evaluation of scientific evidence. However the three prong test was not uniformly applied by

the courts (*United States v. Two Bulls*, 940 F.2d 380, 381 (8th Cir. 1991); *State v. Bible*, 858 P.2d 1152 (Ariz. 1993); *Hopkins v. State*, 1991; *State v. Ferguson*, 2000; *State v. Vanderbogart*, 1992; *State v. Cauthron*, State v. Cauthron, 846 P.2d 502, 506 (Wash. 1993)). There were courts which only applied the first two prongs of the test and argued that it was the responsibility of the prosecution and defense attorneys to argue the specifics of each set of results and the probability of human error, sample degradation, statistics and other sources of error.

In Canada the admissibility of expert testimony was at the discretion of the trial judge as there was no rule regarding expert testimony. The case law mostly opted for a liberal "relevancy" test, emphasizing the qualifications of the expert and adding to that a dose of the flexible "reliability" test which focused on whether the expert's testimony was based on proper scientific methodology and reasoning (Bernstein 1996; Gold 2003). As in the U.S., Canadian courts considered variations on the tests including "trustworthiness", "relevancy and helpfulness", "beyond the ken of the jury" and hybrids of the three (Bernstein 1996).

In 1993, The U.S. Supreme Court mandated that scientific validity be the hallmark of expert testimony in its interpretation of the Federal Rules of Evidence in the case of *Daubert v*. *Merrel Dow Pharmaceuticals Inc.*, 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993). The U.S. Supreme Court was called upon to determine the standard for admitting expert scientific testimony in a federal trial with regard to Rules 702 to 706 of the U.S. Federal Rules of Evidence (*Daubert v. Merrel Dow Pharmaceuticals Inc.*, 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993)). The Court proposed a two-part analysis. First, the Court must decide whether the expert testimony reflects scientific knowledge, derived by acceptable scientific methodology, and whether the work amounts to "good science". Second, the Court must determine if the proposed expert testimony is relevant to the task at hand (*Daubert v. Merrel Dow*

Pharmaceuticals Inc., 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993)). The Court suggested several factors that judges should consider when deciding to admit expert testimony: "whether the theory or technique employed by the expert is generally accepted in the scientific community; whether it's been subjected to peer review and publication; whether it can be and has been tested; and whether the known or potential rate of error is acceptable" (Monahan, 1998, Daubert v. Merrel Dow Pharmaceuticals Inc., 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993)). The difference between *Daubert* and *Frye* is that under *Frye*, the party proffering the scientific evidence had to show it was based on the method generally accepted in the scientific community (Frye v. United States 293 F. 1013 (D.C.Cir.1923)). Under Daubert, the focus is on the reliability of the methodology; therefore the proffering party must offer an explanation of the expert's methodology and demonstrate in some "objectively verifiable way" that the expert has both chosen a reliable scientific method and followed it faithfully. Sent back by the Supreme Court to the Ninth Circuit Court, in 1995, it affirmed the Daubert ruling and "stressed the importance of the "fit" between the testimony and the case," in that the standard requires a clear valid scientific connection to the inquiry because scientific expert testimony can be both powerful and misleading (Daubert v. Merrel Dow Pharmaceuticals Inc., 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993); Monahan 1998). The ruling in Daubert appoints the trial judge as the gatekeeper of the court in assessing expert testimony for scientific validity, proper methodology and application to the issue at hand (Monahan 1998). The standards of appellate review of such testimony were established in 1997 in General Electric Co. v. Joiner, 522 U.S. 136 (1997). On the heels of Joiner, the U.S. Supreme Court decided in Kumho Tire Co. v. Carmichael that equal admission standards should hold true for both scientific and non-scientific expert testimony under the *Daubert* test and that the "gatekeeping" function applied to all expert

opinion evidence; technical, scientific or other specialized field (*Kumho Tire Co. v. Carmichael*, 526 U.S. 137 (1999)). "The *Daubert-Joiner-Kumho* trilogy provides a set of basic principles that now control expert testimony in U.S. federal courts" (Gold, 2003). The amendments to the U.S. Federal Rules Evidence in December 2000 use the trilogy principles as the framework for expert testimony.

Not until after *Daubert* did Canada start to make changes, though not as revolutionary, to the admission of expert evidence (*R*. v. *Mohan*, 2 S.C.R. 9. (1994)). In the spirit of *Daubert*, the court in *R*. v. *Mohan* distinctly restated and updated the test for admissibility of expert testimony (*R*. v. *Mohan*, 2 S.C.R. 9. (1994)).

Subsequently, appellate and trial courts in Canada recognized the need to embrace scientific methods and procedures in admitting expert testimony (*R. v. M.(B.)*, S.J. No 124 (C.A.) (1998); *Wolfin v. Shaw*, 21 C.R. (5th) 324 (Ont. C.A.) (1998); *R. v. F.(D.S.)*, 23 C.R. (5th) 37, 132 C.C.C (3d) 97, 169 D.L.R. (4th) 639 (Ont. C.A.) (1999); *R. v. J.E.T.*, O.J. No 3067 (Gen, Div.), Hill J. (1994); *R. v. Hughes (Ruling No. 2)*, B.C.J. No 1699 (S.C.), Romilly J. (1999); *R. v. Murrin*, B.C.J. No 2715 (S.C.), Henderson J. (1999); *R. v. C.J.S.*, A.J. No. 634 (Prov. Ct. (Youth Div.)), Landerkin Prov. Ct. J. (1995); *R. v. Sood*, O.J. No. 5580 (Gen.Div.), Donnelly J. (1997); *R. v. Campbell*, O.J. No. 6229 (Gen.Div.), Chilcott J. (1998); *R. v. Tercetra*,15 C.R. (5th) 359 (ont. C.A.) aff'd [1999] 3 S.C.R. 866 (1998)). Six years later, the Supreme Court removed any doubt by specifically referencing *Daubert* in *R. v. J-L.J.* (*R. v. J-L.J.*, 2 S.C.R. 600. (2000)). The court cited *Daubert* "as a relevant authority and referred to many of the same factors for analysis referenced by the *Daubert* court" (Gold 2003).

In Canadian law the concept of relevance links the admissibility of expert opinion evidence with the demands of good science. For the evidence to be relevant it must affect the

probability of whether a fact in issue is true or not. Relevance includes both the relevance of content and the method or procedure by which the opinion was formed. The essence of scientific information is its falsifiability or testability (Gold 2003). An improperly founded opinion, one where the opinion or information is not scientifically reliable or valid, is not logically relevant, will not assist the trier of fact and thus the issues of validity and reliability are tantamount.

According *R*. v. *Mohan* as elaborated by *R*. v. *J-L.J*. the admission of expert evidence in Canada requires consideration of four components: necessity (in aiding the trier of fact), relevance, the absence of an exclusionary rule, and an appropriately qualified expert (Gold 2003). The necessity of expert evidence is based on whether the judge or jury would be unaware of the information to be provided as a result of their own experience or knowledge (Gold 2003). Necessity has two aspects: the characterization of the evidence as being outside the experience and knowledge of the trier of fact and that the expert evidence testimony will assist in achieving the correct conclusion on the matter and secondly, that the information be received expressly by way of expert evidence and not simply that it is received at all. "...[W]here the trier of fact can form its own opinion on the matter without any help, the proposed expert evidence is unnecessary" (Gold, 2003). If the trier of fact can receive the required information in some other fashion then the expert evidence is unnecessary. Thus to be admissible expert evidence must be necessary in that the jury would be unable to reach a correct result in its absence and that it is in the form of expert testimony.

Relevance in a judicial context is composed of both logical and legal relevance. First, in a relevance inquiry, there must be a finding of logical relevance. Logical relevance is established via examination of the reliability and validity of the testimony offered, which is brought about by the adherence to the rigors of science. Scientific methodology and adherence

to it is designed to assure validity and reliability. Legal relevance is then assured by way of a harm-benefit analysis. The analysis pits the benefits of the evidence (in terms of materiality, weight, and reliability) against its harms (in terms of the dangers associated with expert testimony, such as uncritical acceptance by the trier of fact, its potential prejudicial effect, and the practical costs of presentation) and is essential for the determination of legal relevance. The harm-benefit analysis evaluates whether the probative value of the evidence is outweighed by its prejudicial effect; whether the time and effort involved in receiving it is commensurate with its value; whether prestige of the witness has an influence; the complexity and potential for confusion of the evidence, or if any other cause will be out of proportion to its reliability (Gold 2003). The rigorousness of the exclusionary analysis is applied less stringently in favor of the defense (Gold 2003). "A trial judge should be particularly cautious when excluding expert defence evidence on the basis of a cost benefit analysis... [T]he exclusionary rule only operates to exclude defence evidence where the prejudice effect of the evidence substantially outweighs its probative value" (*R*. v. *M.(B.)*, S.J. No 124 (C.A.) (1998)).

Exclusionary rules apply to any evidence proffered to the court thus occasionally expert opinion evidence will be rendered inadmissible even when it otherwise complies with the requirements of admissibility (Gold 2003).

The test for a "properly qualified expert" as laid out in *Mohan* and states that "[T]he evidence must be given by a witness who is shown to have acquired special or peculiar knowledge through study or experience in respect to the matters on which he or she undertakes to testify" (*R.* v. *Mohan*, 2 S.C.R. 9. (1994)). Further, the qualifications of the expert must be proved on a balance of probabilities (*R.* v. *Terceira*, 1998). Thus numerous aspects of a witness's background are relevant to determining expertise such as academic qualifications,

publications, books and articles authored, papers and presentations at conferences and meetings, additional training, practical experience, and professional memberships in societies and organizations. "Expert witnesses must not only be qualified, but also qualified with respect to the areas or subjects upon which they are to testify" (Gold 2003). Novel scientific measures, in particular, should undergo "special scrutiny" (Giannelli 2000). The requirements outlined above are strictly applied in the case of new forms of expert evidence regarding novel scientific measures (Giannelli 2000; Gold 2003).

1.2.1 Legal Background on DNA as Scientific Evidence

As few, if any, rulings on DNA testimony have been handed down in either U.S. or Canadian federal courts, the introduction of DNA as evidence in the courtroom is occurring primarily at State and Provincial levels. This section identifies the legal issues pertaining to the admissibility of and the weight of DNA profiling evidence.

1.2.1.1 Admissibility

In the United States introduction and admission of DNA evidence into the courtroom had a rocky beginning due mostly to reservations about the accuracy of the tests and the complexity with respect to its presentation to a jury. Though the initial introduction was difficult DNA was eventually accepted. The first case, a sexual assault, in which DNA typing, specifically RFLP, was proffered and admitted to the court was *Andrews* v. *State* (Florida) in 1987 (*Andrews* v. *State*, 533 So. 2d 841, Fla Dist. Ct. App., 1988). The initial trial resulted in a hung jury, as they were unable to reach a verdict solely on the DNA evidence, but in the subsequent retrial, with the DNA and the match probability evidence, Andrews was convicted. On appeal both the ruling and the conviction were upheld. Thereafter, forensic DNA evidence was accepted without dispute at trial until two years later when the challenge of DNA testing began in earnest. The

experts selected a variety of dimensions upon which to challenge the infallibility of DNA profiling, from the care with which laboratories developed standards and executed procedures, to the appropriateness of the assumptions about the population genetics.

In the decision of *People* v. *Castro*, 144 Misc.2d 956, 545 N.Y.S.2d 985, 995 (Sup.Ct.1989), the judge rejected the admission of DNA evidence because it failed the third prong of the *Frye* test that deals with the performance of accepted techniques by the laboratory (Gass and Schultz 1992). "In a piercing attack upon each molecule of evidence presented, the defense was successful in demonstrating to this court that the testing laboratory failed in its responsibility to perform the accepted scientific techniques and experiments" (*People* v. *Castro*, 144 Misc.2d 956, 545 N.Y.S.2d 985, 995 (Sup.Ct.1989)). The attorneys in the *Castro* case Mr. Peter Neufeld and Barry Scheck were the principle players involved in orchestrating the collusion of the experts for the defense and prosecution that jointly issued a statement refuting the corporate laboratories results (Tucker 125; Hubbard and Wald 131; Lander 1992; Parloff 1989). The New York Superior Court in a *Frye* hearing, separate from the initial case, was the first to reject DNA evidence in a criminal proceeding. The separate hearing to determine the admissibility of the DNA data as evidence generated 5000 pages of transcript and suggested detailed procedures for future courts (Gass and Schultz 1992).

Following *Daubert* one of the Justices commented "[o]ur task, is to analyze not what the experts say, but what basis they have for saying it" (*Daubert* v. *Merrel Dow Pharmaceuticals Inc.*, 43 F.3d 1311 at 1316 (1995), on remand from *Daubert* v. *Merrel Dow Pharmaceuticals Inc.*, 509 U.S. 579, 113 S.Ct. 2786, 125 L.Ed.2d 469 (1993)). From this it is not surprising that some courts insisted that part of the demonstration of scientific soundness required under *Daubert* was a showing that the proper application of an approved method occurred on the

particular occasion (United States v. Martinez, 3 F.3d 1191 (8th Cir.1993)). In State v. Schwartz, the court conceded that DNA typing had gained general acceptance however "the laboratory in this case did not comport" with "appropriate standards" and thus the evidence was excluded (State v. Schwartz, 447 N.W.2d 422,428 (Minn.1998)). It has been further argued that insufficiently rigorous DNA testing procedures are issues of admissibility under Rule 702 (Imwinkelried, 1991; Scheck 1994; Thompson 1997), however, this interpretation of Daubert was rejected in United States v. Shea (United States v. Shea, 957 F.Supp. 331, 340-41 (D.N.H. 1997)). Instead the proficiency and protocols of a specific laboratory are factors that affect the probative value under Rule 403 (Berger 1997; Imwinkelried 1996). Though the decision in Daubert was handed down in 1993, many state courts continue to maintain the Frye standard (Alaska, Arizona, California, Colorado, District of Columbia, Florida, Illinois, Kansas, Maryland, Michigan, Minnesota, Mississippi, Nebraska, New Jersey, New York, Pennsylvania, and Washington) instead of conforming to the new federal standard adopted by other states and codified in the amendments to the Federal Rules of Evidence in 2002 (Inman and Rudin, 2002). Regardless, it is evident that demonstration to the court that the proper application of an approved method was conducted on the particular occasion is an essential component necessary for balancing the probative and prejudicial value of DNA testimony.

By contrast to the U.S. courts in Canadian courts it is likely that failure to comply with approved methodology would render DNA testimony inadmissible under a relevance inquiry, as there must be a finding of logical relevance. In *R.* v. *F.*(*D.S.*) the trial judge held, in rejecting the evidence, that absence evidence of an objective test of the reliability of the opinion and of an objective means to evaluate the reliability of said opinion disqualified it as evidence (*R*. v. *F.*(*D.S.*), 23 C.R. (5th) 37, 132 C.C.C (3d) 97, 169 D.L.R. (4th) 639 (Ont. C.A.) (1999)). Logical

relevance demands reliability and validity from the testimony offered, if the evidence is unreliable due to failure to follow standard protocol, adherence to the rigors of science demand that the testimony be considered irrelevant and therefore inadmissible (*R. v. Sood*, O.J. No. 5580 (Gen.Div.), Donnelly J. (1997)). "Relevance in respect of an expert opinion is a function not just of the substance of the opinion, but rather essentially of the methodology utilized to reach it" (Gold 2003). Thus in Canadian law, the concept of relevance links the admissibility of expert testimony with the demands of good science to exclude evidence that is unreliable and invalid and therefore irrelevant.

In the United States several cases explicitly challenged the statistical approaches to DNA probabilities and in several the evidence was held inadmissible (NRC II 1996; Kaye 1993). Arguments were made that the statistics involved, that often submit probabilities in the range of one in a million, would unduly influence jurors as to the absolute guilt of a defendant. In truth DNA data's actual power is that it can absolutely exonerate a suspect but not absolutely condemn them. As such the probability estimates are limited by test performance and the prevailing error rate, which ground the statistics in the real world, despite DNA testings theoretical reliability and limitations (Thompson et al. 2003).

The California Supreme Court closely examined the probabilistic evidence, in *People* v. *Collins*, which turned out to have been based on "grossly inaccurate statistical data" and subsequently disallowed the admission of the DNA testimony (*People* v. *Collins*, 438 P. 2d 33 (68 Cal. 2d 319 1968; Schultz 1992). The Massachusetts Supreme Court in the *Commonwealth* v. *Curnin* reversed a lower courts ruling and insisted on an inquiry equivalent to a *Frye* test (*Commonwealth* v. *Curnin*, 409 Mass. 218, 219 n. 2, 565 N.E.2d 440, 441 n. 2 (1991)). The judge stated as reason for his decision "the potential prejudicial effect of DNA evidence, which

has "an aura of infallibility" (*Commonwealth* v. *Curnin*, 409 Mass. 218, 219 n. 2, 565 N.E.2d 440, 441 n. 2 (1991); Gass and Schultz 1992). The Massachusetts Supreme Court reversed the conviction based on poor substantiation by Cellmark laboratory of its statistical evidence. "In *United States* v. *Jakobetz*, 747 F.Supp. 250, 258 n. 17 (D.Vt.1990), a federal appellate court upheld the admissibility of RFLP DNA evidence, including statistical calculations to assess the significance of the results" and the following year in *People* v. *Axell* (235 Cal. App. 3d 836, 1 Cal. Rptr. 2d 411 (1991)), DNA and the population statistics were admitted as well (Inman and Rudin 2002).

The debate centered on the simplified population-genetics model used to determine the frequencies of DNA RFLP profiles (Faigman et al. 2002). The contention was that the assumption of statistical independence of the genetic loci didn't take into account possible substructure within the population (United States v. Yee, 134 FRD 161 (ND Ohio 1991); Kaye 1993; Thompson 1993). The statistics, it was argued, were not based on sufficient study of actual allele distribution in various ethnic subpopulations thus the statistics presented by the laboratories in court might significantly misrepresent the real-world frequency with which a match would occur (Moonkin 2002; Thompson 1996). For scientists to provide information as to the meaning of a match they must be able to assess how often a match might occur. This is done, by determining how frequently each allele is found in the population of interest and combining the likelihood of each of the alleles into a single combined statistic. If loci are inherited independently, the probabilities for each loci can be multiplied according to the product rule, but if loci are not independent, using the product rule will result in an inaccurate probability statistic that underestimates the frequency with which matches will occur in the actual population (Mnookin 2002; Kaye and Sensabaugh 2000).

The arguments being raised in trials, pre-trial admissibility hearings and appeal cases defined the scope of the disputes in the scientific community about DNA profiling. The debate was played out in the pages of Nature, Science and other more specialized journals and crossed back and forth between the boundaries of science and the law (Lewontin and Hartl 1991; Charkraborty and Kidd 1991). In an effort to generate consensus on the applications of DNA profiling the US National Research Council convened a panel to investigate the contentious issues (NRC 1992). "Although the instruments and techniques used in forensic DNA profiling were commonplace in biomedical research and diagnostics, the investigations brought into relief questions about the specificity of forensic practice" (Lynch 2003).

With respect to the population frequency estimates the 1992 National Academy of Sciences report proposed a compromise with conservative computational methods. The intention of the proposed methods, the ceiling principle and the interim ceiling principle, were to set a lower limit on the size of the profile frequency. This would be accomplished by setting threshold values for allele frequencies used in calculations thus the overall profile frequency would be capped. However, some population allele frequency research was necessary to enact the ceiling principle. In the meantime, the interim ceiling principle would be pertinent. "In applying the multiplication rule, the 95% upper confidence limit of the frequency of each allele should be calculated for separate US 'racial' groups and the highest of theses values or 10% (which ever is larger) should be used" (NRC 1992). Critics, namely scientists, were vocal against the excessively conservative methodology, which put an artificial limit on the rareness of a particular genetic profile, as baseless with no scientific foundation (Faigman et al. 2002; Chakraborty and Kidd 1991; Cohen 1992; Morton 1992, 1995; Evett, Scranage and Pinchin 1993; Kaye 1993, 1995a; Lempert 1993; Weir 1993a; Balding and Nichols 1994, Devlin, Risch

and Roeder 1994; Lander and Budowle 1994; TWGDAM 1994c). They claimed that the 10% value was completely arbitrary, and there was no scientific justification of its choice as a ceiling value. Further the calculation of an upper 95% confidence limit for an individual allele was justified as a standard statistical procedure but manipulation of those values was not. The ceiling principles failed to make use of the contemporary allele-frequency data available from different groups and subgroups and standard procedures long used by population geneticists to study subdivided populations were ignored. In light of the critiques a second National Research Council panel convened (NRC 1996). The second panel, in addition to reviewing other issues, agreed with the critics and the recent TWGDAM report that there was sufficient data to negate the need for either ceiling principle and that they were flawed. They further reminded the forensic community that "[t]he ceiling principles were intended for VNTRs with many alleles, no one of which has a very high frequency. They are not applicable to PCR-based systems, which ordinarily have few alleles" (NRC 1996).

Modifications in the technical procedures have allowed the use of contemporary molecular DNA profiling methodologies that have evaded/dodged/eluded previous sources of conflict. The multiplex short tandem repeat system emerged in the mid-1990's as the methodology of choice for criminal justice systems in the UK, the EU and the US. The STR methodology, which uses hypervariable DNA sequences of relatively short length as markers, combines multiple markers to generate a profile as individual markers have a limited discriminatory power. "STR markers chosen for a profile system – such as the FBI's system which uses 13 different markers – are taken from distinct chromosomal sites…" (Lynch 2003). However, the change in technology and marker type brought about new issues and challenges.

Polymerase chain reaction-based testing, STR and mtDNA profiles in general, faced different obstacles than RFLP testing, focusing more on the specific application of the methodology, the electronics and software of the automated systems, disclosure of developmental validation and proprietary reagents and less on population genetics and substructure (Inman and Rudin 2002). Many courts in the US have admitted STR typed DNA evidence but contradictions within the same state, and sometimes the same jurisdiction, have created confusion in the legal community (Inman and Rudin 2002; People v. Hill, 89 Cal.Appl. 4th 48, 107 Cal.Rptr. 2d 110 (Ct.App.2001).; People v. Allen, 72 Cal. App. 4th 1093, 1999; State v. Roth, 2000; Commonwealth v. Rosier, 1997; State v. Jackson, 1998). The first ruling declining STR evidence was People v. Bokin (No. SCN 168461. Cal. Sup. Court, San Francisco Co., 1999) in 1999. Bloodstains from the trunk of a car were analyzed using two STR systems; neither was admitted by Judge Dondero due to lack of peer-review and failure to meet general acceptance standards under Frye. However, DNA results using other generally accepted systems were admitted. Almost simultaneously conflicting decisions were rendered in People v. Bertsch and Hronis, People v. Elizarraras, People v. Hill, People v. Hackney, and People v. Hunt, all in the state of California (People v. Bertsch and Hronis, No. 94F07295, Cal. Super. Ct., Sac. Co., 1999; People v. Elizarraras, No. 50651 Cal. Super Ct. Tulare Co. 2000; People v. Hill, No. 232982, Cal. Sup. Court, Santa Barbara Co. 2000; People v. Hackney, No. 97F02466 cal. Sup. Court, Sacramento Co. 1999; People v. Hunt, et. al., No. SA034500, Cal. Sup. Court, Los Angeles Co., 2000). Despite a 1999 California Court of Appeals ruling that STR analysis was generally accepted in the scientific community (People v. Allen) conflicting rulings within the same jurisdiction occurred (People v. McClanahan (No. 162412, Cal. Sup. Court, San Francisco, Co., 1998) and People v. Moevao, (No. 168277, Cal. Sup. Court, San Francisco, Co., 2000)). In

the following year, a Vermont District Court failed to admit STR testing while admitting other DNA results. Again general acceptance standards were not met, but this time Judge Kupersmith used *Daubert* criteria as adopted by the State of Vermont. He opined that the newer systems lacked substantial peer-review publications and failure to provide the court with the primer sequences gave credence to the general lack of acceptance (State v. Pfenning, 2000). In People v. Shreck (Colorado) STR typed DNA failed admission under both Frye and Daubert. Judge Hale of the Colorado District Court based his ruling on the test kit providers failure to furnish the validation studies and primer sequences as well as the lack of scientific publications (People v. Shreck, No. 98CR2475, Co. District Court, Boulder County, 2000; Reversed, Case No. 00SA105, Co. Supreme Court, 2001). Again alternate DNA typing systems were admitted and a conviction was tendered. A contrary ruling admitting STR analysis, the same system that was declined in Shreck, was issued in People v. Flores by a Colorado district court in 2000 (People v. Flores, No. 99-CR2022, Co. District Court, Adams Co.). The appeal of the verdict in Shreck was upheld but in 2002 the Colorado Supreme Court vacated the original trial court's ruling asserting that, under *Daubert* and Federal Rule of Evidence 702, all STR systems should be admissible.

Mitochondrial DNA (mtDNA), one of the newer DNA technologies, was admitted in 1996 at the trial court level in *State* v. *Ware* (WL 233592 Tenn. Crim. App., 1999 WL 233592 (NO. 03C01-9705CR00164). The DNA from root bulbs of several hairs linked a suspect to the rape of a 4-year-old girl. The decision was upheld on appeal and a second Tennessee decision *State* v. *Scott* was also upheld (S.Ct. of Tennessee, No.96-C-1362, 2000). Overall the latest DNA typing technologies are gaining judicial approval (*Adams* v. *Mississippi*, No. 200-KA-00242-COA, Miss App., 2001; *State* v. *Underwood*, 518 S.E.2d 231, N.C. Ct. App., 1999; *State*

v. Ware, WL 233592 Tenn. Crim. App., 1999 WL 233592 (NO. 03C01-9705CR00164);

Curriden, 1996). Mitochondrial DNA has been admitted as evidence in 18 states in trial courts and affirmed in appellate courts in Tennessee, North Carolina and South Carolina. In the *State of Florida* v. *James Deward Crow*, the Honorable Judge O.C. Eaton excluded mtDNA, the results of which did not meet the Frye Standard and was therefore inadmissible as evidence. His ruling was based on his understanding that the FBI database was too small and was insufficient to provide reliable statistical conclusions. In addition, Judge Eaton held that the "counting method" failed to provide a meaningful comparison and would more likely confuse the jury than assist it.

All in all, mtDNA profiling has different capabilities and limitations as compared with nuclear DNA testing. A number of issues pertaining to mtDNA have been raised, including contamination, population genetics, and methodology. Though these are similar to the inquiries of nuclear DNA the specifics pertaining to mtDNA are unique. The chief concern, however, is the interpretation of evidence samples that exhibit heteroplasmy (an authentic mixture of types) (Inman and Rudin 2002).

1.2.1.2 Weight

The majority of the admissibility debate has centered on the DNA data itself but not on the influence it can have on a jury. As mentioned above the weight accorded to the probability statistics must be considered. There are some serious concerns regarding jury comprehension of DNA data as evidence and the weight it should be granted when deliberating a verdict (Lewontin 1994; Thompson and Ford 1989). The 1996 National Research Committee's report articulated three major sources of prejudice: that the jury will be over awed by the small numbers and ignore other aspects of the case, that the jury will misconstrue the probability of a random match as the probability that the defendant is not the source of the incriminating DNA and that the statement

of a probability ignores the possibility of a match being declared due to sample mishandling or other "blunders" (NRC II 1996).

The first suggestion, that jurors do not understand probabilities in general, and that infinitesimal match probabilities will so impress jurors that they will not appreciate the other evidence in the case or any innocent explanations for the match, is not entirely unfounded (Government of the Virgin Islands v. Byers, 35 VI 240, 941 F.Supp. 513 (DVI, 1996); Commonwealth v. Curnin, 409 Mass. 218, 219 n. 2, 565 N.E.2d 440, 441 n. 2 (1991); Faigman et al. 2002). The limited research conducted on this hypothesis to date has been inconclusive (NCR II 1996; Goodman 1992; Faigman & Baglioni 1988; Thompson and Schumann 1987; Kaye and Koehler 1991); Schklar and Diamond 1991; Faigman et al. 2002). In the case of People v. Wesley, 83 N.Y.2d 417, 423, 611 N.Y.S.2d 97, 100 (1994) a mildly retarded man was convicted of murder based on a match of blood residue on the defendant and that of the victim. The defendant claimed he was present at the scene, but denied committing the crime. Wesley asserted that someone else committed the crime and evidence of another person was retrieved from the crime scene (Shultz 1992). In this instance the jury placed undue weight on the evidence that merely indicated that the defendant was present at the crime. The judge in the case was clearly influenced by the evidence proclaiming DNA to be the "single greatest advance in the 'search for truth'...since the advent of cross-examination" (People v. Wesley, 83 N.Y.2d 417, 423, 611 N.Y.S.2d 97, 100 (1994)). The court did, however, order the statistical probability evidence be reduced by a factor of ten to take into account that the population used in the calculations might not be in Hardy-Weinberg equilibrium (Shultz 1992). In so doing the judge tried to limit the possibility of prejudice and lessen the weight of the statistics under jury consideration (People v. Wesley, 83 N.Y.2d 417, 423, 611 N.Y.S.2d 97, 100 (1994)). "However

despite their lay status with reference to scientific and mathematical data, judges are legal professionals experienced in assessing legal concepts like relevance and prejudice. They have the responsibility for protecting individuals' rights against the risks of jury error or prejudice" (Shultz 1992). A few of the suggestions offered by the NRC II to counteract the influence of small probabilities were rigorous cross-examination, utilizing defense experts to oppose evidence and jury instruction (NRC II 1996). The NRC II went so far as to draft exemplar instructions for judges to convey to jurors information regarding match probabilities (NRC II 1996). Exclusion of the random match probabilities has been suggested as a way to avoid the statistical issues which are in some cases as difficult to present as they are for jurors to understand (Clark 1997; McCormick 1992; United States v. Martinez, 3 F.3d 1191 (8th Cir.1993).; State v. Foster, 1996). Suggested exclusion of the random match probabilities has not been well received by the courts (Unites States v. Chischilly, 1994; Hughes v. State, 1999; State v. Weeks, 1995). Exclusion of the random match probability resulted in the abortion of the initial trial, in the State of Florida v. Andrews in 1988, the jury was unable to reach a verdict based only on the DNA evidence with no the statistical data (Andrews v. State, 533 So. 2d 841, Fla Dist. Ct. App., 1988). The retrial, which included both the DNA and the probability statistics as evidence, resulted in a verdict.

It has been further argued that jurors will be confused as to the meaning of the random match probability - by thinking that it gives the probability that the match is random or the probability that the defendant is not the source. The random match probability is the conditional probability of a matching profile, given that the individual tested is not the source of the profile. "Likewise, the random match probability P(genotype given unrelated source) does not necessarily equal P(unrelated source given genotype)" (Faigman et al. 2002). This fallacy is known as the fallacy of the transposed conditional or the inverse fallacy (Koehler 1993). "To

avoid this fallacious reasoning by jurors, some defense council have urged the exclusion of random match probabilities, and some prosecutors have suggested that it is desirable to avoid testimony or argument about the probabilities, and instead to present the statistic as a simple frequency- an indication of how rare the genotype is in the relevant population (Clark 1997; Kaye 1987; Faigman et al. 2002). It is clear from cases such as the *State of Florida v. Andrews* that jurors need some information concerning the frequencies of proffered profiles to allow them to contextualize the DNA evidence. The 1996 NRC report noted that "few courts or commentators have recommended the exclusion of evidence merely because of the risk that jurors will transpose a conditional probability" and it observed that "the available research indicates that jurors may be more likely to be swayed by the 'defendant's fallacy' than by the 'prosecutor's fallacy'" and underestimate the weight that should be accorded to the evidence (NRC II, 1996; McCormick 1992; Faigman et al. 2002 (defendants fallacy)).

In addressing concerns that the random match probability ignores the possibility of a match being declared due to sample mishandling, mislabeling, cross-contamination, the possibility of false incrimination or other events that would yield a false positive result the court has recognized the need for a distinction between minor mistakes and those that are so fundamental as to preclude admissibility. "Shortcomings such as mislabeling, mixing the wrong ingredients, or failing to follow routine precautions against contamination may well be amenable to evaluation by jurors without the assistance of expert testimony. Such readily apparent missteps involve 'the degree of professionalism' with which otherwise scientifically accepted methodologies are applied in a given case, and so amount only to '(c)areless testing affect[ing] the weight of the evidence and not the admissibility'..." (*People v. Venegas*,118 Cal.4th 47,74 CalRptr.2d 2623,954 P2d 525 (Cal 1998)). In the *State v. Kinder* the justice noted that "the

manner in which the tests were conducted goes more to the weight of the evidence, which is in the first instance a discretionary call for the trial court and ultimately for the jury" (*State* v. *Kinder*, 924 S.W.2d 313 (Mo.1996)). It is not entirely clear whether the proper application of the approved method and thereby any mistakes that were made are part of the demonstration of soundness under *Daubert* or weight as suggested by the courts (*United States* v. *Martinez*, 3 F.3d 1191 (8th Cir.1993).; Berger 1997). Further, mislabeling or mishandling may not necessarily be amenable to detection when limited sample precludes the possibility of re-testing, as well as limited resources of the defendant (Goncalves 2008; Khanna and McVicker 2007; Thompson 2006; Seattle Post Intelligencer 2004).

It has been asserted that when the probability of an error or false positive is larger than the match probability the match probability is logically irrelevant and if not irrelevant, prejudicial (Koehler 1995; Lewontin and Hartl 1991; Thompson, Taroni and Aitken 2003; Thompson 1994; Thompson and Ford 1989; Lempert 1991, 1997). The match probability on its own cannot provide the trier of fact with the requisite information to determine if a laboratory or other such error has taken place. Excluding the match probability would eliminate a juror's ability to assess the possibility of kinship or the coincidence of a match at random (NRC I 1992; NCR II 1996; Higuchi 1991). Probability estimates of a false positive, however, would allow the jury to assess the chance that the defendant is not the source even though the forensic sample is reported to match the defendant's DNA profile. As for the prejudicial impact of the random match probabilities it is possible, as described previously, that juries might be so taken with the small match probability that they ignore all other dissuasive evidence including the false-positive estimate. It has been suggested that the potential for prejudice is so grave as to warrant the submission of the laboratory's false positive rate exclusively (Lampert 1991, 1997). This

argument, though, has not been considered substantial enough to eliminate random match probabilities as evidence and if anything research shows that both estimates, not a combined figure, are useful for jury's decision making (Schakar and Diamond 1999). The prejudicial impact of the random match probability can be addressed directly and counterbalanced by judicial instruction (NRC II 1996).

Disagreement exists as to how the probability of a false positive should be determined either through quantitative estimates or proficiency testing and to what extent it should apply to the case at bar (Balding 1997; Berger 1997; Thompson, Taroni and Aitken 2003; Thompson 1997; Kaye 1997). Proficiency testing to determine the probability of false positives raises a variety of legal issues. It has been suggested as a prerequisite to the admission of DNA evidence that forensic laboratories participate in a program of proficiency testing, that the proficiency results be admissible as estimates of the likelihood of laboratory error in the case at bar, and that random match probabilities be inadmissible unless presented with proficiency tests to estimate the probability of a false match (McCormick 1992; Jonakait 1991; Scheck 1994; Thompson, Taroni and Aitken 2003; Thompson 1997). A figure that combines error rates and match probabilities has been rejected as 'inappropriate' and juror's discretion as to the weight accorded to the individual estimates supported (NRC II 1996; Morton 1997; Balding 1997).

Placing admissibility of DNA evidence subservient to proficiency testing is unusual practice in the United States. Typically issues relating to the proper and faithful conduct of the accepted and validated protocol is usually said to affect the matter of evidentiary weight rather than admissibility (*United States* v. *Shea*, 957 F.Supp. (D.N.H. 1997)). However, in extreme cases the court may decide that the laboratory erred in such a fashion as to render the probative value so slight as to warrant inadmissibility of the DNA profile (Imwinkelride 1991, 1996).

The National Research Council's first report stressed that proficiency testing be done to estimate laboratory error rates and that this information be presented in court alongside DNA testing data but the 1996 report failed to echo this assertion. Instead it raised questions as to the probative value of proficiency testing as an estimate of laboratory error (NCR 1996). By raising concerns as to the probative value of estimates of error rates based on proficiency tests they became susceptible to exclusion under the Federal Rule of Evidence 403 relating to probative value versus prejudice. Exclusion of proficiency testing results may also be achieved through restrictions of character evidence. "If the theory of logical relevance is merely that the laboratory's past commission of errors increases the probability that the laboratory erred on the occasion in question, then the theory amounts to forbidden character reasoning" and is thus prohibited under Federal Rule of Evidence 404 (Faigman et al. 2002). Further, an attempt to show a general tendency to commit errors on the part of the laboratory by introduction of proficiency testing is likely inadmissible under character Rule 405. Proficiency testing, however, may be admissible as a counterbalance to miniscule random match probabilities and in situations where genuine disagreements as to the profile of a DNA sample exist results of proficiency testing may support a false positive finding. Moreover, Federal Rule of Evidence 403 may support admission of proficiency testing results when calling into question the probative value of the testimony regarding industry error rates and the quality of the individual laboratory's testing.

1.3 Ethics and DNA profiling

With respect to DNA in the justice system there has been little written concerning ethics as compared to the technological, procedural and statistical issues. In part this stems from the interface between forensic science and the justice system, in that the focus for scientists has been

science education of the attorneys and judges while the justice system has focused on procedural issues. Judicially it has been assumed that procedural exactitude will eliminate errors thereby ensuring that decisions are ethical (Walsh 2005). In forensic science, however, the evaluation of practices within the judicial context have led those in the field (forensic scientists, population scientists, geneticists, attorneys and judges) to raise issues not just of reliability and validity of methodology but to concerns of protections of privacy and confidentiality. The balance between protection of the group versus protections of the individual means that in the justice system personal freedoms and security are pitted against one another.

An ethics analysis of the use of DNA technology should in general cover more than just privacy and confidentiality concerns, and move beyond issues of stigmatization and discrimination to evaluate the underlying principles of respect for persons and justice. In chapter 4, I attempt to address some of the unprobed ethics areas.

The National Research Council's 1992 report tried to address some of the issues: ethical, legal and societal, surrounding the introduction of DNA technology in the criminal justice system. Though they tried to scrutinize the ethics in a fundamental manner they were unable to go beyond comparing the use of DNA with other identification technologies employed in forensic science, allowing custom or established practice to stand for proof that no fundamental ethical principle was violated. "A threshold question for any ethical inquiry is whether the action or practice under discussion is intrinsically wrong. An action is fundamentally wrong if it violates fundamental ethical principles" (NRC I). Fundamental principles include respect for persons, nonmaleficence, beneficence, and justice (Beauchamp and Childress, 2004). Violations of these principles include torture, the exploitation of humans as means, and enslavement.

This is not to say that their conclusion, that the use of DNA is unlikely to violate any fundamental ethical principal, is wrong but they failed in their analysis to establish the veracity of their assertion. The sole argument posed was that DNA "techniques are extensions and analogues of techniques long used in forensic science, such as serological and fingerprint examinations, handwriting analyses, photography, and examination of teeth. Ethical questions can be raised about other aspects of this new technology, but it cannot be seen as violating a fundamental ethical principle" (NRC I 1992). In light of the questions raised concerning the reliability and validity of forensic handwriting analyses and fingerprinting, comparisons and prior acceptance lend little support (Harris 1958; Risinger et al. 1989; Evett et al. 2000; Faigman et al. 2002; Mnookin 2003; Gold 2003). In a scientific examination of signatures, Harris falsified the core claim of handwriting identification, that no two signatures are alike, when he noted the indistinguishable similarity of many signatures (Harris 1958). Further, the paucity of valid scientific evidence supporting fingerprint identification has led to comments on the subjective and ill-defined nature of fingerprint examination; with one author concluding that "A vote to admit fingerprints is a rejection of conventional science as the criterion for admission" (Faigman et al. 2002; Lewis 1997; Saks 1998).

The NRC I proposed to extend their ethical analysis by utilizing two opposing ethical perspectives: an examination of the rights of those affected and a consequentialist harm benefit analysis. With respect to moral rights the NRC I examined whether new rights were generated by the introduction of DNA technology and if established rights were diminished, endangered or enhanced. The Council determined that no new moral rights were generated through the implementation DNA applications in forensic science but they did not include a discussion of their reasoning process. The assessment of potentially endangered rights focused on the right to

privacy, confidentiality and the right not to be wrongly convicted. Again, the comparison with established techniques was employed as means to assert that intrusion into privacy and the resulting confidentiality breach was not enhanced despite acknowledging, in the first paragraph, that for many "DNA typing technology has the potential for uncovering and revealing a great deal of information that most people consider to be intensely sensitive" (NRC I 1992). The NRC I stated that "[t]he current use of DNA technology appears to pose no greater threat to the right to privacy than does normal fingerprinting, placement of photographs in evidence, collection of blood or saliva samples, or other established forensic techniques. DNA technology is no different in principle from those other techniques, although it holds the promise of providing a more definitive identification than most others" (NRC I 1992). As for the right not to be wrongly convicted of a crime the Council asserted that a high standard of proof imposed before a person may be found guilty would serve as protection. They stressed that the techniques used must have proven reliability (comprising accuracy, precision, specificity, and sensitivity) and consensus of the scientific community (NRC I 1992).

The Council acknowledged the potential for misuse of DNA samples retained by the criminal justice system, focusing on confidentiality and privacy. They cited the President's 1983 Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, which noted the increased "risk of breaches of confidentiality and misuse of the information" (President's Commission 1983; NRC I 1992). The difficulty is ensuring that the DNA samples and generated data are secure thereby avoiding privacy intrusions and violations of confidentiality. The NRC I acknowledged that failure to safeguard information and prevent its unauthorized release or dissemination would be seen as a violation of individual rights in the forensic context (U.S. Congress, Office of Technology Assessment 1990; de Georgey 1990). It

is expected that the potential risks of forensic DNA technology will increase as our knowledge of the genome expands and as more databanks that retain samples, not just profiles, are established (Simoncelli 2006; Simoncelli and Wallace 2006; Annas 1993). The loci, presently used for DNA profiling, are not known to be associated with any physical or behavioral traits (Steinhardt 2004; Fox 2001). In the future, however, it may be feasible to relate DNA profile information with physical characteristics, race, parentage or disease susceptibility consequently facilitating suspect identification by police (Butler, Colbe, and Vallone, 2007; Dale, Greenspan and Orokos 2006; Spencer 2004). The NRC I cautioned that "[w]ithout strict limits, however, DNA information can be more intrusive into privacy, in that it provides more information about a person".

Despite their cautions, the Council went on to suggest that DNA technology, in one way, may be less intrusive as searching the databank for a profile match is less inconvenient for potential suspects than detention and questioning. On the other hand, it has been argued by others that the collection of DNA from all persons in an area as part of a "DNA dragnet or sweep" to identify a potential suspect is inconvenient for all even if the sample is just a buccal swab (Walker 2004). Refusal to provide a sample is viewed as suspicious and will automatically warrant investigation concerning that individual thereby effectively eliminating the individuals right to refuse (Electronic Privacy Information Center 2005). The choice no longer becomes a voluntary one (Steinhardt 2004). In addition retention of samples and profiles used for elimination places people who have had no reason to be involved with the criminal justice system to be considered as suspects every time a profile is compared with theirs, regardless of whether they are notified or personally inconvenienced each time (National Commission on the Future of DNA Evidence 2000 hearing; Steinhardt 2004).

The National Research Council I did have some foresight into these issues and raised several questions related to function creep, DNA sweeps, suspect-elimination databases and disclosure of data not related to law enforcement. They didn't however address them in their harm benefit analysis. Instead the Council was content to note that novel practices and the uncertain projected consequences thereof lead to problems when trying to balance good and bad consequences (President's Commission, 1982). Thus the consequentialist analysis was limited to queries of balancing positive identifications versus erroneous conclusions, the weight to be accorded to the interests of various parties, and the superiority of DNA identification technology. Once more no genuine analysis was put forth on these issues saving the description and comparison of various identification technologies with DNA methods. Issues of stigmatization and discrimination were subsumed under privacy and largely ignored.

1.4 Policy and Legislative Background

1.4.1 Policy Recommendations for Protections in the Forensic Context:

As of 1986 DNA typing was in use in commercial laboratories in the US. (It was unclear initially what external regulation or supervision would be required of these facilities. Only after the successful challenges to DNA testimony in courts did the issues begin to cryatalize/materialize.) There have been several advisory bodies instigated to supervise the safe and rigorous implementation of DNA into forensic applications. The many groups include TWGDAM/SWGDAM, NRC I and II, the proposed NCFDT, the DAB, and the NCFDE. The first group established to facilitate the introduction of DNA methods was the Technical Working Group on DNA Analysis Methods (TWGDAM).

The Technical Working Group on DNA Analysis Methods (TWGDAM) was formed in 1988 at the behest of the Federal Bureau of Investigation (FBI). As a forum for forensic

laboratories TWGDAM encouraged discussion on issues, collaboration on studies, and consensus building as to the DNA methodologies to be used in North American crime laboratories. Coincidentally, 1988 was the same year that the FBI began using DNA methodology in casework. By the summer of 1989, questions with respect to scientific, legal, societal, and ethical issues had begun to arise as a result of the novel application of this technology.

The questions not only focused on laboratory analyses and dependability but also public perception of the reliability and validity of DNA as evidence. To ensure the technique's credibility, a group of federal agencies called upon the National Academy of Sciences to examine the unresolved issues surrounding DNA testing and recommend guidelines for its beneficial and competent use. The National Research Council (NRC) an arm of the National Academy of Sciences assumed the task of addressing the general applicability and appropriateness of DNA technology to forensic science. The NRC formed the Committee on DNA Technology in Forensic Science which held its first meeting in January 1990, chaired by Victor McKusick, and issued its first report on forensic DNA with recommendations in 1992 (Rudin and Inman 2002; NRC I 1992). The NRC Committee's report entitled, DNA Technology in Forensic Science, tendered recommendations in six separate areas: technical considerations, statistical interpretation, laboratory standards, databanks and privacy, legal considerations, and societal and ethical issues (NRC I 1992). The NRC report also suggested the formation of a National Committee of Forensic DNA Typing (NCFDT) under the auspices of an appropriate government agency to provide advice on scientific and technical issues as they arise (Rudin & Inman 2004; NRC I 1992; NRC II 1996). The two agencies proposed to provide this service

were the National Institutes of Health or the National Institute of Standards of Technology. Neither agency was given the mandate nor funded for such an undertaking.

The committee's recommendation for the statistical basis for interpretation of a DNA profile sparked heated debate and generated the paramount impetus for the second committee's constitution and direction. The "ceiling principle" and "interim ceiling principle" were conceived to ensure that the rarity of any individual profile in the population was not underestimated. The calculations, unfortunately, had no scientific basis nor did they constitute a ceiling. The rationale was to provide protection from infinitesimal probabilities by providing conservative allele frequencies in the calculation, until population sampling could provide a sound genetic and statistical basis for the use of actual allele frequencies. The committee also weighed in on laboratory error rates recommending that they "be measured with appropriate proficiency tests and should play a role in the interpretation of results of forensic DNA typing" (NRC I 1992). However, failure to define an "error" and any specification for an acceptable or unacceptable risk of error led to confusion.

The controversies surrounding the first NRC report prompted Judge William Sessions, then Director of the FBI, to request in April 1993, a second examination by the NRC (Rudin and Inman 2002). The new NRC Committee established under a different chair (Dr. James Crow), retained only two members from the previous committee and was tasked to resolve, specifically, the statistical controversies provoked by the NRC I and to incorporate new data on population substructure that had been accumulating (Jasonoff 2004; Inman and Rudin 2002; NRC II 1996). The first meeting of the NRC II was held in September 1994 and the final report, entitled *The Evaluation of Forensic DNA Evidence*, was published in November 1996. As stipulated above the scope of the second NRC Committee was much narrower than the previous one and

addressed primarily population statistic issues in addition to risk of laboratory errors and laboratory operations. They concluded that the calculations for the "ceiling principle" and the "interim ceiling principle" were both unnecessary and inadvisable. Instead the Committee proposed standard statistical procedures based on population genetics, and offered advice on a few of the unique circumstances that arise in forensic applications. Use of the product rule (multiplication of allele frequencies from genetically independent loci) was fully endorsed by the NRC II with a few caveats. However, the Committee strongly discouraged combining the error rate with the estimated profile frequency to generate an estimated risk of error. Instead both estimates should be presented singly. The publication of the NRC's second report put to rest the statistical controversies of DNA test interpretation and sample processing and it concluded, overall, that "The technology for DNA profiling and the methods for estimating frequencies and related statistics have progressed to the point where the reliability and validity of properly collected and analyzed DNA data should not be in doubt" (NRC II 1996).

During the period that the NRC II was formulating its recommendations, the DNA Identification Act of 1994 (42 USC §14131, 1995), was passed by Congress, providing for a DNA Advisory Board (DAB) to be appointed by the Federal Bureau of Investigation from nominations submitted by the National Academy of Sciences and other organizations (NRC II 1996; Rudin and Inman, 2002). The board, which replaced the proposed NCFDT, was established to set standards for DNA testing and provide advice on other DNA-forensic matters. Originally the duties of the DAB were scheduled to revert to TWGDAM in 1999 however the mandate was extended through 2000. The DAB provided guidelines for both forensic DNA testing and DNA database typing on a wide variety of quality control issues. The work performed by the DAB resulted in the publication of two sets of quality assurance standards.

Quality Assurance Standards for Forensic DNA Testing Laboratories was issued in October, 1998 by the director of the FBI and a separate but overlapping set of standards, *Quality Assurance Standards for Convicted Offender DNA Databasing Laboratories*, was released in April 1999, for groups maintaining DNA databasing (Quality Assurance Standards for Forensic DNA Testing Laboratories 2000; Quality Assurance Standards for Convicted Offender DNA Databasing Laboratories 2000).

The standards were developed to help ensure the highest quality laboratory performance and to unify the field as a whole. The influence of DAB has been substantial, as compliance with the standards set by the advisory board is necessary for any agency to receive federal laboratorydevelopment funds for forensic DNA testing or DNA databasing (NRC II 1996). One of the ways compliance with DAB standards is assessed is via objective auditing. Forensic scientists, either internal or external to the laboratory perform the laboratory audits and are typically inspectors from the American Association of Crime Laboratory Directors-Laboratory Accreditation Board (ASCLD-LAB), or more recently from the National Forensic Science Technology Center (NFSTC). The NFSTC has agreed to perform certification audits on DNA sections of laboratories for compliance with DAB and ASCLD-LAB standards as the ASCLD-LAB accreditation program does not allow laboratories to obtain accreditation for specialized services exclusively. To obtain ASCLD-LAB accreditation a laboratory must comply with standards for all forensic sciences services offered. The NFSTC offers it's auditing service to private sector DNA laboratories as well as government laboratories. The quality assurance guidelines promulgated by TWGDAM, the DAB and ASCLD-LAB require laboratories to document personnel qualifications and training, laboratory organization and management, facilities, evidence control and procedures, validation of methods and procedures, equipment

calibration and maintenance, analytical procedures, standards for case documentation and report writing, procedures for reviewing case files and testimony, proficiency testing, corrective actions, audits, safety program and review of subcontractors (Faigman et al. 2002). In addition, both the DAB and TWGDAM recommend that every analyst regularly undergo open proficiency testing, twice a year, and that whenever proficiency testing discrepancies or casework errors are detected, corrective measures, by the laboratory, occur in a timely manner. The American Board of Criminalistics also offers certification, as a specialist in forensic biology DNA analysis, requiring a single proficiency trial per year (Faigman et al. 2002). The standards promulgated by the DAB, just as the previous TWGDAM guidelines, have become *de facto* for any facility furnishing DNA services to courts regardless of formal accreditation or the receipt of federal funds. To further bolster the quality of forensic DNA services, the DAB encouraged laboratories to seek accreditation and at least two states require such accreditation of forensic DNA facilities (DAB Standards, FBI July 15,1998 p1; N.Y. Executive Law § 995-b (McKinney 1999); Cal DNA and Forensic Identification Data Base and Data Bank Act of 1998, Cal Penal Code § 297 (West's 1999)).

Unfortunately, the differences in interpretation of the assorted standards by the audit teams (in all disciplines, not just DNA) appeared to have exceeded the differences in implementation of those same standards between laboratories. As a consequence, laboratories were expending more energy in second-guessing the expectations of their audit team than in the creation of a quality program. In an effort to minimize interpretation variability and clarify the intent of the various standards the FBI, in collaboration with ASCLD/LAB, NFSTC, and multiple forensic DNA laboratories undertook the development a comprehensive audit

document. *The Quality Assurance Audit for Forensic DNA and Convicted Offender DNA Databasing Laboratories* document was published in December 2000.

Meanwhile, at the behest of Attorney General Janet Reno, the National Institute of Justice appointed a National Commission of the Future of DNA Evidence in 1998. The task of the National Commission was to suggest ways to improve the investigative utility of DNA and aid "in the operation of the criminal justice system, from the crime scene to the courtroom" (National Commission on the Future of DNA Evidence 2000). The constitution of the Commission included not only forensic scientists but also representatives from law enforcement, municipal politics, legal aid, victim advocacy, and academic law. The multidisciplinary makeup of the Commission, chaired by Shirley Abrahamson, chief justice of the Wisconsin Supreme Court, and its early organizational choices exhibited a manifold understanding of its advisory mandate. Five working groups, formed from the twenty members, selected topics they viewed to be most urgent for further review. These included research and development, postconviction issues, crime scene investigation (improvement and standardization), legal issues, and laboratory funding for database backlogs. The formal work of the National Commission ended in November 2000 when it presented its final report to Attorney General Janet Reno at a special conference at Harvard University.

In 1999, TWGDAM changed its name to the Scientific Working Group on DNA Analysis Methods (SWGDAM). The tradition cast by TWGDAM and the DAB was assumed by SWGDAM in 2000. SWGDAM continues to update guidelines for quality assurance, proficiency testing, statistical treatment of forensic DNA data, and interpretation and has issued several consensus statements regarding the validity and reliability of various DNA techniques, calculations and responses to DNA testing challenges. Through the means of various

subcommittees issues of quality assurance, quality control, database developments, and new technologies are critically examined before implementation. Like TWGDAM before it, SWGDAM provides laboratories throughout the country with a forum for the exchange DNA testing data, sponsorship for inter-laboratory studies and has played an important role in establishing guidelines for working forensic DNA labs.

1.4.2 Policy Recommendations for Protections in Genetics Research

The Canadian Tri-Council Policy statement provides principled guidance in the area of human genetics and though it does not address the forensic use of molecular genetics directly it provides a framework for policy development. This summary of the issues is modest mostly to provide a sense of the scope of the issues related to human molecular genetics in research. Though there has been a decade of experience and analysis since its inception nuances further challenge genetic research.

Section 8 of the Canadian Tri-Council Policy statement on human genetic research involves the study of genetic factors responsible for human traits and the interaction of those factors with each other and, in some instances, with the environment. Research in this area includes identification of the genes that make up the human genome, the functions of the genes, and the characterization of normal and disease conditions in individuals, biological relatives, families and groups. Observation of different forms of the gene may be important among biological relatives and within and among different groups. Accordingly, human genetic research is concerned with the use of genetic material. Genes and their alleles are being identified as part of the Human Genome Project, but the function of each gene and its relationship to human health may not be clear. Although the research is both exciting and

rapidly changing, the recently acquired knowledge regarding genes and their mutations is not yet matched with a full understanding of the implications for human subjects.

Because genetic material is by its very nature shared by biological relatives, identifying a genetic causative agent has implications beyond the individual. Thus, issues of privacy and confidentiality may affect the individual, the family and the group to which the individual belongs. For example, in population studies, a particular group can be identified by common descent, geographic location, ethnic origin, etc. The results, if revealed and publicized, may stigmatize the other individuals in that group.

The potential ability to identify all human genes and their mutations has profound social implications. Misunderstanding or misuse of the results of genetic testing has the potential to interfere with an individual's self identity and sense of self-worth, and to stigmatize the entire group to which that individual belongs. A number of issues remain unresolved and require continuing deliberation by the research community and the public.

1.4.3 Legislation

The nature of the law is that it is a mutable entity, constantly adapting to the needs of the community it serves. As such legislation is constantly changing. The legislation in the area of DNA profiling and banking is no different. Understanding that the state of the scientific art is also in flux, legislators were cautious and though there have been some modifications in the overarching legislation authorizing the collection of DNA and establishing DNA banking facilities; the collection and storage of DNA samples themselves has been largely left to the provinces and states. As such there is no legislation in Canada or the US that directly addresses in one comprehensive piece the issues under analysis. The following are the federal laws that address the establishment of forensic DNA banks and privacy legislation where available.

1.4.3.1 Canadian

In Canada, Parliament enacted Bill C-3 for the establishment of a National DNA database in December 1998. In November 1999 amendments were added to Bill C-3 under Bill S-10 to include the taking of fingerprints for identification purposes, the inclusion of designated offenders convicted in military courts and a full legislative review after five years, to be conducted by the Senate. On June 30, 2000 the bills were officially proclaimed. In July 2000 the National DNA Data Bank opened its doors for business. "The Data Bank will include DNA profiles from young offenders as well as adult civilian and military offenders who are convicted of serious crimes. It will also include a crime scene index, which will contain DNA profiles from unsolved crime scenes. The information can be cross-referenced to find a match in the system" (RCMP press release, 2000). The national DNA data bank is part of the Royal Canadian Mounted Police's (RCMP) National Police Services which includes the Criminal Intelligence Service of Canada, the Canadian Police Information Center (CPIC), Forensic Laboratory Services, The Canadian Police College and Identification Services. The DNA Identification Act allows for the retention of samples and DNA profiles of convicted persons indefinitely. If however the individual's conviction is reversed their sample is destroyed, subsequent to satisfying discharge conditions. Youthful offenders also fall under the Act but retention of samples and profiles fall under the Young Offenders Act. "Information collected by the National DNA Data Bank will be used strictly for law enforcement purposes. All other uses including medical research, are strictly prohibited and punishable by law" (RCMP 2000).

<u>The Canadian Privacy ACT.</u> The purpose of the Act was to extend the laws of Canada to protect the privacy of individuals with respect to personal information about themselves held by government institutions and to provide individuals with a right of access to that information. Of

course the Act limits individuals from access to information contained in databases designed for criminal detection.

1.4.3.2 United States

In the United States the FBI CODIS databank commenced operations in October 1998. All states, along with the Federal Government, have implemented laws to collect DNA profiles from convicted offenders and place them in a database. The DNA Identification Act of 1993 (H.R.829,S.497,103d Cong.,1st sess.) allowed the expansion of state data banking capabilities through federal funding. (McEwen and Reilly 1995).

The CODIS database has three hierarchical tiers - local, state and national and is installed in 137 forensic laboratories in 47 states and the District of Columbia. To date, CODIS has assisted in over 1,900 investigations in 31 states. The national tier of CODIS holds over 600,000 convicted offender DNA profiles, along with 26,000 crime scene samples. All of the States in the United States have state DNA laboratories but not all of them have the funding to support those labs (Hibbert 1999; Stevens 2001).

Currently there is no federal privacy legislation protecting individuals from the intrusions of government on their rights to privacy (Editorial B 1997; Stevens 2001). Individual states have enacted legislation but most of them limit the use of DNA information by group health insurers not life insurance agencies. In 1996, the Health Insurance Portability and Accountability Act came into effect, which prohibits the use of only genetic test results to apply "preexisting condition" exclusions by group health insurers. This is designed to limit exclusions of individuals in group insurance to genetic conditions that are indicated by genetic tests and not by any actual symptoms (Hall and Rich 2000).

1.5 Summary

DNA profiling has for the most part met the legal standards for scientific evidence. There is still debate regarding the statistics and weight of match probabilities but the science itself is sound. The courts, however, have a responsibility to ensure that the specific evidence at bar meets evidentiary standards. Errors have plagued DNA testing facilities that are not related to the scientific validity of the technique but the actual execution. Testing facilities vary in their application of quality assurance measures, rate of mix-ups and cross contamination, integrity of employees and proficiency testing. All of which can have very serious legal ramifications. Error rates and proficiency test results must be available to the court and jurors to allow proper assessment the weight DNA evidence should be accorded. The solid scientific foundation for DNA testing is no guarantee that DNA testing will be reliable and produce accurate results. It is the responsibility of attorneys, judges and legislators to press for reforms through transparency, the creation of independent commissions to supervise forensic laboratory operations and external proficiency testing.

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CHAPTER 2: DNA BANKING

2.1 Preface

In order to ferret out the potential ethical and legal issues involved in the mass use of DNA in forensic and molecular genetics applications, it was important to assess the changes that had occurred in the field since the 1995 survey of McEwan and Riley. This follow-up project, A Survey of DNA Banking Worldwide, helped to focus an ethics inquiry on consent, privacy and future use concerns within the forensic banking community. This paper was previously published in Populations and Genetics: Legal and Socio-Ethical Perspectives. Martinus Nijhoff Publishers, Linden, 2003 and as such the data is not current. While there have been changes, both technological and social, since that time, the ethical issues raised are still pertinent (Bianchi and Lio 2007; Maschke 2005). In addition to the expansion of DNA or biobanks, the numbers of samples included in these banks has increased exponentially (Nuffield Council on Bioethics 2007). The FBI boasts that the National DNA Index (NDIS) contains over 6,031,000 offender profiles and 225,400 forensic profiles as of June 2008, each of these profiles has a corresponding DNA sample stored in a DNA bank (http://www.fbi.gov/hq/lab/codis/clickmap.htm). The United Kingdom biobank has on it's home page the number of people who have submitted samples (168,353 as of August 2008; http://www.ukbiobank.ac.uk/) and the UK forensic DNA bank now hosts 6% (four million samples) of the UK population (Nuffield Council on Bioethics 2007; House of Commons 2007). The legal and regulatory information has changed rapidly in response to the expansion of DNA technology in forensics and is addressed in more detail in chapter 4.

2.1.1 Introduction

DNA banking is not a new topic but it has recently become evident that there is more global DNA banking occurring than anyone might have suspected. The advances in science and technology that have allowed the sequencing of the Human genome to become a reality have also inspired commercialisation. Commercialisation includes, but is not limited to, intellectual property rights, the development of diagnostic tests, public offerings of genotypic information and the development of genetically based pharmaceuticals.

It seems that there are weekly reports of new findings, new genes and new tests that have captured the public's attention and focused it on DNA and the facilities that store it. A quote from Dr. Philip Reilly at the 1999 National Commission on the Future of DNA Evidence speaks to that very point. "We have actually arrived at universal DNA databanking. It's just no one's talking about it" (Hembree 2002). The inspiration for this work was a paper written by McEwen and Reilly in 1995, cited above, which surveyed research laboratories that banked DNA (McEwen and Riley 1995). None of the facilities considered themselves to be DNA banks. The study presented below was an initial effort to assess DNA banking in the public domain, as well as changes in the field from 1995 to 2003.

Not only is DNA banking going on globally but in many cases there are no unified standards governing the practice. There are measures under way to unify law enforcement standards but the commercial and public health initiatives have little motivation to standardize their procedures (Carracedo 2002). The International Society of Forensic Genetics, the European Network of Forensic Science Institutes, and the European DNA Profiling Group are pushing for a unified number of STR's to be run and a compatible platform to be used to share information (Carracedo 2002). "Guthrie" cards, which are available around the world, a rich source of

unbiased genetic material collected and stored uniformly, are currently being used in genetic research. There was a need to assess this field so as to determine the risks that may be associated with the utilization of this rapidly changing technology. DNA can be a powerful tool but its benefits to society must be weighed against the risks to individuals and communities.

The aim of this study was to survey a variety of DNA banking facilities throughout the world to determine their banking procedures, duration of specimen storage, what security and privacy measures were in place and what quality assurance practices were available. The goal of the project was to make an inventory of these resources; to compare and contrast individual data banks within and between categories of banking; and to evaluate the manner in which privacy and security concerns are addressed in each bank.

The survey defined DNA banking as the collection and storage of DNA or of profiles derived from DNA marker analysis. DNA banking was defined in such a broad format in order to draw attention to the idea that marker profiles are being used for more than just identification in the forensic sense. They are being used to estimate drug sensitivity and minimize adverse reactions, to trace the transmission of disease alleles through generations and help to target alleles that may be associated with disease. With the knowledge that there are only 30,000 genes in the human genome, it is conceivable that the DNA that is today considered junk may well be implicated in the regulation and function of genes in the future. Thus, the anonymous sequences that are being used today solely for identification may well become informative of the individuals' health.

The banking facilities surveyed comprised commercial ventures, public health initiatives and forensic facilities. Commercial facilities consisted mostly of fee-for-service establishments but also include research facilities looking for intellectual property rights. Public Health

facilities included public health initiatives such as newborn screening, commercially operated public health bodies and government funded research. Forensic facilities comprised provincial, state or federally funded facilities designed solely to identify individuals through DNA.

Within these domains, forensic units perform only one function, while commercial ventures perform forensic services, public health research as well as offering services to the general public. Many of the commercial facilities functioned in more than one capacity.

2.2 Materials and Methods

This survey was conducted in 2003 by means of a structured questionnaire that was filled out through the use of on-line sources, brochures and telephone interviews of 36 facilities conducting DNA testing, profiling and banking. The survey sampled laboratories and banking facilities from 14 different countries, broken down into three categories or groups; exclusively forensic (25%), commercial (58%) and public health initiatives (17%).

The survey instrument was developed after conducting a review of relevant literature. Pre-interview validation of the survey instrument included review of multiple drafts of the questionnaire by a geneticist and by a bioethicist. The survey consisted of 23 questions most of which were close ended, i.e., with a finite number of standard answer choices. Respondents were encouraged to elaborate and room was left to fill in additional information, so as to enhance validity by ensuring that respondents were not forced into answer choices that did not accurately reflect their policies. The questionnaire, which took approximately 15 minutes to complete, was designed to elicit information about storage practices, sample usage and access including security measures, quality assurances and personnel qualifications. Again, the survey defined DNA banking as the collection and storage of DNA or of profiles derived from DNA marker analysis. The survey was conducted in the first half of 2002.

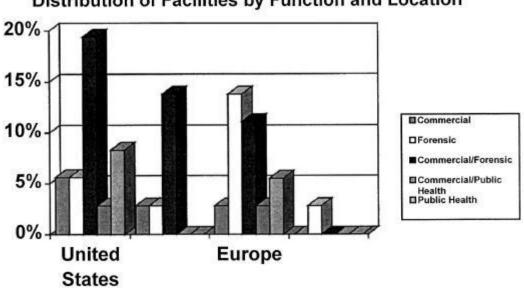
2.3 Results

Of the facilities surveyed 15 (42%) were from the US, 13 (36%) were in Europe, 7 (19%) were in Canada, and 1 (3%) in Australia. The distribution of facilities by location and function is graphically presented in Figure 1. The European component consisted of facilities in Austria, Belgium, the Czech Republic and Slovakia, Iceland, Italy, Malta, the Netherlands, Portugal, Spain, and the United Kingdom. Commercial facilities constituted 58% of the sample while public health initiatives and exclusively forensic facilities comprised 17% and 25% respectively.

The source of DNA sample can vary from tissue to a single cell. Each facility banks specimens according to which type will optimize results, as each type of facility has a somewhat different agenda. The survey revealed that banking facilities stored a variety of different specimens with 18 facilities (51 %) storing more than one type. Six (17%) of the banking facilities stored tissue, mostly in the form of buccal swabs, 19 (53%) stored isolated DNA, 8 (22%) stored blood spots or "guthrie" cards, 18 (50%) stored marker profiles and 2 (5%) stored lymphocytes (Figure 2). Of the 20 facilities that perform marker profiles, 90% store them and 90% use 13 or more markers.

2.3.1 Storage of Samples

The duration of storage of specimens varied greatly among commercial facilities from 3 months to an indefinite period. This was mostly reflective of the stated use of services; provided by the facility. Many of the facilities offered different periods of storage for different types of samples or for different priorities. For commercial facilities this translated into 2 facilities limiting storage to between 0-6 months while 2 facilities limited storage to between 11-50 years.



Distribution of Facilities by Function and Location

Figure 1. The distribution of facilities by geographical location and function

The other 16 (71%) facilities would potentially retain specimens indefinitely though their standard storage times may be shorter. In contrast, most of the forensic and public health facilities surveyed stored specimens indefinitely. Only 1 public health facility specifically limited storage to 10 years. The US Armed Forces Repository of Specimen Samples for Identification of Remains is the only forensic facility that will remove a sample after 30 years, once the individual has been discharged, but only if a written request to do so is submitted. Otherwise the samples will remain on file for a minimum of 50 years and no date is specified for destruction. In total 30 (83%) of the labs surveyed store DNA or marker profiles indefinitely.

The conditions under which specimens are stored vary considerably mainly based on the type of sample being banked. Of the 8 facilities storing FTA or "Guthrie" cards, 6 keep samples at room temperature. All of the 18 facilities storing isolated DNA samples store them in the freezer with the median temperature of -20C. Only two (6%) facilities indicated that they used liquid nitrogen as a storage medium; these were also the facilities that banked lymphocytes exclusively. Five (14%) facilities indicated that they did not store physical samples but retained marker profile information.

2.3.2 Use of Samples

The intended use of the specimen can range from genetic research, drug and product development, public health initiatives, and civil issues regarding paternity, estates and immigration, as well as forensic issues of criminal investigation, perpetrator detection, and remains identification.

2.3.3 Facilities

2.3.3.1 Commercial Facilities

Of all of the facilities surveyed, commercial entities performed the greatest variety of services, including forensic profiling, paternity and relationship testing, genetic disease screening, drug interaction profiling, as well as research on specific populations and specific disorders. More than half (57%) of the 21 commercial facilities surveyed perform research. Of the commercial facilities performing research, 5 indicate that product development is one of their aims. Three of the commercial research facilities are dedicated to the study of a specific population and could alternately be considered to be population banks. Forensic profiling is conducted in 16 of the commercial facilities while personal and civil matters are attended to by 16 of the surveyed facilities. Eighteen (52%) of the total queried facilities performed or offered more than one service.

2.3.3.2 Forensic Facilities

The 9 exclusively forensic facilities surveyed included 8 (89%) law enforcement facilities and 1 (11%) repository for the identification of human remains. In addition, 16 commercial facilities provide forensic testing on a contract basis. Though there is discussion by all law enforcement facilities to store DNA in a limited capacity, mainly so that as technology changes they are able to update their banks without having to perform repeated sample collections, 4 (44%) facilities clearly state that they bank a physical sample. The other 5 (56%) bank only the marker profile.

2.3.3.3 Public Health

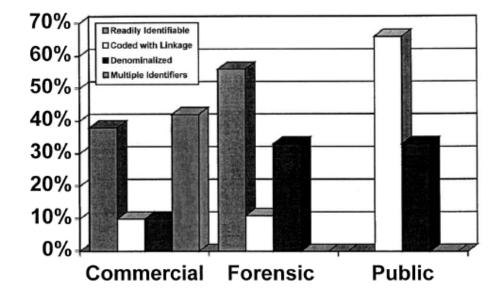
Five (83%) of the 6 public health initiatives are state interest disease groups. All of these initiatives provide DNA for research purposes. Three (50%) of the facilities provide DNA via access to "guthrie" cards, 2 (33%) store lymphocytes and 1 (17%) provides extracted DNA.

2.3.4 Access to Samples and Data

All of the facilities surveyed limited access to both the specimen and the data associated with it. Select employees were granted access to the sample in 35 (97%) of the surveyed facilities. Data access in 30 (83%) facilities was limited to select employees. All access was regarded as work related.

Only 1 (8%) of the 12 commercial research facilities granted sample access to external researchers, while 3 (25%) of the 12 granted external researchers access to data. Access to the samples in exclusively forensic facilities was restricted to lab personnel and access to the marker profile was limited in all cases to authorized law enforcement officials. The public health initiative laboratories limited sample access to select employees and external researchers. Three (50%) of the public health facilities performed genetic testing in addition to storage, and thus data was accessible to select employees, while the other 3 (50%) facilities accede to data requests from external researchers only after publication of results.

Of all of the surveyed facilities, only 4 (11%) completely anonymize their research samples; all four are commercial facilities. Ten (28%) facilities have specimens that are denominalized, but linkable to data. One of these also banks samples that are coded with full linkage. Still 8 (22%) other facilities code specimens with full linkage between marker information and personal data. Of these, 3 also store specimens that are readily identifiable.



Use of Identifiers

Figure 2. Extent of denominalization of samples as a function of type of banking enterprise

Thus a total of 21 (58%) facilities have their samples or profiles readily identifiable by name and pertinent information (Figure 2).

Sixteen (94%) of the 17 commercial facilities that provide services will grant access of the remaindered sample to the donor of samples or to the donor's representative with a written request, typically provided at the time of consent. Only 1 (12%) of the exclusively forensic facilities allows access to the sample, and then only with a court order. To access samples from any of the public health initiatives requires a letter of intent, and all requests must go through at least one Internal Review Board (IRB) before access is granted.

Access to the data revealed as a consequence of the provision of commercial services is outlined in the consent forms of the 17 facilities, and all of the commercial facilities surveyed issue a report to the individual who submitted the sample or to a designated representative (e.g., the physician). Access to the data in all the forensic law enforcement facilities is restricted to authorized officials only. The 3 (9%) commercial facilities that perform marker profiles for law enforcement submit the results directly to the database. The Armed Forces Repository of Specimen Samples for Identification of Remains submits its data to the US Armed Force branch concerned, and they in turn notify the appropriate living relatives. The data derived from the public health research initiatives becomes public domain after the publication of the results by researchers, while the test results of newborn screening are reported to the physician, hospital or patients for confirmatory testing (Mandl 2002).

2.3.5 Security

Both physical and electronic security is of concern in a post September 11th environment. Physical access was restricted through securing the perimeter of facilities in 35 (97%) cases; badges were utilized in 4 (11%) of those facilities. Electronically the majority (26, or 72%) of

the surveyed facilities protected data by both encryption and passwords. A computer systems firewall was specifically mentioned in 10 facilities with either encryption or password protection. Secure socket links were also available in 4 of the commercial facilities. One commercial facility stores nothing electronically and one exclusively forensic facility stores biologic material and identifiers in separate locations.

2.3.6 Legislation

Specific legislation covers 22 (61%) of the 36 surveyed facilities. All public health initiatives are covered under mission mandates or state legislation, but 9 (25%) facilities are unregulated either by legislation or mandate. The regulatory bodies vary drastically worldwide. Most of the survey facilities indicated a specific regulatory body to which they subscribe, and which provides some form of accreditation,

2.3.7 Personnel Qualifications

All of the facilities surveyed had minimum standards for personnel, including supervisors, technicians and result analysts. With respect to database managers, 33 of the 36 facilities specified minimum requirements. One facility keeps no electronic records, thus eliminating this issue. Information was not known or could not be provided by two respondents, both of which are part of newborn screening efforts.

2.3.8 Oversight

Twenty-six (72%) of the surveyed respondents indicated that there was some form of oversight, either through legislative bodies, regulatory agencies or compliance with accreditation facilities. Of these, 17 undergo annual performance visits, 6 undergo biennial checkups, and 4 did not specify the evaluation periods. Only three facilities indicated that they complied with more than one type of performance visit (Figure 3).

Oversight

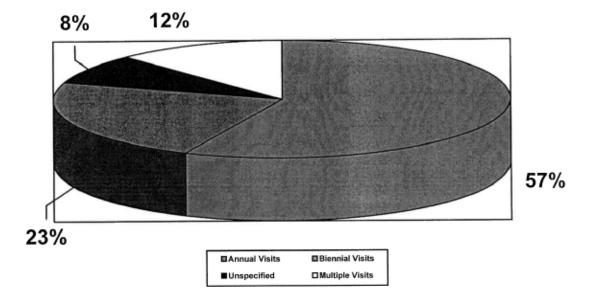


Figure 3. Variation between laboratories in use of quality control measures

2.3.9 Quality Assurance

Proficiency testing and quality assurance testing was reported in 32 (88%) of the surveyed facilities. Twenty-seven (75%) indicated how often this testing occurred, while 3 of the remaining 10 indicate internal auditing, but fail to give a time frame.

Thus, 7 facilities perform daily quality control, 1 performs semi-annual exercises, 9 perform annual quality checks, 3 perform biannual quality assessments and 2 perform quarterly proficiency measures. Test validation was performed in 30 (83%) of the 36 sampled facilities (Figure 4).

2.3.10 Consent

Written consent was required in 29 (81%) of the 36 surveyed facilities. The exclusively forensic facilities, of which there are 9 (25% of the total), have legislation granting them powers to compel a sample when an individual declines to give consent. All of the commercial facilities were clear about the usage of the samples and the need to obtain new consent for further examinations. Only the public health facilities were vague regarding consent.

A closer examination reveals that the public health section of the survey separated into three subgroups: government funded research, public health initiatives, and commercially operated public health bodies. The consent requirements varied with the subgroups. The two government subsidized research facilities provided a broad discussion of the research aims, but intentionally left the consent document vague so as to allow "mining" of the samples to their fullest potential. It was unclear if researchers associated with these public health ventures were planning to update consent for each individual research project. The single public health resource, which was commercially operated, indicated that consents are updated as research agendas change. The last three facilities were established as public health initiatives for newborn

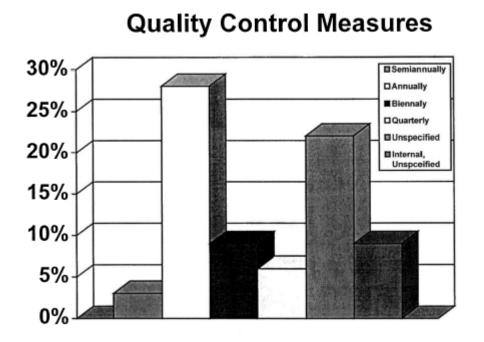


Figure 4. Variation between laboratories in the use of quality control measures

screening, and though no consent was required at the time of testing, no further attempt was made to contact the individuals when the samples were donated for research. It is considered ethically acceptable in research to use these samples without consent provided that all individual identifiers are removed. As population screening for genetic disorders becomes more prevalent, population consent may become an issue (European Society of Human Genetics 2000).

2.4 Discussion

Thirty-six facilities conducting DNA testing, profiling and banking were surveyed by means of a structured questionnaire. Facilities from 14 different countries, broken down into three categories or groups [exclusively forensic (25%), commercial (58%) and public health initiatives (17%)] comprised the sample population. Although there are a few areas in which all or most facilities follow similar procedures, the survey more frequently revealed an immense degree of variation between facilities with respect to virtually all practices and procedures. For example, while the most common source of DNA for marker testing is blood, many facilities store several different types of blood derivatives, while a minority store tissue or tissue derivatives as well. The duration of specimen storage varied greatly among commercial facilities from 3 months to an indefinite period, while the majority of public health and forensic facilities typically store specimens indefinitely. In general, variations in storage conditions principally reflected the type of sample being banked. Commercial facilities had the greatest variability with regard to the use to which DNA samples are put, ranging from forensic profiling, to paternity and relationship testing, to genetic disease screening or drug interaction profiling, as well as research on specific populations and specific disorders. The forensic facilities use samples to evaluate individuals as suspects or to identify them for burial. The public health

initiatives all use samples for individual clinical queries, but most are also involved in research as well.

With respect to regulatory control, there was generally less variability than might have been anticipated. All of the surveyed facilities limited employee access to both the specimen and the associated data and all access was regarded as work related. Physical access to the facilities was restricted through securing the perimeter in almost all of the cases. Electronically, the majority of the surveyed facilities protected data by both encryption and password protection. All respondents defined minimum standards for

personnel, including supervisors, technicians and result analysts and more than three-quarters specified minimum standards for database managers as well.

Specific legislation and mission mandates covered 75% of the surveyed facilities, while 25% were unregulated. The majority of surveyed facilities indicated some form of oversight, either through legislative bodies, regulatory agencies or compliance with accreditation facilities. Quality assurance and proficiency testing was reported in 89% of the surveyed facilities. Where consent was required, all of the surveyed facilities were in compliance.

A surprising finding was the comprehensive security measures in place to protect both physical specimens and electronic data. In most cases it should be a matter of course in that the commercial facilities are protecting their livelihood and researchers rely on the samples for their work. Law enforcement as well has a vested interest in maintaining the integrity of their systems. The one area where security may still be an issue is in independent or university research initiatives. This subgroup was not represented in this sample and is the most likely to be vulnerable due to lack of resources. Individual investigator archiving of DNA should be the subject of more detailed study. Overall, security appears to be taken quite seriously.

In light of the current job market, it is not surprising that minimum standards are imposed on personnel. All technical personnel in the surveyed population were expected to have a Bachelors degree in science or computer science and a minimum of six months experience. Supervisors, technicians and results analysts were at a minimum required to hold a Masters degree in a related science. In addition, many facilities required confidentiality agreements as part of employment.

Consent is a matter of particular importance and has been stressed repeatedly in the bioethics literature. Most of the facilities surveyed recognize the necessity to inform research subjects and clients of their intentions and duties. The area of true concern is the "mining" of samples from public health initiatives, particularly the newborn screening programs, with the goal of using those samples in ways other than those originally intended. Written informed consent is not currently required from parents for the screening of detectable genetic disorders, for which there are treatments (World Health Organisation, 1997; Therrell 1996). Typically, hospitals which are involved in the collection of such samples do require a consent document, although at best it is sandwiched in at the time of delivery or shortly after birth, or at worst, the expectant parents are completely uninformed, even though their signatures are on the documents. It has become an ethically accepted procedure to anonymize "guthrie" cards when they are to be used in research and renewed consent is not possible (Therrell 1996). Of concern here is not that newborns are being tested without consent but that the sample is then being retained indefinitely and possibly distributed to researchers.

The "Guthrie" cards stored at a facility are reflective of the newborn infants of that region. When considering genetic information it is important to keep in mind the possible predictive outcomes of genetic research results. By using and testing the DNA from previous

births, that population - presumably the current population - may be characterized genetically. Further, if testing is occurring on recently collected specimens, are future adult-onset illnesses being foretold? The genetic characterization of a population may not be of concern to some groups, but others may feel as if their dirty laundry is being aired in public. These populations may perceive themselves, rightly or wrongly, to be the target of stigmatization, or worse yet, discrimination (Andrews 2001). Genetic characterization of a population may be of positive or negative value to that population, but the outcome cannot be known in advance. Bartha Knoppers et al. make a valid point that, since no consent is required for newborn screening, researchers could be jeopardizing such initiatives by using the samples for other purposes (Knoppers et al. 2002). Knoppers and colleagues suggest guidelines for an ethical approach to such research projects, with informed, written consent playing a major role (Knoppers et al. 2002). There are projects currently underway that are following these guidelines and appear not to be hindered by obtaining this type of consent (Atkins et al. 2001). Further, there has been at least one study indicating that screening numbers did not diminish when consent was requested for newborn testing (Levin 2002).

It is expected that each country will have its own regulatory agencies that monitor genetic testing and banking practices. Even if a country as a whole has no specific legislation, the states or provinces have licensing agencies with their own set of regulations. It is thus understandable that there are no uniform oversight bodies. With respect to quality assurances, accreditation facilities help to provide some guidelines for "good practice," but not all accrediting bodies are equal in their standards or stringency. The forensic science community might serve as a role model for an approach to standardizing practices so that quality control and quality assurance programs can be developed. Scientific bodies can agree on standards and norms for DNA testing

and storage. The forensic community has already lead the way through their efforts to ensure the admissibility of DNA in courts by establishing criteria that meet Daubert tests for peer reviewed accurate science. Both technical and procedural standards were addressed at the International DNA Users Conference, which discussed the strides made in standardization to ensure result integrity (Levin 2002). Due to the differences in legal systems worldwide standardization of common forensic analysis has not been easy. The International Society of Forensic Genetics has worked as facilitator through national and international working groups that are actively involved in establishing common standards. Several other groups have contributed to this effort and coordination of these groups is underway (Levin 2002). If the forensic standards withstand court scrutiny why shouldn't those be the minimum standards for all facilities storing DNA? Once norms and standards are adopted, it should be easier to establish internationally consistent oversight bodies.

Enrolment in an accreditation program is a good first start but guidelines ought to be established to facilitate the ability of IRBs to evaluate research proposals and methodology, while still allowing them to reflect the beliefs and norms of the surrounding culture. Each facility must verify kits and reagents within its own laboratory setting and be able to show that the methodology is working properly, even if the manufacturer or the test developer previously verified the method. Tests should be validated along all of the following parameters: linearity with respect to sample, time, and reagent; reproducibility; analytical or reportable range; accuracy; stability of specimens and reagents; interfering substances; reference range; rates of false positives and false negatives; sensitivity; specificity and positive predictive value (Prence 1999; Center for Disease Control 2002;. Association for Molecular Pathology Statement 1999; Deaucker et al. 2001; McGovern et al. 1999; Robinson 1999).

Again, the Forensic Science community could be a role model for establishing minimum standards for quality assessment. There is a necessity for scientific credibility and validity in all testing and banking. The methodology is intrinsically the same and there are some agreed upon standards in the areas of DNA analysis. Regulations might be implemented through the use of international licensing bodies that monitor facilities via accrediting agencies. In this case, accrediting agencies would have to provide similar quality assessment schemes with proficiency testing and only those facilities that maintain acceptable levels of performance are licensed. Accrediting agencies would also ensure that newly developed protocols are compared with older established methods before they can be implemented.

Developing an External Quality Assessment (EQA) scheme designed to educate researchers and IRB members on quality assurance measures could be mutually beneficial; such an EQA scheme has been in place in the UK since 1988, established in recognition of the need for national standards and external quality assessment (Deaucker et al. 2001). Proficiency testing schemes should be a part of all accreditation requirements /standards (McGovern et al. 1999).

Attempts to provide guidelines that allow researchers to exercise creativity and reflect the norms and beliefs of their culture is a complex goal, but could foreseeably be accomplished. It is important that a balance be struck between the desire for progress in research and the need to safeguard individual clients and study populations.

In no way is this survey representative of all facilities that store DNA. Individual researchers were not represented in this survey and this could conceivably have an impact on results pertaining to sample access, security and personnel qualifications. The surveyed population consisted of a variety of types, countries and clients. Although the survey

covered a range of facilities in multiple countries, the sample size was fairly small and limited in scope. A broader selection from the sample population would help the results to be more fully representative of the population as a whole. In the interests of time, the assessment measure was brief as it was administered by an interviewer, and a mass mailing was not an option. A mailed, self administered survey would be ideal, as it would allow for a more comprehensive range of questions, less potential for administrator bias and an increased range of responses. This survey is a small extension of work started earlier by McEwen and Reilly, and reflects the changes that have occurred during that time. Further, it highlights the need for more work to be done to develop an informed public policy.

In summary, the research suggests that the duty to obtain informed consent in most cases was being adequately discharged. Practices with respect to the access of public health samples need to be re-examined with the interests of local populations considered. Further attention to the development of guidelines governing standards for banking practices and the implementation of uniform quality assessment measures is necessary.

Concerns regarding quality assessment have lead to further investigation of the methodology used in forensic settings where the implications though not life threatening in a clinical sense, may curtail ones liberty and in those States where the death penalty is permitted life threatening (Canada is opposed to the death penalty). In the following chapter comparison data will be presented, using state of the art equipment and current methodology, on panels of STR markers in an effort to examine marker performance and reliability.

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CHAPTER 3: COMPARATIVE PERFORMANCE AND IMPLICATIONS OF FORENSIC AND GENOMIC MARKERS IN A FAMILY-BASED SAMPLE

3.1 Introduction

DNA profiling is now being hailed by many as the gold standard in forensic science, replacing the position where fingerprinting once held sway (Lynch 2003). The shift in focus has allowed the problems inherent to fingerprinting to become more commonly acknowledged. Recognizing these limitations, which mostly involve extrinsic error and technician interpretation, allows for the modification of procedures to make the practice more precise. The field of human identification has made remarkable improvements since CellMark's poor proficiency testing and *State of California* v. *John Ivan Kocak*² case involving CellMark (Thompson and Ford 1988; Cotton 1995). In recent years the development of powerful PCR-based DNA typing systems has enhanced forensic DNA identification significantly, capturing both length and sequence genetic variation (Leclair et al. 2004; C. Oz et al. 2003; Peterson et al. 2003). Rapid genetic profiling from minute amounts of biological material is possible through the use of Short Tandem Repeat (STR) loci and highly sensitive instrumentation based on fluorescence detection (Leclair et al.

²There were two different proficiency tests administered by the California Association of Crime Lab Directors in 1988 and 1989. Cellmark made one false match in each of these tests. These false positives are discussed in more detail in a paper by W. Thompson and S. Ford [In: *Forensic DNA Technology* (Proceedings of the American Chemical Society, Science and Law Conference, Los Angeles, CA, 1988), M. Farley and J. Harrington (eds), Lewis Publisher, Chelsea, Michigan].

In the *State of California* v. *John Ivan Kocak*, a sexual assault case Cellmark analyzed, the labels on the known reference samples from the victim and suspect were switched. The vaginal swab apparently had DNA from only the victim. However, due to the error Cellmark declared a match between suspect and the DNA in the sperm fraction of the vaginal swab. Charlotte Word at Cellmark caught the error while testifying in court. Cellmark went on to claim that there was no harm committed as they (i) the ultimately caught the mistake and (ii) the DNA technology worked OK there was only a transcription error. Their response does not take into consideration the possibility of a plea bargain being struck before the Cellmark employee was asked to testify? In the end a report that erroneously says a person matches some evidence DNA does as much damage whether the error was clerical or had some deep scientific genesis.

2004). STR markers target repeated motifs in normal DNA such as CA and GATA; these vary in both the number of repeats and thus in length, and usually have many alleles.

Most forensic facilities have moved away from slab-gel sequencing and genotyping to an automated process of generating STR profiles which yields higher throughput and requires less manual intervention, allowing samples to be processed faster and at a lower cost (Butler 2004; Koumi et al. 2004; Schneider et al. 2004; Swerdlow et al. 1990). The capillary array electrophoresis (CAE) instruments separate DNA by size, through the application of a current across a non cross-linked polymer matrix. Depending on the instrumentation used, this allows the simultaneous resolution of between 1 and 96 DNA samples for up to 10 STRs within separate capillaries. The ABI Prism 3700 instrument can process 96 samples in 2.75 hours and can run four 96-well plates from each setup, allowing overnight runs without supervision. Thus approximately 700 samples per day (corresponding to 7,000 STR genotypes) can be run, including positive and negative controls, per instrument (Shadravan 2004).

These technological improvements have secured STR marker dominance in forensic DNA identification testing (Amorim and Pereira 2004; Jobling, 2001) despite both intrinsic and extrinsic limitations that continue to plague the field. Recent public attention has begun to focus on the limitations of forensic DNA profiling and these limitations are receiving the same scrutiny and public disclosure as fingerprinting (Liptak 2003; Liptak 2004; New York Times 2004; Sessions 2004). Particular limitations of STR markers with respect to forensic applications include stutter (Leclair et al. 2004]; Walsh 1996), allelic drop-out or allelic imbalance (Cotton et al. 2000; Hendrickson et al. 2004; Leibelt et al. 2003; Nelson et al. 2002; Rubocki et al. 2000; Schneider et al. 2003), anomalous peaks or triallelic peaks (Clayton et al. 2004), non-specific peaks (Gill and Kirkham 2004, Kimpton et al. 1996), n-peaks (Koumi et al. 2004) and within-run frameshifts (Kimpton et al. 1996; Koumi et al. 2004).

As part of a comprehensive exploration of the parameters of DNA profiling, a comparison between the performance of markers typically used in scientific genotyping facilities and forensic markers was performed. Since DNA profiling can be a powerful tool in establishing a prosecution's case (although by itself not always sufficient) it is crucial that the markers perform consistently and reliably.

The hypotheses tested in this study are: (1) The older markers would be harder to read comparatively due to their structure as tetra-repeats and the presence of stutter peaks; (2) The newer markers will be more reliable; (3) Does the technique/procedure always yield the same results; i.e., how consistent are the results? (4) There will be limitations on the ability of the marker panels to distinguish closely related individuals.

3.2 Genomic Materials and Methods

3.2.1 *Families*

Cell line DNA for the 93 members of the CEPH pedigrees was obtained from CEPH (www.cephb.fr). CEPH families are multigenerational kindreds originally collected in the early 1980's for the purpose of establishing a genetic map (White et al. 1985; Dib et al. 1996). In this study, we used two three-generation families consisting of 14 and 16 individuals respectively. Family 1 consists of 2 parents, 4 grandparents and 8 children. Family 2 consists of 2 parents, 4 grandparents and 8 children. Family 2 consists of 2 parents, 4 unrelated individuals.

3.2.2 Molecular techniques

All of the genotyping was performed at the McGill University and Genome Québec Innovation Centre. The focus of the center is genomic research and as such is not a forensic facility. The Center's Genome-wide Scan Platform uses an ABI-3700 DNA Analyzer to obtain the genotyping results. Thirty-two fluorescently labeled microsatellite markers were chosen. The markers were segregated into two panels. One representing the 16 markers suggested for use in forensic genotyping and profiling by Applied Biosystems and The Federal Bureau of Investigation. The other 16 markers (genome scan markers) were a random selection via computer algorithm from the 300 most current and commonly run markers at the McGill University and Genome Québec Innovation Center.

The markers were all amplified on GeneAmp PCR System 9700 (ABI) thermocyclers under uniform conditions. Five nanograms of DNA was added to a reaction mixture containing 3.0mM MgCl2, 0.1 mM dNTPs, 0.1 uM of each primer and 1.0U *Taq* polymerase. The reaction was initiated by denaturing the samples at 96 C for 10 min, followed by a touchdown procedure comprised of 40 cycles of 30s denaturation at 94 C; 30 s primer annealing at 60 C (3 times), 59 C (2 times) and 54 C (35 times); and 1 minute extension at 72 C. Final extension was done at 72 C for 10 minutes. All reactions were carried out in 96 well plates with 93 samples, 2 positive controls and 1 negative control. Allele binning and mendelian error checking was done using PEDMANAGER software.

3.2.3 Experimental design

All of the forensic markers were run on the 93 samples of related and non-related individuals a minimum of 4 times. The panels were run sequentially for the first two runs and the last two runs were performed simultaneously with the same reaction mixture and analyzed in

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the same machine. The genome scan markers were run three times and in all other respects under conditions identical to those used for the forensic panel.

3.2.4 Data Analysis

URL: PEDMANAGER software is available at http://www-genome.wi.mit.edu/ftp/distribution/software/pedmanager/.

3.3 *Results*

The overall results are presented in the summary table. Of the 5320 genotypes performed for the forensic markers, 5181 generated an answer. Thus 91% of the time a call was generated. Of the 4560 genotypes performed for the gene scan markers, 4144 generated an answer, roughly 91%. Overall the forensic markers performed better with respect to generating a call. The rate of concordance and discordance for both marker panels were similar, with 98% of the genotypes matching across runs and 2% of the genotypes being discordant across runs. For the forensic markers, 5102 genotypes were concordant across 4 runs and 79 genotypes were discordant across the 4 replicates. The gene scan marker genotypes were concordant for 4070 genotypes and discordant for 75 genotype determinations. There was no significant difference in rates concordant and discordant between panels (P concordant 0.90, P discordant = 0.74).

Table 2 lists the mean performance of each marker. Markers from the forensic panel are shown on the left and those from the genescan panel are on the right. The forensic panel shows a broader range (0.794 to 0.961) as compared to the genescan panel (0.856- 0.965). As is clearly indicated the average performance of the two panels is equivalent. The forensic marker TPOX never yielded an amplified product during PCR despite multiple attempts at optimization. It was later determined that the TPOX primers were degraded and this locus was therefore excluded from the forensic panel.

| SUMMARY | | | | |
|------------|----------|--------|----------|--------|
| | FORENSIC | % | GENESCAN | % |
| | | | | |
| CALL RATE | 5181 | 0.9089 | 4144 | 0.9088 |
| CONCORDANT | 5102 | 0.9848 | 4070 | 0.9821 |
| DISCORDANT | 79 | 0.0153 | 75 | 0.0181 |
| | 5320 | | 4560 | |

TABLE 1: Summary of Genotyping Performance

| MARKER SU | MARKER SUMMARY TABLE | | | | | | |
|-----------|----------------------|------------|------------|----------|--------------|------------|------------|
| | CALL RATE | CONCORDANT | DISCORDANT | | CALL RATE | CONCORDANT | DISCORDANT |
| FORENSIC | | | | GENECSAN | | | |
| D13S317 | 0.9395 | 0.9944 | 0.0056 | D17S1298 | 0.9193 | 0.9961 | 0.0038 |
| D21S11 | 0.8526 | 0.9812 | 0.0185 | D17S1301 | 0.9649 | 0.9782 | 0.0218 |
| D5S818 | 0.9184 | 0.9943 | 0.0057 | D7S3051 | 0.8561 | 0.9877 | 0.0123 |
| THO1 | 0.9395 | 0.9944 | 0.0056 | D16S403 | 0.8702 | 0.9798 | 0.0202 |
| TPOX | | | | D10S1426 | 0.9579 | 1 | 0 |
| D16S539 | 0.9316 | 0.9972 | 0.0028 | D16S518 | 0.9193 | 0.9847 | 0.0153 |
| D3S1358 | 0.9263 | 0.9972 | 0.0028 | D5S1480 | 0.8947 | 0.9843 | 0.0196 |
| D7S820 | 0.8842 | 0.9940 | 0.0059 | D1S1677 | 0.9333 | 1 | 0 |
| D19S433 | 0.8947 | 0.9559 | 0.0441 | D11S4949 | 0.8877 | 0.9684 | 0.0316 |
| AMELXY | 0.9605 | 1 | 0 | D7S1824 | 0.9263 | 0.9962 | 0.0038 |
| D18S51 | 0.9500 | 0.9972 | 0.0028 | D1S1622 | 0.8632 | 0.9797 | 0.0203 |
| D2S1338 | 0.8868 | 0.9881 | 0.0119 | D5S1462 | 0.8877 | 1 | 0 |
| VWA | 0.7947 | 0.8974 | 0.1027 | AMELXY | 0.9509 | 0.8856 | 0.1144 |
| CSF1P0 | 0.9447 | 0.9944 | 0.0056 | D19S586 | 0.9228 | 0.9924 | 0.0076 |
| FGA | 0.8974 | 0.9941 | 0.0059 | DXS7132 | 0.9228 | 0.9962 | 0.0038 |
| D81179 | 0.9132 | 0.9769 | 0.0231 | D14S617 | 0.8632 | 0.9878 | 0.0122 |
| MEAN | 0.9089 | 0.9838 | 0.0162 | MEAN | 0.9088 | 0.9823 | 0.0179 |
| SD | 0.0432 | 0.0264 | 0.0264 | SD | 0.0350 | 0.0274 | 0.0274 |

TABLE 2: Individual Marker Performance

Though there was variation in performance of the individual markers, each panel had a similar mean call rate of 91%, a concordance rate of 98% and a discordant rate of 2%.

The apriori probability of a match on all markers and thus an identical profile is $.25^{16}$ and $.25^{15}$ respectively or $.25^{n}$, where n = the number of markers. Individuals who are first degree relatives are assumed to share 50% (.5) of their DNA, identical by descent, and the probability of sharing 2 alleles at a given locus identical by descent is .25 or 25%.

The family data for each marker panel was assessed for the ability to distinguish between first degree relatives, specifically between siblings, by comparing the marker profiles. Family 1 has 8 siblings and the profile for each sibling was compared in a pair-wise fashion to determine the number of diplotypes per marker. Of the 16 genescan markers between 5 and 12 identical diplotypes were identified in each pair of individuals. Of the 15 forensic panel markers for family 1 between 1 and 7 identical by descent diplotypes were identified for each pair of individuals. The percentage range for the forensic panel was smaller than that of the genescan panel.

With respect to family 2 between 0 and 10 identical diplotypes were identified with the gene scan panel. Of the 15 forensic panel markers for family 2 between 2 and 8 identical by descent diplotypes were identified for each pair of individuals. In no case was a completely identical profile generated between siblings. Siblings were always different at a minimum of 4 diplotypes with the genescan marker panel and 7 diplotypes with the forensic marker panel.

Our experience of examining 15 STR markers suggested for use in forensic genotyping and profiling by Applied Biosystems and The Federal Bureau of Investigation, yielded single run call rates that were suboptimal. In our recent tests using the ABI 3700 CAE technology we determined that the single-run call rate ranged from 79.5% to 96.3% with an average of 91%.

There was a significant difference between the single-run call rate and triplicate or quadruplicate consensus rates (F 2.42 = 24.19, p < 0.0001). The overall results are presented in the Table 3.

One of the goals of this experiment was to test the threshold for acceptable sensitivity as depicted in figure 1 and 2. The ability to identify genotypes with confidence improves when panels are run repeatedly. Individual runs show significant variation in performance when compared with a composite call rate. The figures also clearly demonstrate the variation possible between runs and highlight individual marker performance.

3.4 Discussion

The principal finding of this study is that the forensic marker panel performed as well, if not better, than the more contemporary genescan marker panel. The call rate for the forensic panel was the same as that for the genescan panel, indicating that overall genotypes were likely to be equally assigned. This performance alone is not sufficient to claim superiority, as both the call rate and the discordant rate could be high. Were this to be the case, a wrong answer would be assigned more often than no answer. In the present study, however, the concordant and discordant rates between panels are almost identical. It is also important to note that none of the markers were enhanced for performance with the application of poly-A tails. These synthetic additions help to optimize amplification and increase the amount of PCR product. In forensic settings this addition is common to boost marker performance. Since the McGill University and Genome Québec Innovation Centre is not a forensic facility with technicians trained in forensic applications the conclusions are useful to illustrate issues related to STR genotyping, but should not be used to generalize how state of the art forensic facilities/labs operate nor be used to draw conclusions as to accuracy rates obtained in these facilities.

| FORENSIC MARKER | % AVERAGE CALL RATE per SINGLE RUN | CONSENSUS RATE (Triplicate replicates) | CONSENSUS RATE (Quadruplicate replicates) |
|--------------------|--|--|---|
| D13S317 | 93.95 | 98.95 | 100 |
| D21S11 | 85.26 | 95.79 | 96.84 |
| D5S818 | 91.85 | 95.79 | 100 |
| TH01 | 93.95 | 100 | 100 |
| D16S539 | 93.16 | 98.95 | 100 |
| D3S1358 | 92.11 | 97.89 | 100 |
| D7S820 | 88.42 | 95.79 | 100 |
| D19S433 | 89.47 | 95.79 | 96.84 |
| AMELXY | 96.32 | 100 | 100 |
| D18S51 | 95 | 100 | 100 |
| D2S1338 | 88.95 | 96.82 | 98.95 |
| vWA | 79.47 | 90.53 | 91.58 |
| CSF1PO | 94.47 | 100 | 100 |
| FGA | 89.74 | 94.73 | 95.79 |
| D8S1179 | 91.58 | 98.95 | 98.95 |

TABLE 3: Repilcate Runs Improve Genotyping Reliability. The percent average call rate per run represents the arithmetic mean of the single-run call rates for each marker. The consensus rate for triplicate runs was scored when at least two of three calls were in agreement. The quadruplicate score differs from the triplicate rate in situations where additional data made it possible to arrive at a consensus score.

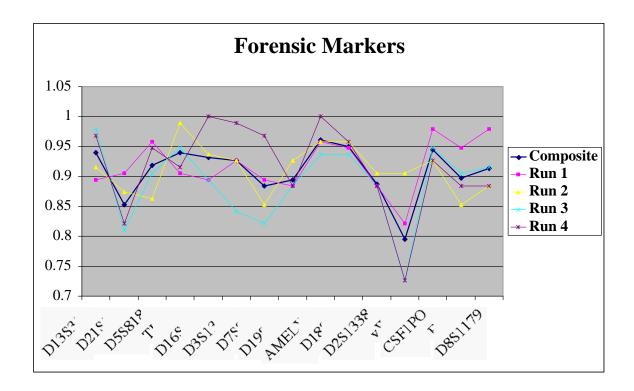


Figure 1: Comparison of Marker Performance from Run to Run. The percent call rate for each replicate run as compared to the average call rate across runs (composite).

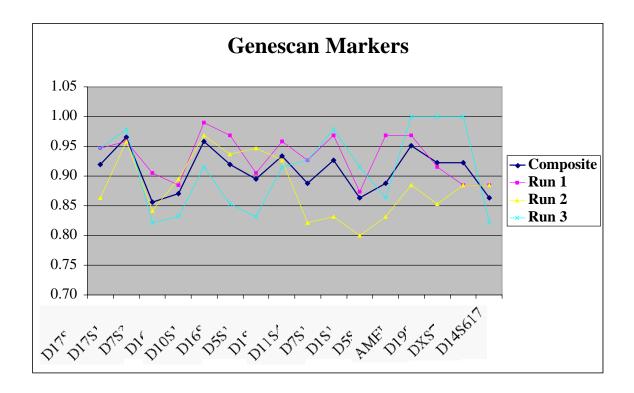


Figure 2: Comparison of Marker Performance from Run to Run. The percent call rate for each replicate run as compared to the average call rate across runs (composite).

It is also important to note that all of the samples and markers were multiplexed and as such the optimal temperature for amplification for each individual marker was not achieved. This shortcoming was offset by running the touchdown procedure, which uses multiple denaturation temperatures to increase the amplification product during PCR.

During the analysis of the data some of the limitations of STR markers presented themselves. Stutter was encountered particularly with D21S11. Stutter, a classic example of STR-related artifacts, is the presence of minor peaks one or more repeat unit length shorter than the parent allele. It is the result of a test-tube anomaly of DNA polymerase extension that mimics the naturally occurring process thought to be the source of STRs in the first place (Leclaire et al. 2004; Koumi et al. 2004). Stutter can be particularly difficult to control when alleles are one or two base pairs apart, as one allele can be masked by the other and misidentified (Moretti et al, 2001). Forensic typing of complex repeat sequences is particularly jeopardized. This is the case with marker D21S11, a putative tetranucleotide repeat marker for which the observed allele separation is often only two base pairs (FBI CODIS Core STR Loci STR Fact Sheet--D21S11). The potential of mistyping is further enhanced by "within-run" frameshifts of one base pair (Koumi et al. 2004) or the presence of "non-integer" or microvariant alleles (Weber and Broman 2001; Brinkmann et al. 1998). A recent population study examining 15 autosomal STRs (~12,000 allele measurements) revealed 160 heterozygous alleles that required a 1, 2 or 3 bp resolution up to about 300 bp due to microvariant alleles (Butler et al. 2004).

The most substantial result of this experiment, however, is to highlight the importance of performing multiple runs. It is clear from the figures that no single run yielded a complete, comprehensive genotype for each individual. Given that the information is being used in a forensic setting, less than 100% accuracy is not acceptable. Furthermore it is not just sufficient

for a run to yield an answer for each sample. Rather, it is essential that the answer be accurate. Thus even if a run performed perfectly, and all samples were designated with a call, how would we be sure that the calls obtained were accurate? Keeping in mind that there were no synthetic additions to the markers to increase performance it is possible to improve on the results, but it is unlikely to take care of all possible problems. So the real question to be asked is how many repeat runs are necessary to be confident in the results?

The sensitivity of individual markers is also clearly presented in both the tables and figures. Most of the forensic markers performed in the 90's or higher but there were a few markers that consistently performed less well. In light of the forensic application, prudence calls for the re-examination of at minimum the D21S11 marker in the forensic panel. Other, more reliable, markers with equivalent polymorphism rates are surely available. The individual genescan markers were more variable with fewer markers performing consistently high. The D17S1301 marker performed the most consistently high followed by D1S1677 and D10S1425. Further it is important to remember that the 16 genescan markers selected were done so randomly and as such there was no performance bias. It is possible that a panel of contemporary markers with the degree of polymorphism and reliability necessary for forensic profiling could be generated.

The family data were interesting and further highlight the importance of maintaining a minimum of 13 markers in a panel to ensure reliable identification. It is unlikely that first-degree relatives would match on all markers of a 16-marker panel but from the data it is clear that for some individuals it is not inconceivable to have a match up to 10 or 12 markers. The families selected are Mormon and as such have close kinship relations. Several populations throughout the world have greater consanguinity and would be at risk for misidentification if smaller marker

panels are employed in forensic analysis. The United Kingdom has already experienced such an event. An individual matched on 8 markers, was wrongly accused, but fortunately had an airtight alibi. Re-testing with more markers resulted in dismissal of charges. The culprit was later discovered and matched on 11 markers.

To date very little work has been done on this topic, save for the validation of the forensic markers and the frequencies of the polymorphisms in various ethnic populations (FBI, 1999). There is little in the literature about why these markers were selected for forensic use except that in the late nineteen-eighties, when they were developed for this use, they were state of the art (Butler 2001). The current literature on forensic DNA profiling focuses mainly on the need for accreditation and the problems related to extrinsic error (Liptak 2003; Liptak 2004; New York Times 2004; Sessions 2004; Scheck, 2003).

In summary we have shown that the markers that are currently used to generate DNA profiles perform as well as a randomly chosen set of contemporary markers. The real issue is how do we determine if the answer obtained is correct without performing multiple runs? The results of this study suggest that multiple runs are necessary to ensure the reliability of the profiles generated. Furthermore, it is necessary to determine the optimum number of runs required for reliability.

3.5 Implications and Perspectives

An alternative to STR markers in forensic work might be the use of Single Nucleotide Polymorphisms (SNPs). SNPs are single base differences in the DNA sequence and are usually diallelic. There are estimated to be over ten million SNPs with minor allele frequencies greater than 1% in the world population, as compared to STR estimates of 330,000 possible markers. Few SNPs had been well characterized until recently (Hinds et al. 2005; Weiner and Hudson

2002; International SNP MAP Working Group 2002; Cooper et al. 1985). Now, data from more than 11.5 million independent SNP loci (common and rare) are freely available from databases such as NCBI dbSNP (http://www.ncbi.nlm.nih.gov/projects/SNP/) and The SNP Consortium (http://snp.cshl.org) (Phillips 2005). Thanks to the rapid increase in validation, over a million SNPs have been genotyped in samples from four populations (International HapMap Project 2005).

Using SNPs in a forensic contexts is not a novel idea (Amorin and Pereira 2004; Jobling 2001; Gill 2001; Le Hellard 2002), but until recently, these markers were not considered to be a viable alternative to the predominant STR markers (Cooper et al. 1985; Sorbino and Carracedo 2005; Brion et al. 2004; Ye et al. 2002; National Center for Biotechnology Information 2005). The two-allele nature of SNPs is obviously disadvantageous for a single marker as compared to multi-allelic STRs in regard to their information content. In order to obtain equivalent discriminant power, 45-50 SNPs would be necessary to match the currently used 15 STR CODIS panel (Amorin and Pereira 2004; Gill, 2001). This ratio improves as the heterozygosity of SNPs increases: a simple problem to resolve given the hundreds of thousands of validated SNPs in public databases with high minor allele frequencies (between 0.4 and 0.5). Furthermore, given that new technologies (described below) allow efficient testing of large number of loci at low cost, one can design forensic SNP panels to provide significantly higher information content than microsatellites (John et al. 2004).

More importantly, the diallelic nature of SNPs makes them much more amenable than STRs to automated genotyping as no electrophoresis is required. A variety of assay methods such as single base extension (SBE), allele specific oligonucleotide hybridization (ASO), oligonucleotide ligation and invasive cleavage are available for SNP detection (see Sorbino &

Carracedo (2005) and Sorbino et al. (2005) for complete assay descriptions). Many instruments are available to detect genotypes generated using these different assays (Table 5). One can see that some instruments allow multiple SNPs to be tested in parallel, ranging from a handful to over 100,000. The multiplexing of marker loci is extremely efficient in terms of DNA consumption and cost per genotype making multiple runs viable under the constraints of limited sample size.

Of greatest concern to the field of forensic sciences is the accuracy of the genotypes. Genotyping accuracy rates of 99% or greater have been obtained for multiple technologies using different SNP assays (Table 1, Sorbino and Carracedo, 2005). In a comparison of genotyping error, SNPs performed significantly better than STRs [1/10,000 or 0.01% (Oliphant, 2002) versus 70/10,000 or 0.7% Weber and Broman, 2001)]. A side by side comparison of STRs and SNPs generated call rates of 95.57% and 99.96% respectively (International Multiple Sclerosis Genetics Consortium 2004). The SNP error rate was 0.02% compared to 1% for STRs (International Multiple Sclerosis Genetics Consortium 2004). The better accuracy of SNP genotyping assays is related to many factors. SNPs have lower mutation rates (International SNP MAP Working Group 2002). SNP assignment is unimpaired under conditions in which STRs face problems of strong peak imbalance, artifact signals and allelic dropout (although both marker types will show null alleles in cases where an amplification primer overlaps with an unknown polymorphic site).

With the advent of robust multiplexing technologies one can now see an obvious leap towards testing over 1,000 SNPs per forensic sample with high completion rates and a range of SNP markers which, tag sex and other demographic variables of forensic utility. Because of the extensive linkage disequilibrium in the human genome, the spacing between markers should be

far enough to avoid linkage disequilibrium among markers. These large marker panels could provide near-perfect discriminatory power for the purposes of forensic studies.

SNP genotyping will face the same intrinsic difficulties of working with forensic samples. These include contamination or adventitious transfer (Kimpton et al. 1996; National Research Council 1992; National Research Council 1996), mixtures (Duewer et al. 2001; Torres et al. 2003), inhibitory substances, poor sample quality or degradation (Kimpton et al. 1996; National Research Council 1992; Schneider et al. 2004) and limited sample size (National Research Council 1996). Forensic DNA typing will also involve matching genotype data obtained from an unknown suspect sample to samples drawn from potentially large populations, which may lack built-in consistency checks (National Research Council 1992).

It is not uncommon in forensic settings to encounter mixed samples (Torres et al. 2003). These samples are often typed with the expectation of excluding the victim's profile and generating a perpetrator profile to compare to profiles of potential suspects. In the case of SNPs, the diallelic nature makes mixture profiles difficult to distinguish without a priori knowledge of a mixed sample. One way to overcome this limitation will be to examine the entire array for increased heterozygosity (Gill 2004). Determining the genotypes of individuals once the presence of a mixture has been established will also be a challenge. It is possible to determine the mixture composition of heterozygous loci by focusing on allelic imbalance within the heterozygote samples, as indicated by a ratio of alleles that is different than the expected 50:50 seen with true heterozygotes. Quantitative genotyping methods of SNPs to detect imbalances are commonly used in many research applications that study allelic expression (Pastinen et al. 2004, Pastinen and Hudson, 2004), copy number variation in genomic DNA (J. Huang et al. 2004), and loss of heterozygoity in tumors (Zhou et al. 2004; Wong et al. 2004).

| Platform Name | Parallel Testing | Description of Assay | Measures of Accuracy |
|--|------------------|--|---|
| Taqman SNP Genotyping Assay (ABI) | Single-Plex | Fluorogenic 5' nuclease Assay | Error rate of <0.05%; Call rate of 99.95%; data concordance for duplicate runs and direct sequencing was 100% [Ranade et al. 2001] |
| Invader (Third Wave) | Single-Plex | Structure-specific activity of Cleavase [®] DNA enzyme and Sequence-specific hybridization | Genotyping efficiency 99.5% for duplicate runs [Nevil et al. 2002]; Accuracy rate 99.2% compared with genotypes from established sources; Failure rate 2%-3% [Mein et al. 2000]; 98% Genotyping accuracy with independent genotypes [Onishi et al. 2001]. |
| SNP Stream UHT (Beckman) | Up to 12-Plex | Tag array single base extension and hybridization capture of tagged SNP-IT [®] primers to array plate | Call rate 96% and Concordance rate 99.5% [Bell et al 2002]; 98.18% genotyping success (call rate) and 99.88% data concordance for previously genotyped samples [Jiang et al. 2004]; Confirm Bell accuracy rate of 99.5% [C.Y.A. Huang et al. 2004]. |
| MassARRAY™ with MassEXTEND (Sequenom) | Up to 15-Plex | MALDI-TOF MS (matrix-assisted laser desorption/ionization time-of-flight mass spectrometry) and Primer-extension chemistry MassEXTEND TM ; where each allele-specific primer extension product has a unique molecular weight allowing mass spec. genotyping | 90% successful genotypes (call rate) 99.7% accuracy with known genotypes [Beaulieu and Hong 2004]; 99.6% accuracy of genotype calls; Call rate 94%; Independent error rate 0.4% [Gabriel et al. 2004]. |
| ParAllele BioScience | 1000-Plex | Molecular inversion probe assay and microarray hybridization | Call rate 95%; Accuracy (compared with independent sequencing) 99.4% [Hardenbol et al. 2003]. |
| GoldenGate™assay Bead Array (Illumina) | 1536-Plex | Oligonucleotide labeled beads compatible with single- base extension, oligonucleotide ligation and allele- specific extension SNP Assays | Reproducibility and Call rates: 99.83% and 99.6% for 173,473 alleles; Reproducibility and Call Rates: 99.59% and 99.9% for 565,278 alleles; Strand correlation 99.35% and Call rate 97.7% [Oliphant et al. 2002]; Call rate 99.94%; Reproducibility 99.99%; Mendelian inconsistencies 0.005% and Double recombinant errors 0.03% yielded an estimated error rate of 0.035% [Murray et al. 2004]; Call rate 99.5%; Reproducibility below 0.1%; Mendelian error rate below 0.1% [Saba et al. 2005]. |
| GeneChip 10K Array (Affymetrix) | 6 to 10K-Plex | Parallel genotyping with GeneChip Tag Array | Call rate 92.7%; Concordance rate for repeated samples 99.7% [Sellick et al. 2003]; Call rate 95.8%; Concordance rate 99.1% with independent genotyping method [Kennedy et al. 2003]; Concordance rate >99.3%; Call rate of >97.5% [Lui et al. 2003]; Genotyping Accuracy (concordance) >99.5%; Reproducibility (consensus of 9 replicates) 99.99%; Call rate 95.9% [Matasuzaki et al. 2005]. |
| MegAllele™ (ParAllele BioScience) | 3 to 10K-Plex | Molecular inversion probe assay and GeneChip Tag Array hybridization | Completeness 98%; Accuracy: Repeatability 99.5-99.9% & Trio Concordance 99.6% [Hardenbol et al. 2005]. |
| GeneChip [®] Mapping 100K (Affymetrix) | 100K-Plex | GeneChip Tag Array | Call Rate 95.91%; Concordance with HapMap reference groups is 99.81%; Mendelian Error Rate 0.018%, At $\alpha = 0.05$; Inconsistency errors 47/4,976,938 with a consensus call rate 94.13; Consensus rate between DM and MPAM 99.90% [Di et al. 2005]; Concordance 99.93%; Median call rate per chip 99.1%; Minimum call rate per chip 95.6%; Median call rate per individual 99.1%; Minimum call rate per individual 96.7%; Reproducibility Xba Repeat concordance (4 replicates) 99.886%; Xba Positive control concordance (2 replicates) 99.870%; 10K concordance (3 replicates) 99.767% [Klein et al. 2005]. |
| Human-1 BeadChip (Illumina) | 100K-Plex | Sentrix Human-1 exon-centric genotyping BeadChip | Call Rates >99.9% and accuracy of 99.7% to 99.9% [Gunderson et al. 2004]; Concordance >99.7%; Accuracy 99.7 to 99.9% [Fan et al. 2003]. |

An alternate way to circumvent the difficulties of mixtures for SNPs would be to incorporate established Y-SNP (Vallone and Butler 2004) and X-SNP marker sets; as males are hemizygous for these loci and should only generate single alleles for these markers; heterozygosity at these loci could allow profiles to be deduced more easily from individual contributors.

A probable advantage of SNPs in the context of testing forensic specimens is their diallelic nature which may make them more amenable to typing under degraded conditions or poor sample quality, or where the quantity of DNA is significantly limited, as STRs require a minimum fragment length range of 100-360 base pairs of template DNA versus 57 to 146 base pairs for SNPs (Dixon et al. 2005; Divine and Allen 2004; Lessig et al. 2004; Sanchez et al. 2003).

Recent commentary on the use of SNPs vs. STRs in forensic DNA testing suggests that SNPs will occupy a niche domain but will not be widely adopted over STRs (Butler, Colbe and Vallone 2007). While acknowledging the technological advances, the improvement in multiplex amplification and the selection of optimal SNP loci (Dixon et al. 2006; Sanchez et al. 2006; Kidd et al. 2006), these authors suggest that miniSTR markers work equally well on, the primary reason to consider SNPs, degraded samples. The drawbacks cited as most intractable are the increased number of loci ³ and mixture decoding. Further, and more importantly they discuss the difficulties in deciphering and manipulating more information related to the increased number of markers necessary for robust identification with SNPs, the lack of a standardized forensic SNP platform and the existing infrastructure supporting STRs.

Niche applications such as Y-SNPs, mitochondrial DNA (mtDNA), ancestry informative markers (AIMs) or phenotypic markers were suggested for future SNP forensic genotyping.

³ Work has been done to generate a 50 SNP multiplex (Sanchez et al. 2006).

3.6 Conclusion

DNA profiling is considered to be a powerful forensic tool and in a justice system that claims that it would rather let a guilty person go free than incarcerate an innocent person, the standard for DNA evidence needs to be 100%. The results of the marker comparison clearly indicate that the markers that are currently used to generate DNA profiles perform as well as a randomly chosen set of contemporary markers. However, errors continue in proficiency testing, the switching of evidence samples, and false positives to name a few⁴ (Teichroeb 2004a; Teichroeb 2004b). Alternative markers might help to redress some of the lab errors and provide additional forensic information. SNPs are thought to have a promising future in forensic and anthropological genetic analysis (Butler, Colbe and Vallone 2007; Le Hellard et al. 2002; Vallone and Butler 2004). SNPs are more abundant, more robust, have a lower mutation rate and though their lower polymorphism information content requires more alleles to be interrogated, this can be easily accomplished with recent technological advances. Genotyping costs for SNPs are low due to easy automation, high-throughput, efficient use of sample, and reduced error rates. Most of the current challenges in regards to SNP use-- limited multiplex marker panels, mixtures, discriminatory power and cost--can be or are in the process of being overcome. The argument most intractable against the use of SNPs in forensics is the investment already made in STR profiling, the CODIS profile database and implementing the necessary information technology infrastructure attendant with database searching that would need to be done to modify CODIS. The importance of accuracy and reliability cannot be overstated in forensic work, as the results have a profound impact on victims, the accused, their families, and

⁴ A false match was made in a proficiency test by SERI in California in September of 1997, (ii) evidence samples were switched at the Minnesota BCA in October of 1997, (iii) the errors in the APEX proficiency tests, and (iv) the Philadelphia Police Department false positive in 1999. See http://darwin.bio.uci.edu/~mueller/error_rate.html.

other potential victims to name only a few. As the investment in SNP technology increases and multiplex panels become more refined, SNPs will likely compliment STRs in forensic DNA profiling. Until the time that multiple runs are the standard in forensic case work the estimate of laboratory false positive rates and match probability rates need to be presented at trial so that the evidence can be given the proper weight.

Though the DNA profiling paradigm may be imperfect, it is still better than previous techniques and of great benefit to society in two ways: the detection of perpetrators of criminal offences and the exoneration of the innocent. How this powerful tool impacts society is of paramount importance with far reaching consequences, not just in terms of altering criminal behavior, but in terms of altering the behavior of all citizens.

Little has been written on the broad societal impact of profiling outside of the exoneration and detection of individuals. How will this change the relationship between the citizen and the state? How does this potentially impact our conceptualization of civil liberties as opposed to our concerns for security? Will this impact privacy and change the fundamental relationship of the citizen to the state? Will social and racial inequities lead over-representation of groups within the forensic DNA databanks? Will unbridled use of DNA profiling lead to the erosion of our social pact? What areas should be of most concern as we seek to minimize potential undermining of our social contract?

These questions are the topic of the next chapter of this thesis.

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CHAPTER 4: ETHICAL AND LEGAL FRAMEWORK FOR FUTURE POLICY DEVELOPMENT

4.1 Introduction

"Baton Rouge, Louisiana

Of the more recent cases involving police use of DNA searches, the Baton Rouge, Louisiana search has drawn considerable criticism. In the search for a serial rapist, based on a forensic profile, in 2002 Baton Rouge Police began gathering DNA samples from white males. Additional information directed police to individuals who may have driven a white pick up truck. Men who refused were fearful of public exposure as a suspect and complied with police requests. Critics of the Baton Rouge search claim that many were coerced into giving samples. The use of DNA was criticized due to lack of other investigative activity that would have cleared individuals. In all, over 1200 men were sampled. A suspect was later arrested, convicted and sentenced to death in the case. Derrick Todd Lee is an African American and did not drive a white pick-up truck. His arrest and conviction came through the investigation of an unrelated incident. The DNA samples gathered were not returned or destroyed after suspects were eliminated. Some of those who were sampled have filed suit to have their DNA returned or destroyed" (Walker 2004). Mr. Shannon Kohler was one of those men. He initially declined to let police take a DNA sample and in November 2002, the Baton Rouge Police Department obtained a seizure warrant to force Mr. Kohler to submit his DNA sample for the investigation. At that time, Mr. Kohler was identified by the police and news media as a suspect in the highly publicized search for a serial rapist-murderer. Mr. Kohler was cleared by police as a suspect in the investigation.

Subsequently, "Mr. Kohler filed a Section 1983 claim in the Middle District of Louisiana alleging that the seizure warrant used to obtain his DNA lacked the required probable cause. He has asked for his DNA profile to be removed from any state or federal database and has requested damages for the invasion of his privacy in violation of the Fourth Amendment. In February 2005, the District Court dismissed Mr. Kohler's claim on a Motion for Summary Judgment, finding that police had probable cause based on two anonymous tips (a 1982 burglary conviction, Mr. Kohler's place of employment in 1991) and the fact that Mr. Kohler met "certain elements of an FBI profile," which the Court characterized as "so broad and vague that it cast a net of suspicion over thousands of citizens." The Court rejected Mr. Kohler's request for a new trial on the issues. Mr. Kohler has filed an appeal with the Fifth Circuit Court of Appeals" (Electronic Privacy Information Center 2005a,b).

Innovations in molecular biology have led to dramatic changes in the practice of forensic

sciences and the relationship of science to the criminal justice system. DNA profiling has not

only become the gold standard of forensic evidence (Lynch 2003), but has also facilitated the apprehension of culprits, the conviction of the guilty and the exoneration of the wrongfully suspected and the wrongly convicted (Innocence Project 2007; Scheck, Newfeld and Dwyer, 2000). Despite these obvious benefits, the application and expansion of this technology raise serious ethical issues. These issues are mentioned repeatedly, but few have been examined in depth from an ethical perspective. Can the existing legal framework address key issues related to the endless expansion of forensic DNA identification or is an innovative approach required? The analysis below will outline the philosophical and legal underpinnings and the following arguments will draw on law, political philosophy and ethics.

4.2 The Philosophical Underpinnings

Political philosophy is the foundation on which any theory of criminal justice rests, as it is the study of the fundamental questions about government, politics, property, law and the enforcement of law (Bowie 2008). Broadly, political philosophy concerns the nature and forms of power and more specifically, it concerns the principles for proper governance (Bowie 2008). The bases for the majority of political philosophies in the present era are the ideals of liberty, property and rationality extolled in The Age of Enlightenment (1600's-1800's) (Dupré 2004). The belief in a rational, orderly and comprehensible universe inspired Enlightenment thinkers to form a rational and orderly organization of knowledge and the state (Dupré 2004). The Enlightenment movement began from the assertion that law governed both heavenly and human affairs. Law was conceptualized as a relationship between individuals, with a refined focus on individual liberty as a fundamental right of man, given by "Nature and Nature's God" (US Declaration of Independence 1776). In the ideal state, individual liberty would encompass as many people as possible. Thomas Hobbes in 1651 argued that if we lived in a state of nature,

without society, we would each have unlimited natural freedoms, but at the cost of security, because anarchy and general warfare would reign (Hobbes, edited by Curley 1994). As a political theory, Hobbes proposed the social contract whereby people ceded their individual rights to the state, creating the sovereign state, in return for protection and a functional society (Hobbes, edited by Curley 1994). In this implicit social contract we each gain civil rights in return for accepting the obligation to honor the rights of others, consequently giving up some freedoms to do so. Thus the social contract evolved out of pragmatic self-interest. However, according to Hobbes, any abuses of power by the sovereign authority were to be accepted as the price of peace (Hobbes, edited by Curley 1994).

Though a contemporary, John Locke's version of Hobbes' social contract theory was distinctly different. Besides believing that man was "naturally" good, he understood the natural state to be the source of all rights and unity, with the government's role being to secure the state of nature and to allow it to flourish (reprinted in Shapiro 2003; reprinted in Yolton 2004). The cession of individual rights forms the state, which is then obliged to secure the rights of individuals. In contrast to Hobbes, Locke asserted that citizens contracted with one another to form a particular kind of government, and that they had the right to rebel against tyranny, to modify or even abolish their government (reprinted in Shapiro 2003). Accordingly, to some philosophers (including Locke and Rousseau) the civil rights granted under the social contract are not permanently fixed but rather are a means to an end, whereby the contract is only legitimate to the extent that it satisfies our goals. Thus the contract can be renegotiated, to the benefit of all, using means such as the legislature and elections (Shapiro 2003; Rousseau, cited in Bertram 2004).

4.2.1 Foundational Philosophy, Deontology and Utililatrianism

In addition to yielding novel theories of governance the Enlightenment also generated new theories governing behavior within society. The application of order to society lead to Foundationalist theory, which was rooted in the idea that one must create a foundation from the most general and abstract truths and then reason deductively to work out the particulars (Hyland, Gomez and Greensides 2003). The two main western foundationalist philosophical theories are deontology and utilitarianism. Deontology alleges that there are a set of permanently defined, unwavering principles by which people ought to live. The existence of *a priori* moral obligations implies that decisions should be made by considering one's duties, the rights of others and fulfillment of one's obligations. Participation in civil society, argued Immanuel Kant, is undertaken not for self-preservation, as Thomas Hobbes believed, but as a moral duty (Kant, translated by Wood 2002; Kant, translated by Gregor 1998). Kant, a German philosopher, examined the structure and meaning of obligation and asserted that particular kinds of acts are morally wrong because they are inconsistent with the status of a person as a free and rational being. He cautioned that immoral actions could never be justified by their consequences and should therefore never be carried out under any circumstances. Kant is probably best known for his admonishment of the treatment of people as means: "Act as to treat humanity, whether in your own person or in that of any other, never solely as a means but always also as an end" (Kant, translated by Wood 2002). Jeremy Bentham, on the other hand, criticized deontology on the grounds that the unchanging principles were subjective opinion and that it was essentially a variation of popular morality.

Conversely, utilitarianism is a consequentialist approach in which the ends justify the means because decisions are judged primarily in terms of their consequences. The basic

principle of utility is often summarized as "the greatest good for the greatest number" but was most clearly stated by John Stuart Mill, whereby "[a]ctions are right to the degree that they tend to promote the greatest good for the greatest number" (Mill, edited by Sher 2001).

Utilitarian balancing consists of considering the needs of everyone affected, the goals and the resources available, then calculating a course of action. Even when utilitarian philosophers disagree as to whether we ought to maximize happiness or other values, they all agree that the assessment of the greatest good should be in terms of the total intrinsic value produced. Mill argued that when rights and duties conflict, deontologists often fail to specify which principles should take priority, thereby offering incomplete moral guidance. There are however problems with consequentialist arguments in that there are actions that would be considered by most to be immoral that would still be tolerated, maybe even obligatory, because they maximized the good, such as killing, torture and a duty to commit suicide. In these cases the rights of the minority are subrogated to the interests of the majority. Further, utilitarianism is indifferent to unjust social distributions because justice is not given independent weight (Beauchamp and Childress 2004). The principle of utility can, however, play a meaningful role in formulating public policy via the objective assessment of everyone's interests and the impartial maximization of positive outcomes for affected parties. As utilitarian theory is about promoting welfare, it can also be characterized as beneficence-based as well as consequentialist (Beauchamp and Childress 2004).

We do not live in either an entirely deontological or an entirely utilitarian society. Because we live in political systems, we do not have the luxury of examining moral theories out of context, but rather must contend with the framework within which they are situated. We as individuals have selected aspects of these theories and employed them to help make, justify and defend difficult moral choices. Rights language has been used to buttress political morality by

protecting individuals from societal intrusions through legitimizing the role of political, civil and legal rights. "No part of the moral vocabulary has done more to protect the legitimate interests of citizens in political states then the language of rights" (Beauchamp and Childress 2004). The rights language was employed by Hobbes in his contractarian framework where all human beings have rights or moral claims that protect their basic interests. However, Hobbes' concern was with who determines what the rights are and who will exercise the vital political powers when we all share the same entitlements (Hobbes, edited by Curley 1994).

4.2.2 The Liberal Political Framework

Canada and the United States are both liberal political societies. They embrace social systems that define value as determined by the individuals living in the society. The foundation of society in general is rooted in mutual consent for mutual advantage (Locke, reprinted in Shapiro 2003; Rawls 1971:4). "The life of the free individual cannot be separated from the protections afforded through social order" (Novak 1989). The liberal political context governs our social relations and is limited by the political rights of individuals. We are obliged, as free persons relishing liberty and finding its justification in our endowment with "unalienable rights," to recognize these same rights in other human beings (Novak 1989). Thus a plurality of values must coexist, with none given a privileged position over any other. Ensuring that this remains so is the responsibility of the community. It is the community that secures the rights of individuals through institutions worthy of human dignity. Institutions are conceived as a way to secure both the public good and private rights. For pluralistic societies to exist and secure the conditions under which personal liberties thrive, they must establish institutions and rules to proscribe particular behaviors that threaten the benefits of mutual cooperation. "At the political level coercive rules in liberal societies are, in theory at least, subject to ethical constraints. These

derive from the premise that the liberal state exists to support individual autonomy and freedom rather than to restrict it" (Wilson 2002; Kant as cited by Paton 1948). The proscriptive rule, far from restraining liberty, secures it for all, leaving free a wide expanse of liberty for creativity and enterprise. Liberty in this sense is forever tied to law, as it rests upon law. Individuals are protected by coercive rules and are able to use these rules as standards by which to conduct themselves with the minimum of state interference (Wilson, 2002). Thus institutions and rules allow for the defense of a full pluralism of intentions, aims and purposes when they are appropriately delimited (Novak 1989). The basic institutions of society must be agreed upon by rational members of the society to secure mutual advantage. Each member must internalize the concept that he or she will be better off under the terms of the institution than in its absence. 4.2.3 *Liberalism, The Social Contract and the Justification of Punishment*

Accordingly basic institutions like medical care, education, national defense and law enforcement are examples of systems to which we consent, or would if the choice were offered, as rational agents living within a liberal society. The pluralist society, with its institutions and rules, is entirely rooted in choice (autonomy) and the ability of its members to give consent and it is this society which is the guardian and nurturer of autonomy. This theory implies that there is a choice; there is somewhere else to go, when in reality tacit consent maybe coerced because there is no alternative. However, it also presupposes that the choice is not limited to tacit consent or expatriation, but includes accepting the contract in principle while participating in the political process to alter parts that are disputed. Regardless of whether we make a contract with each other or the state, it would not do for one to feign consent to gain the benefits of mutual cooperation, but then defect and behave in ways that damage such cooperation, hinder mutual benefit and subjugate the public good. There must be some way to ensure continued compliance

with the social contract. The absence of any form of punitive measure for violations of the social contract would entirely eliminate the possibility of social cooperation and indeed society as a whole. "Any voluntary agreement must set out consequences for violations, along with a plausible enforcement mechanism for detecting violations and imposing the announced penalties" (Finkelstein 2005). As any agreement, commercial or otherwise, would unravel without the threat of enforcement for nonconformance, a system of punishment will be a prerequisite of the contract that sets out substantive rules of compliance. The social contract is no different; in theory the self-imposed threat of punishment is the assurance that is offered to fellow citizens that one will not defect (Finkelstein 2005). Some argue that punishment is meant to act as a deterrent the moment one enters into the original social contract and all of social order is grounded on consent, just as punishment is consented to by the offender at the moment he enters into the contract. Alternately, it can be asserted that punishment is "mutual coercion, mutually agreed upon", as rights come from agreeing to the social contract, and these rights can be revoked when violations of contractual obligations occur (Hardin 1968). Those who fail to conform to the contract risk losing some of their rights. Defectors who receive punishment under either system still fare better under a punitive agreement than they would under a system devoid of all social order. As members of society we further its interests by following its rules and accept responsibility along with the threat of punishment for violation of the rules (Rawls 1955).

The paradox of autonomy as the guarantor of the legitimacy of state coercion is reflected in the harm principle (Wilson 2002). The harm principle originally discussed by John Stuart Mill in his essay 'On Liberty' has at its core two concepts: that the state has the authority (1) to criminalize the causing of harm and activities leading to harm and (2) to accomplish this goal with minimal interference while guaranteeing maximization of individual autonomy. In general,

we are comfortable with laws punishing harmful behavior and carried out by the institution of criminal justice because we are concerned about others harming us and we do not plan on harming others. The harm principle not only limits state coercion, but also serves to maintain moral neutrality and ensure that the interests of the citizenry are served rather than the interests of the state (Wilson 2002; Raz 1986).

4.2.4 Rights and Moral Principles

Western liberal societies are based on recognizing the rights of the individual and securing them. Rights are claims that are justified by principles or rules either moral, political or institutional. Within a society rights exist or fail to do so as long as the relevant rules allow or disallow the claim in contention. Autonomy and the right to self-determination are key in liberal democratic societies. The tension between asserting individual rights and securing society implicates more than just autonomy. The overarching dilemma is composed of other rights, each of which should be elaborated upon within the context to elucidate the pertinent ethically legitimate claims involved. North America and many other constitutional democracies in general have agreed upon principles of human rights enshrined internationally in the Universal Declaration of Human Rights and UNESCO policy documents. The guiding principle in each of these documents is respect for persons or human dignity, which has its roots not only in moral philosophy but in the philosophy of law as well (Morris 2005; Beauchamp and Childress 2004). This principle forms the foundation of our primary ethical obligations and the correlative principles of justice, the right to life, security of the person, respect for vulnerable persons, respect for privacy and confidentiality, and in the application of these principles the necessity to balance harms and benefits. The analysis presented draws on the relevant key principles.

4.2.5 Ethical Principles

Respect for human dignity, as a principle, strives to protect the numerous and often interdependent interests of the individual, bodily, psychologically and culturally. The cornerstone of human dignity is autonomy. Securing autonomy in its various forms by the state is not always clear cut but instead requires balancing interests both between individual citizens and the state. The primary way in which the state secures autonomy is through the granting of rights. All rights are presumptively valid (prima facie) claims that must sometimes yield to other claims. They capture the purpose of morality in securing liberties and other benefits for the citizen and form the justification of obligations (Dworkin 1978; Beauchamp and Childress 2004). They should, however be considered the minimal enforceable standard for the mutual treatment of individuals and communities. "[S]ocial ideals and principles of obligation are as critical to social morality as rights, and ... neither is dispensable" (Beauchamp and Childress 2004).

The moral imperative of respect for human dignity defines duties and obligations, one of which is ensuring justice. Respect for justice and inclusiveness seeks to regulate behavior in a fair and equal manner. Thus procedural justice requires that the process be fair in methods, standards and procedures, while distributive justice concerns the fair distribution of benefits and burdens. Distributive justice imposes obligations of protection toward vulnerable individuals to ensure that they are not exploited and that no segment of the population is unfairly burdened with harms. At the same time, distributive justice also imposes duties to ensure that the interests of those who may benefit, either individuals or groups, are not abrogated or neglected.

Respect for vulnerable persons is derived from respect for human dignity and the correlating respect for justice. Respect for human dignity imposes strong ethical obligations of

protection toward those vulnerable members of society limited by a diminished capacity for decision-making. Justice or fairness, in this case, demands protections against exploitation, discrimination and abuse of these vulnerable members thereby safeguarding their interests.

Safeguarding interests is paramount to the respect for privacy and confidentiality, which is presupposed as fundamental for the respect of human dignity. Respect for privacy and confidentiality ensures that access, control and dissemination of personal information is protected. In part, respect for human dignity is demonstrated through respecting autonomous wishes not to be observed, touched, or intruded upon. "On this account, rights of privacy are valid claims against unauthorized access that have their basis in the right to authorize or decline access" (Beauchamp and Childress 2004). Rights to privacy are infringed when those who are unauthorized obtain access to personal information, whereas confidentiality is infringed when entrusted information is unprotected and disclosed without consent (Bauchamp and Childress, 2004).

Critical to the application of ethics is the balancing of harms and benefits of an action or proscription, the analysis of which requires a favorable ratio whereby harms should not outweigh benefits. This analysis impacts the welfare and rights of individuals, the ethical justification for various options or preferences and the informed acceptance of harms and benefits. As we move to consider specific ethical issues which arise in the forensic domain, rights to privacy and autonomy and the balancing of harms and benefits are most immediately implicated. In the context of arguments presented in this thesis, to balance harms and benefits we must identify issues that relate not just to the use of DNA, but also to the context in which it is acquired, so as to completely characterize the relationship between the individual and society.

4.3 Forensic DNA Applications

As we examine the ethical principles surrounding the conduct of individuals toward one another and begin to consider the use of DNA in a forensic milieu, several specific questions spring to mind. Does the outcome justify the intervention (Childress et al., 2002)? With respect to identifying the perpetrator of a crime, to what end do we identify him or her? If we assure society that a perpetrator will be identified does this help to deter the commission of crime, or is it simply a means of reinforcing retribution through punishment? If we accept that punishment is a self-imposed assurance and is freely consented to under the social contract, one can argue that it meets both of the chief aims of the criminal justice system, retribution and deterrence (Finkelstein 2005).

Critical to the discussion is our conceptualization of what DNA is at a philosophical level and how it is variously perceived. As noted in the introduction, DNA has been argued to be different in that it is not solely a unique identifier as in the case of fingerprints or voice exemplars but, in addition to its potential accuracy and reliability, it contains information about our genetic basis not just as humans but as unique individuals. DNA has the potential to unlock our body's secrets and reveal a host of current and future health information. This information cannot only impact our choices but the decisions made by our children, siblings and other family members. By allowing the use of DNA analysis, are we allowing the state intimate access to what makes us unique as individuals? Is using DNA for identification equivalent to treating people as objects, equating them to molecules to be codified, dehumanized, commodified, and objectified? What are the limits for the state to identify and ultimately track its citizens? Should there be limits on such surveillance by the state with respect to identifying and tracking its citizens?

Before addressing these questions, it is first necessary to describe the various ways in which DNA samples may enter the criminal justice system.

4.3.1 Forensic Acquisition of DNA

4.3.1.1 Crime Scene

In the forensic setting, the acquisition of a DNA sample can follow several paths. The first and most basic collection occurs when a sample of biological material is left at a crime scene. Thereafter, a DNA profile is determined and placed in a database/databank, either local or federal, to await comparison with other profiles.

4.3.1.2 Voluntary Exclusionary Samples

Often as part of an initial investigation samples are requested from close friends and relatives to exclude their DNA from crime scene material and eliminate legitimate trace evidence from potential suspect trace evidence. The DNA and attendant profile are retained until the closure of the case and may be retained indefinitely depending on the jurisdiction under which it is collected.

4.3.1.3 Warranted Seizure

During the investigation of a crime, the agents of the criminal justice system may uncover enough circumstantial evidence toward a suspect to shift the balance from individual freedom toward group security, thereby justifying a warrant. In this event the evidence is brought before a judge who evaluates, weighs and affirms that it meets the necessary justification to support the infringement of an individual's rights in the interest of security and justice. If the evidence supports the cause for the warrant, in this case the acquisition of a DNA sample, it will be issued and the DNA analyzed to reveal a profile comparable with the crime scene sample. The warrant should stipulate the limits in comparison with the incident or other crime scene

samples and specify the duration of retention, destruction or return of the sample and the maintenance or elimination of the profile from the database.

4.3.1.4 Acquisition on Arrest

The fourth mode of criminal DNA acquisition is available in those regions where arrestee testing has been legally approved, which varies from province to province and state to state. In this instance, if there is probable cause to arrest a suspect of a crime, a sample can be obtained, a profile generated and compared with unsolved crime scene samples. In the most permissive statutes the suspect does not have to be convicted of the crime for which they were arrested and neither the sample nor the profile need be destroyed. Both are retained and the profile is entered into the local database. In the US, arrestee profiles are now a component of the CODIS federal database (Senate Bill 1197, 2005).

4.3.1.5 Postconviction

For individuals who have already been convicted of those specific crimes deemed by the state to be either particularly heinous or to have a high probability of yielding a trace biological sample, DNA samples are typically collected, processed and placed in a profile database during the period of incarceration (Axelrad 2005).

4.3.1.6 DNA Dragnets

This collection pathway is from the community at large in what have been called DNA "dragnets" or "sweeps" (Walker, 2004). Under these circumstances individuals are asked to provide a sample for elimination purposes. By not specifying the specific comparison case, the profile can be compared with all crime scene samples and both the sample and profile are typically retained. It has been argued that the consent obtained in these circumstances is neither

informed nor free from coercion (American Civil Liberties Union 2003; Wilson, 2002; Halbfinger 2003).

4.3.1.7 Discarded DNA Acquisition

Lastly, sample collection can occur via the acquisition of discarded items such as coffee cups, chewing gum, used tissue and in some cases even fingerprints can yield DNA (Phipps and Petricevic 2007; Balogh et al., 2003). In this instance the potential suspect is unaware that a biological sample has been collected for DNA isolation, or that a profile has been obtained and compared with that left at a crime scene. Sample and profile retention in these cases varies with jurisdiction. Considering the financial expense involved in processing samples, there is no reason to believe that the sample and profile are not retained and placed in the databank based on the treatment of voluntary samples from DNA sweeps (Walker, 2004; *Shelton* v. *Ann Arbor Police Department*, 1995⁵).

As we are at the interface between the use of forensics in law enforcement and the law it is important to assess and clarify the differences between them. Law enforcement is the physical application of the law and must base decisions and actions on information available at the moment. The law has time to reflect and carefully weigh positioned arguments and as such has time to consider subtle nuances that maybe lost in the application by law enforcement.

⁵ From 1992 to 1994, a serial rapist sexually assaulted or attempted to sexually assault up to ten women or more in Ann Arbor, MI. Michigan State Police gathered blood samples, in 1994, from 160 African-American men in an attempt to identify the serial rapist. The description was of a black male between the ages of 25 and 35 years old and between 5'7" and 6'2" tall. The dragnet failed to identify a suspect. The rapist, Earvin Mitchell, was eventually identified when he was arrested for an attack of another woman. During the sweep the police never disclosed that law enforcement agencies would keep the obtained DNA profile on file for possible use in other criminal investigations regardless of whether the profile excluded them as the serial rapist. The DNA and profiles were retained until a civil and class action suit were filed requesting the return or destruction of the DNA. Michigan State Police were ordered to destroy or return the DNA in 1997 as well as paying monetary damages (Hansen, 2004; Robinson, 1997; Sasser-Peterson, 2000; ACLU raises, 2003).

4.4 DNA Analogies

One way in which to explore DNA collection is to examine legal rulings concerning the forensic use of other relevant personal materials or bodily products, such as fingerprints, hair, body parts and waste.

4.4.1 Fingerprints

The most common legal analogy is to compare the use of acquired DNA with the use of fingerprints for identification. This comparison has face plausibility because fingerprints and DNA are equivalently deposited as we go about our daily activities. In the fingerprinting context, there exists a constitutionally significant distinction between the gathering of fingerprints for identification purposes from persons within the lawful custody of the state and the gathering of fingerprints from persons to determine their guilt of an unsolved criminal offense. The Fourth Amendment does not bar the fingerprinting of a properly seized person, so long as the initial seizure of the person is reasonable (a lawful arrest) subsequent fingerprinting is permissible.

The US Supreme Court in *Davis* v. *Mississippi*⁶ suggested that the taking of fingerprints, may "under narrowly defined circumstances", during the course of a criminal investigation, of individuals for whom there is no probable cause for arrest may comply with the Fourth Amendment and would thus not be afforded protection as "Fingerprinting involves none of the

⁶ Meridian Mississippi police, in connection with a rape investigation, brought numerous Negro youths to the police station for questioning and fingerprinting without warrants. The petitioner was questioned, fingerprinted, and released. Thereafter, concededly without a warrant or probable cause for arrest, the police drove petitioner to Jackson and confined him in jail overnight. Subsequently he was questioned, signed a statement and was returned to Meridian and jailed. During confinement he was again fingerprinted and the prints were sent to the FBI for comparison with prints left in the victim's home. The fingerprint evidence was admitted at petitioner's trial for rape, over objection that it was the product of unlawful detention, and he was convicted. The Mississippi Supreme Court upheld the conviction. The U.S. Supreme Court held that: "fingerprint evidence is no exception to the rule that all evidence obtained by searches and seizures in violation of the Constitution is inadmissible in a state court". Pp. 723-724. The Fourth Amendment applies to involuntary detention occurring at all stages of the investigation and detentions for the sole purpose of obtaining fingerprints are also subject to the constraints of the Fourth Amendment requirements could be met by narrowly circumscribed procedures for obtaining, during a criminal investigation, fingerprints of persons for whom there is no probable cause to arrest, since no attempt was made in this case to employ procedures which might comply with the Fourth Amendment. P. 728. As such the verdict was reversed.

probing into an individual's private life and thoughts that marks an interrogation or search" (*Davis* v. *Mississippi*, 394 U.S. 721, 727, 728; 1969).

However, the gathering of fingerprint evidence from 'free persons' constitutes a sufficiently significant interference with individual expectations of privacy that law enforcement officials are required to demonstrate that they have probable cause, or at least a cogent suspicion, that the person committed a criminal offense and that the fingerprinting will establish or negate the person's connection to the offense. (*Hayes* v. *Florida*, 470 U.S. 811, 813-18, 1985⁷; *Davis* v. *Mississippi*, 394 U.S. 721, 726-28, 1969).

Other courts have offered the rationale that because fingerprints are part of routine identification procedures and are easily accessed and constantly exposed to public scrutiny, one cannot hold a reasonable expectation of privacy to such publicly available knowledge (*Smith* v. *U.S.*, 324 F.2d 879, 882 D.C. Circuit 1963; *Napolitano* v. *U.S.*, 340 F.2d 313, 314 1st Circuit Court 1965⁸). Similarly, the Supreme Court has said that individuals do not possess an expectation of privacy in their personal characteristics (*United States* v. *Dionisio*, 410 U.S. 1, 1973⁹). To whit, the police may require individuals to give handwriting and voice exemplars, as

⁷ In Punta Gorda, Florida, the police, without a warrant, went to the home of the principal suspect in a burglary-rape to obtain fingerprints. Arriving at the home, the police spoke to the suspect and when he expressed reluctance to accompany them to the station house, one officer said that they would arrest him. Petitioner replied that he would rather go to the station than be arrested. He was then taken to the station and fingerprinted. When it was determined that his prints matched those taken at the scene of the crime, he was arrested. The trial court denied his pretrial motion to suppress the fingerprint evidence, and he was convicted. The Florida District Court of Appeal affirmed, holding, although finding neither consent by petitioner to be taken to the station nor probable cause to arrest, that the police could transport petitioner to the station house and take his fingerprints on the basis of their reasonable suspicion that he was involved in the crime. The U.S. Supreme Court on appeal held that "Where there was no probable cause to arrest petitioner, no consent to the journey to the police station, and no prior judicial authorization for detaining him, the investigative detention at the station for fingerprinting purposes violated petitioner's rights under the Fourth Amendment, as made applicable to the States by the Fourteenth; hence the fingerprints taken were the inadmissible fruits of an illegal detention".

⁸ See Smith v. U.S., 324 F.2d 879, 882 D.C. Cir.1963) (Burger, J.) ('it is elementary that a person in lawful custody may be required to submit to ... fingerprinting ... as part of the routine identification processes'); Napolitano v. U.S., 340 F.2d 313, 314 1st Cir. 1965) ('Taking fingerprints [prior to bail] is universally standard procedure, and no violation of constitutional rights').

⁹ The compelled production of the voice exemplars did not violate the Fifth Amendment privilege against compulsory self-incrimination, since they were to be used only for identification purposes, and not for the testimonial or communicative content of the utterances. Pp. 5-7. Respondent's Fourth Amendment claim is also invalid as a subpoena to compel a person to appear before a grand jury does not constitute a "seizure" within the meaning of the Fourth Amendment, and the fact that many others besides respondent were ordered to give voice recordings did not render the subpoena unconstitutional. Pp. 8-13.

well as hair, blood, DNA, and fingerprint samples, without complying with the Fourth Amendment's requirements.

4.4.2 Hair

Hair, because of its public accessibility, has also been considered unworthy of Fourth Amendment protection (*Mills*, 686 F.2d 135,139 3d Cir. 1982¹⁰; *Coddington* v. *Evanko*, WL 2416429 3d Cir. 2004¹¹). Sherry Colb argues that Coddington "misinterprets" Fourth Amendment law and that both the Mills and Coddington courts are "wrong as a matter of logic" in that exposure of the item, in these cases hair, does not mean that there is no privacy interest attached or that taking something that is physically attached from one's body does not constitute a seizure (Colb 2004). Unlike fingerprints or voice exemplars which can be given without loss of the items in question, hair once cut is "seized". Further, Colb asserts that the act of shaving one's hair is somewhat intimate and that doing so "without consent may also invade what many of us would consider a reasonable expectation of privacy" (Colb 2004).

4.4.3 Body Parts

Another potential analogy would be to think of DNA as a part of the human body. Historically the law has treated human bodies as property. Slaves were bought, sold and traded as property. The tradition of conceptualizing the body as property owned by the inhabitant,

¹⁰ The Appeals Court concluded that there was no greater expectation of privacy with respect to hair which is on public display than with respect to voice, handwriting or fingerprints. In the case of blood samples and fingernail scrapings, the bodily seizure requires production of evidence below the body surface which is not subject to public view. In the case of facial and head hair, as well as fingerprints, voice and handwriting exemplars, the evidence is on public view. In his concurring opinion, Judge Gibbons makes the distinction that it is only the "appearance of one's hair that we offer to the public's view. The cutting of a few strands of hair is more like the involuntary touching, inking, and pressing of one's finger involved in the process of fingerprinting." If fingerprints can be subject to compelled disclosure by the grand jury without implicating the Fourth Amendment, it follows logically that hair strands can as well.

¹¹ The U.S. Court of Appeals for the Third Circuit ruled that police officers may constitutionally shave large amounts of hair from a suspect's head, neck, and shoulders, without a warrant, probable cause, or any basis for suspecting that the hair would provide evidence of crime. On April 5, 1999, William Coddington, a Pennsylvania State Trooper, reported for work at which time several of his superior officers relayed that a confidential informant indicated he was using cocaine. Coddington was ordered to submit to hair sampling so that his hair could be tested for cocaine and other drugs. A sergeant at the police station cut hair from Coddington's head, neck, and part of his left shoulder blade. Coddington was then informed that additional hair had to be taken and the subsequent shaving resulted in bare spots on his scalp. When police sent the hair to the laboratory to test for cocaine (along with other illicit drugs), the results were negative.

though modified, still exists today (Boyd 2003; Carney, 2007a,b,c). The law in this area recognizes quasi-property rights in the disposition of one's body through donation, but forbids the sale of organs (Rao, 2000; The National Organ Transplant Act 2005; Andrews and Nelkin, 1998a,b; Brotherton v. Cleveland, 4th Circuit 1991¹²). The body and its parts are now viewed by most as special gifts in accordance with the Kantian perspective, in which "things that are beyond price have dignity" (Kant, translated by Gregor 1998). Under this conceptualization we have a stake in the respectful treatment of body parts as per Human Tissue Gift Act legislation, The Uniform Anatomical Gift Act (1987) and other provincial and state statutes (Uniform Anatomical Gift Act, 1987; Canadian Council for Donation and Transplantation, 2003; Quebec: Civil Law of 1993 Articles 42, 43, 44). Renewable bodily products such as blood, semen and, more recently, ova have been characterized historically as commodities to be sold, but this characterization has been highly criticized (Andrews and Nelkin, 1998a, b). In the US there have been rulings on the ownership of cell lines generated from individuals. In the case of John Moore, for example, samples from his hairy cell cancer were removed and transformed into a cell line without his consent, then subsequently made commercially valuable. The case went to the Supreme Court of California where it ruled that the doctor was at fault for failing to inform Moore and getting false consent from him. However, the court did not find for Moore on the issue of conversion (i.e., the conversion of his cells into a pharmaceutical product), in which

¹² After Mr. Brotherton was found dead in a car, his body was taken to a hospital. His wife expressly forbade the hospital from harvesting her husbands organs, saying he had an aversion to such an anatomical gift. Because his death was a suspected suicide, his body was taken for examination by the coroner, who permitted the man's corneas to be removed. State law permits a coroner to remove the corneas of autopsy patients without consent, provided that the coroner had no knowledge of an objection by the decedent, the decedent's next of kin. Though the coroner was never informed of Mrs. Brotherton's views as the tissue harvesting, it was clearly stated on the report of death given to him. Further, it is the custom and practice of the coroner's office to not obtain a next of kin's consent or to inspect the medical records or hospital documents before removing corneas. The plaintiff brough suit under 42 U.S.C. § 1983, alleging that her husband's corneas were removed without due process of law and in violation of the equal protection clause. Her claim was dismissed by the trial court; she appealed to the Sixth Circuit which dismissed and reversed the claim. The Circuit court held that the trial court erred in dismissing wife's claim that her dead husband's corneas were taken under color of state law without due process of law because she had a legitimate claim of entitlement in his body, protected by the due process clause of the fourteenth amendment.

instance Moore claimed that the University of California and the doctor benefited from his property. The court noted that it could be detrimental to research if property status was granted to cells and body parts of the patients (*John Moore* v. *the Regents of the University of California*, 793 P.2d 479 Cal. 1990).

The deceased have been accorded quasi-property rights, "a category that encompasses the right to possession and the right to exclude, but not the right to transfer to others" (Rao 2000). The crucial question, however, is can a body or an organ be abandoned? It would seem that while alive it is impossible to abandon one's body or organs but what of eggs and sperm? Particularly those left with fertilization clinics (Shimron 2008). Further, it has been asserted that privacy is the only interest maintained in abandoned tissue (Levine 1986). How then should the body and its attendant DNA be legally conceptualized? Does the right to control our body extend to DNA? If, as has been stressed, the "body is central to human dignity," it seems odd that DNA, the blueprint for the body, would be accorded less respect and dignity than the body itself (Murray 1986). If there exists a right to control the body and its disposition then there are grounds for asserting that a similar interest exists in the DNA, whether within or separated, from the person, yet DNA is not conceptualized in the same way as the body (Hardcastle 2007). An additional analogy related to abandonment, which could be considered an extension of abandonment, is the characterization of DNA as waste.

4.4.4 *Waste*

Is abandoned DNA really akin to human waste? It is convenient to think about it in that way as it eliminates all Fourth Amendment privacy concerns. The holding in *California* v. *Greenwood* stipulates that once a person has placed garbage on the curb it becomes publicly available to all and there is no longer a reasonable expectation of privacy inherent in divested

items (486 U.S. 35 1988¹³). Courts have also tried to distinguish privacy interests between collecting human waste and the waste itself. In Skinner v. Railway Labor Executives Association the Court noted that the process of collecting and testing a urine sample implicated privacy interests and thus the Fourth Amendment (Skinner v. Railway Labor Executives' Association, 489 U.S. 1989¹⁴). This distinction was echoed in Vernonia Sch. Dist. 47J v. Acton (575 U.S. 646,658 1995¹⁵), another drug testing case. With respect to the waste itself, the Appellate Court in Venner v. State of Maryland "conceded that it was "not unknown for a person to exert a continuing right of ownership, dominion or control... over such things as excrement...or other parts of the body, whether their separation from the body is intentional, accidental or merely the result of normal body functions," while at the same time rejecting claims that the defendant possessed a reasonable claim to privacy in his excrement, stating that such things are abandoned in a legal sense "by the person from whom they emanate" (345 A.2d 498-498 Ct. Spec. App. Md. 1976). Further it has been asserted that "[t]he test for abandonment in the search and seizure context is distinct from the property law notion of abandonment: it is possible for a person to retain a property interest in an item, but nonetheless to relinquish his or her reasonable

¹³ In *California v. Greenwood*, for example, the Court held that that the defendants possessed no reasonable expectation of privacy in the trash bags they had left at the curb, which contained incriminating evidence of their narcotics trafficking. Even though police intercepted garbage intended for city collection, Greenwood could not complain because he had left his items in a place "particularly suited for public inspection." Anyone could delve through his abandoned trash.

¹⁴ After a review of accident investigation reports, from 1972 to 1983, the Federal Railroad Administration (FRA) found, "the nation's railroads experienced at least 21 significant train accidents involving alcohol or drug use as a probable cause or contributing factor," and that these accidents "resulted in 25 fatalities, 61 non-fatal injuries, and property damage estimated at \$19 million (approximately \$27 million in 1982 dollars)." 48 Fed. Reg. 30726 (1983). The FRA adopted safety standards including the administration of breath or urine tests or both to covered employees who violated certain safety rules. The Court of Appeals reversed the initial ruling, holding that a requirement of particularized suspicion was essential to a finding that toxicological testing of railroad employees was reasonable under the Fourth Amendment. The court stated that such a requirement would ensure that the tests, which reveal the presence of drug metabolites that may remain in the body for weeks following ingestion, are confined to the detection of current impairment. Moreover, although the collection and testing of privacy as to medical information and the act of urination that society has long recognized as reasonable. Even if the employer's antecedent interference with the employee's freedom of movement cannot be characterized as an independent Fourth Amendment seizure, any limitation on that freedom that is necessary to obtain the samples contemplated by the regulations must be considered in assessing the intrusiveness of the searches affected by the testing program, however, the compelling governmental interests served by the regulations outweigh employees' privacy concerns.

¹⁵ Veronia school district (District) adopted the Student Athlete Drug Policy, which authorizes random urinalysis drug testing of students who participate in its athletics programs, over concern that drug use increases the risk of sports-related injury. Mr. Acton was denied participation in his school's football program when he and his parents refused to consent to the testing. The Appeals court held that the testing was constitutional under the Fourth and Fourteenth Amendments (Pp. 652–666), that the testing was reasonable as student athletes have even less of a legitimate privacy expectation than the general population, for an element of communal undress is inherent in athletic participation, and athletes are subject to preseason physical exams and rules regulating their conduct.

expectation of privacy in the object. See, e.g., United States v. Colbert, 474 F.2d 174, 176 (5th Cir.1973)" (United States v. Thomas, 864 F.2d 845-846 D.C. Cir. 1989¹⁶).

Thus protection may well turn on what society recognizes as reasonable. "Custom may suggest that we intend to abandon human waste, but the assumption that we do so, and thus implicitly authorize DNA analysis on the same waste is hardly a widely accepted part of our social experience" (Joh, 2006; See, e.g., Abel v. United States, 362 U.S. 217, 241 (1960) finding that government collection of items left in hotel room wastepaper basket was permissible because to the defendant such items were "bona vacantia," or ownerless goods).

Now that we have established the different ways in which DNA may be characterized in the law it is time to examine the impact of DNA's legal standing on the ethics of acquisition and retention from various members of society.

4.5 Legal and Ethical Arguments Arising from Forensic Uses of DNA

4.5.1 Treating People as Means

In the instance of sample collection at the crime scene, commission of a crime by an individual bound by the social contract equals forfeiture of some rights, as the violator chooses to violate the rights of others. By failing to respect the rights of others, an offender can expect to have limits placed on rights such as liberty, privacy, and confidentiality, which are otherwise granted by society. As part of the tacit agreement of living in social groups, certain freedoms are ceded to allow for the expectation of a minimal level of group protection. Thus as part of the commitment to provide basic protection for the rights of all citizens, violation of the rights of others creates a duty to identify the culprit and use appropriate means to bring that person to

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^{..&}quot;Thomas intended for fourth amendment purposes to abandon the bag at the top of the stairs. His actions were in relevant respects similar to those of a person who tosses an object during police pursuit: in order to prevent discovery of his bag by the police, Thomas left it behind in a public place where he retained no reasonable expectation of privacy in it. 5 Cf. United States v. Collis, 766 F.2d 219 (6th Cir.) (per curiam), cert. denied, 474 U.S. 851, 106 S.Ct. 150, 88 L.Ed.2d 124 (1985) (defendant who threw a bag containing cocaine over a fence during pursuit by drug agents had "abandoned the shoulder bag and failed to establish a legitimate subjective expectation of privacy" in it)".

justice and protect society. In the case of DNA, one can argue that collection of a sample when possible is warranted. Though the terms of the social contract may not be explicit, one can argue that interest in materials, biological or not, left at a crime scene are forfeit as commission of a crime shifts the balance between freedom and security. Of course this does not take into account the possibility that there is shed DNA at the scene belonging to others that had a legitimate interest in being there and that they deserve protection in the security of their person. Though the presence of DNA may narrow the range of suspects and provide strong evidence it does not however change the notion of innocence until proven guilty and the requirement of prosecutors to prove their case.

When examining this scenario, it might at first seem as if we are treating people as means. In the Kantian conception, each of us has an interest in being treated with dignity and respect befitting persons not as mere means (Kant, translated by Wood 2002). Recognition of individual autonomy and respect for human dignity acknowledges that we are moral agents with preferences, values, commitments and a conception of "the good" and our legal system protects us from unwanted touching. Within the context of DNA collection or the collection of any bodily sample this would imply that individuals deserve protection from nonconsensual invasion of their bodies, and in most situations in our society (eg., research, medical treatment, or health insurance), they are protected by a requirement for consent and, in criminal law, through assault statutes.

It can be argued that framing the issue initially as a conflict between individual rights and society's protection is not illuminating, as the content and limits of individual rights cannot be determined prior to identifying and judiciously weighing all of the relevant morally legitimate interests at stake. Within the context of crime scene collection, however, utilizing this form of

evidence to identify the culprit is not equal to treating people as means. When we use this phrase it is designed to describe the use of people as objects or means to serve the ends of others without the consent or knowledge of the individual (Beauchamp and Childress, 2004). As part of the social contract we reject the notion of treating people as objects and instead enter into arrangements from positions of respect for the individual and their choices and engage them in the exchange of services. Furthermore, it would be difficult to argue that the culprit would be unaware of the potential for DNA collection to occur (Podlas, 2006a,b; Roane, 2005; Tyler, 2006). So in reality, both of the criteria for mitigating objectification--consent (which was given as assurance against defection and is thereby forfeit through the commission of a crime) and knowledge of the collection--are met and as a consequence none are treated as means. Further, the social contract, in securing the freedoms of the citizenry, obligates the agents of society to identify the culprit. Thus the collection of DNA from the crime scene violates no moral norms and assertions can be made that collection is obligatory under the social contract.

Alternately, it could be argued that once the perpetrator has abandoned articles at the crime scene, they are no longer extensions of the individual but in fact become objects. Using this logic, employing the abandoned articles for identification purposes is not a violation of moral principles. The problem with this assertion is that it assumes that because we shed DNA involuntarily, we no longer have an interest in it. This discussion will be developed further on in this chapter.

4.5.2 Curtailed Rights

Comparison of the convicted offender profile is likely to occur as soon as any crime scene profile is entered into the database. In this case, statutes often exist allowing DNA collection and the courts have ruled that, upon conviction of a serious crime, certain individual

rights are diminished or relinquished. Legally there have been two modes of thought with respect to sample collection without a warrant: diminished rights and probable cause exceptions. Within this legal framework it has been held that the rights of the convicted are "diminished" to the extent that their rights are fundamentally inconsistent with the needs and exigencies of the regime to which they have been lawfully committed (Hudson v. Palmer, 468 U.S. 517,525; 1984¹⁷; Wolff v. McDonnell 418 U.S. 539, 555-556; 1974¹⁸). The exception to the Fourth Amendment of the U.S. Constitution, regarding unreasonable search and seizure, is determined through a balance of intrusiveness versus the government's interests. The U.S. Court of Appeals for the Fourth Circuit held in Jones v. Murray that there is no "per se Fourth Amendment requirement" of individualized suspicion when the purpose is for ascertaining and recording the identity of legally confined inmates (Jones v. Murray, 962 F.2d 302 4th Cir. 1992). To be precise, the Court held that the acquisition of a DNA sample was a search requiring individualized suspicion. However, as there was no jurisprudence requiring probable cause when the purpose of the search was to identify prison inmates, to whit, no Fourth Amendment violation was committed (Jones v. Murray, 962 F.2d 305-6 4th Cir. 1992). The Court further noted that "probable cause had already supplied the basis for bringing the person within the criminal justice system" (Jones v. Murray, 962 F.2d 306 4th Cir. 1992)¹⁹. In dissent Judge

¹⁷ "[W]e [p526] hold that society is not prepared to recognize as legitimate any subjective expectation of privacy that a prisoner might have in his prison cell and that, accordingly, the Fourth Amendment proscription against unreasonable searches does not apply within the confines of the prison cell. The recognition of privacy rights for prisoners in their individual cells simply cannot be reconciled with the concept of incarceration and the needs and objectives of penal institutions. ... A right of privacy in traditional Fourth Amendment terms is fundamentally incompatible with the close and continual surveillance of inmates and their cells [p528] required to ensure institutional security and internal order" *Hudson* v. *Palmer*, 468 U.S. 517,525; 1984.

¹⁸ "But though his rights may be diminished by the needs and exigencies of the institutional environment, a prisoner is not wholly stripped of constitutional protections when he is imprisoned for crime. ... Of course, as we have indicated, the fact that prisoners retain rights under the Due Process Clause in no way implies that these rights are not subject to restrictions imposed by the nature of the regime to which they have been lawfully committed" *Wolff* v. *McDonnell* 418 U.S. 539, 555-556; 1974.
¹⁹ Probable cause for a single act is not a blanket probable cause for all acts. Further the specification of being within the "criminal justice system" leads one to think the record keeping function relates not just to the penal system but the whole of the criminal justice system. However, once the offender is released from the justice system (either prison or parole) there should be no further need to identify inmates as they are no longer within the penal system according the courts' rationale for retaining DNA identification.

Murnaghan argued that the interest of the government in banking samples from non-violent felons did not outweigh the intrusion upon an inmate who was no more likely to commit a violent crime in the future than a member of the general population (*Jones* v. *Murray*, 962 F.2d 308 4th Cir. 1992). A dissenting opinion by Judge Nelson, in *Rise* v. *Oregon*²⁰ contends that the statute for the Oregon DNA data bank should be held unconstitutional²¹. He argues that, because the only function of this statute is to aid in solving future crimes, there should be no exception to Fourth Amendment protection for prison inmates (*Rise* v. *Oregon*, 59 F.3d 1568 9th Cir. 1995²²).

4.5.3 Diminished Privacy

It has been asserted that once an individual has been convicted of a crime, expectation of privacy is diminished and this is clearly true during incarceration (Lippke 2002, *Hudson* v. *Palmer*, 468 U.S. 517, 525; 1984, *Wolff* v. *McDonnell* 18 U.S. 539, 555-556; 1974). From a social contract perspective, conviction of a crime implies a violation of the contract, a failure to respect others and by extension their choices or autonomy thereby forcing society to curtail the liberty, choices and actions of the individual. It could be argued that conviction implies the need

²⁰ Rise, Durham, Rhodes and Milligan brought a civil rights action against the State of Oregon and others, alleging that Chapter 669; a 1991 Oregon Law requiring persons convicted of murder, a sexual offense, or conspiracy or attempt to commit a sexual offense to submit a blood sample to the Oregon Department of Corrections for the creation of a DNA data bank; violates the Fourth Amendment's prohibition against unreasonable searches and seizures and constitutes an ex post facto punishment as applied to them because they were convicted prior to the law's enactment. They also maintained that the Due Process Clause required the State to provide a hearing before drawing blood pursuant to Chapter 669. Milligan also alleged that the State violated his right to due process by ordering him to submit a blood sample even though he had not been convicted of a predicate offense and by placing him in administrative segregation for refusing to comply.

²¹ "In reaching this conclusion, the majority brushes aside Supreme Court and Ninth Circuit precedent that recognizes invasion of the body as an intrusion of a scope fundamentally different from the capture of visual images or fingerprints, in which there is a minimal expectation of privacy because that information ordinarily is held out to the public. Because there is no justification for this unprecedented departure from settled Fourth Amendment jurisprudence, I respectfully dissent".

 $^{^{22}}$ "The majority's approach stands Fourth Amendment jurisprudence on its head by suggesting that statistical probabilities of future conduct can suffice in lieu of the probable cause requirement in the traditional law enforcement context. ... The majority does not and cannot rely on this "prison inmate" exception, because the sole purpose of the statute is to facilitate future criminal investigations. The statute allows authorities to conduct the blood test at any time, even immediately prior to the convict's release. See Or. Rev. Stat. S 137.076(2)(c). The creation of the data bank has nothing to do with prison administration; in fact, the Department of Corrections indicated at hearings on the proposed legislation that it neither supported nor opposed the bill, and its practice has been to obtain the sample only at the point that the inmate is to be released from its custody. Thus, the statute falls squarely under traditional law enforcement analysis requiring probable cause for invasions of bodily integrity".

for greater societal scrutiny of the individual upon release, as there has already been a failure to abide by the social contract.

In the arguments presented above, a balance between the special needs of society to track these individuals was weighed against the privacy or dignitary needs of the individual. A DNA profile, like the notion of fingerprints, provided reliable identification of the individual with minimal intrusion of their dignity and was not seen to curtail their choices and actions within the community. Further it is possible that the increased surveillance would act as a deterrent and boost the probability of identifying the perpetrator for repeat offences. Retention of the DNA itself raises other problems and is outside the scope of the current discussion.

The legal conception of privacy is explicitly defined in both the Unites States and Canada, it derives from several sources. Specific privacy rights have been granted through statutes such as The Privacy Act of 1974 and the Right to Financial Privacy Act of 1978, tort law, the amendments to the federal Constitution, the Bill of Rights in the U.S. and the Charter of Rights and Freedoms and The Personal Information Protection and Electronic Documents Act (PIPEDA) in Canada (http://laws.justice.gc.ca/en/P-8.6/text.html). However privacy is not explicitly mentioned in the amendments to Constitution but the First, Fourth and Fifth Amendments enumerate "privacy" guarantees along with the implied rights relating to privacy and autonomy from the specific guarantees of the Bill of Rights. The same is true of the Charter guarantees whereby the Second enumerates fundamental freedoms of religion, thought, expression, communication, assembly and association, Seven secures the right to life, liberty and security of the person and Eight affirms the right to be secure against unreasonable search and seizure.

4.5.4 DNA Retention for the Non-violent

As outlined, a reasonable argument can be made for retaining a DNA profile from violent and recidivistic offenders for identification purposes from a social contract perspective. It is less clear how to justify profile acquisition for individuals who have committed a non-violent or nonrecidivistic offence. These offenders have presumably served their sentence and paid their debt. Thus the post-incarceration period should see the individual theoretically restored to citizenship. Often as part of the conditions for release, these individuals are placed on parole for a specified period of time. This restriction to their freedom is considered both a security measure for society as well as a support mechanism for the parolee but is not indefinite (Abadinsky 2003). Insisting, however, that a DNA profile be retained from these individuals in perpetuity might violate the presumption of innocence underling the justice systems of most developed countries (Krawczak & Schmidtke, 1998). "Once an individual has been convicted of a crime there is not justification for implying them in future cases" (Krawczak & Schmidtke, 1998). Perpetual retention of a DNA profile is akin to continuing to implicate convicted offenders in the commission of future crimes. Though the intrusion may be minimal, it is arguably not balanced against society's desire to solve future crimes (see Judge Nelson, dissenting, in Rise v. Oregon, 59 F.3d 1568 9th Cir. 1995). Just as past bad acts are not admissible in court to substantiate current charges, DNA profiles should not be accessible without probable cause 23 .

Would the argument above justify intrusion for those convicted of civil disobedience? Would it be permissible to demand DNA from Rosa Parks? Civil disobedience within a social contract is a nonviolent act, performed in public, contrary to the existing law but with the intent to bring about change in policy or government law (Rawls 1988). The justification for civil

²³ A system could be set up whereby the profile was maintained, so as to reduce intrusion, but a showing of cause would be needed to access it and the individual notified of such a search. A cold case exception would be more problematic.

disobedience relies upon moral principles that define a conception of civil society and the public good and should serve as a warning that the conditions of social cooperation are not being honored. It may at first seem as if this is a digression, but it may help to clarify some of the issues underlying indiscriminate DNA sampling. To maintain a society we must have willing cooperation among free men and women. Under the social contract as defined by Rawls there are two principles of justice; first that we all have an "equal right to the most extensive liberty compatible with like liberty for all" and second that "social inequalities are to be arranged so that they are to everyone's advantage" (Rawls 1988). Thus once liberty is restored to a non-violent member of society, he or she should have the same standing as other members (to comport with the first principle of justice) or this imbalance must be justified under the second justice principle so as to be to everyone's advantage or at minimum to the benefit of the least advantaged member of society. If the subject's standing is fully restored, why should suspicion continue through the retention of their DNA profile? If the guiding principle is equal treatment for all, then why not have the profiles of all citizens available for search? If, on the other hand we do not believe that prior offenders are restored as full citizens, then are they to be equated with those who have committed violent offences?

4.5.5 *Liberty and Justice*

It has been argued that the liberty of the individual is not adversely affected by the retention of a DNA profile (CIHR 2000). The individual is still able to make all of the same choices related to liberty as any other member of society. If we accept that liberty or autonomy are not at issue, we must still determine if the inequalities are to everyone's advantage. The rationale for retaining profiles from perpetrators of crimes with a high recidivism rate, crimes likely to yield a biological sample and particularly heinous crimes is obvious and this practice is

advantageous to all. However it is difficult to see a principled justification, under this argument, for retaining the samples or profiles of non-violent offenders, the civilly disobedient and those who were sampled, but not charged. If the intention is to solve crime regardless of type, then the true limitation is our ability to collect and type a DNA sample at the crime scene. If society's goal were to solve all crime, then would it not be to everyone's advantage to have their profile registered and have trace DNA collected for all crimes? If on the other hand the goal of the criminal justice system is solely protection of its citizens and we acknowledge that crime will never be eradicated, then database expansion to include non-violent and exclusionary samples maybe a waste of valuable resources. Further concerns about the duration or perpetual retention of the DNA profile have been raised but are a secondary ethical issue to the just collection in that if collection is unjust it remains so regardless of the length of time it is retained.

4.5.6 Search and Seizure

In the event of a warranted seizure, great care is taken to ensure that the rights of the individual are protected against undue infringement. Only after probable cause has been shown is the justice system granted the authority to contravene the rights granted to the citizen by society (*Katz* v. *United States*, 389 U.S. 347,357, 1967 "searches conducted outside the judicial process, without prior approval by judge or magistrate, are per se unreasonable under the Fourth Amendment – subject only to a few specifically established and well-delineated exceptions"). The "reasonable expectation of privacy" test was articulated by Justice Harlan in *Katz v. United States*, providing the modern analytic structure and definition whereby a "search" has occurred when there is an actual expectation of privacy and one that "society is prepared to recognize as 'reasonable" (389 U.S. 347,357, 1967).

In this case, limits are or may be placed on the range of uses of the sample for comparison and the duration of sample and profile retention. As part of due process the utmost care is taken to secure legal and ethical treatment of suspects. However some have raised the question of whether using DNA evidence at all violates the protections against self-incrimination. *Schmerber* v. *California* held that blood is physical evidence and is not considered testimony deserving of Fifth Amendment protection [that which wards against self incrimination, through testimony, by refusing to compel a person to bear witness against oneself (*Schmerber* v. *California* 384 U.S. 757, 1966)²⁴]. Further examination of Fourth Amendment claims justified the collection of blood for analysis as there was reasonable belief that the delay necessary to obtain a warrant, under the circumstances, threatened the integrity of the evidence, as blood alcohol levels begin to drop after drinking and nearly two hours had elapsed since the accident.

The information contained within a DNA sample is immutable. The identification information contained within DNA will, therefore, not change with time and as a result there is little support for a "special needs" exception to the probable cause requirement based on an urgency to act. This being the case, it stands to reason that issuance of a warrant is most efficient for both law enforcement and the suspect. If the chief virtues of social institutions are justice and efficiency, then the assurance of use within the courtroom and the protection of individual rights via a warrant legitimizes the criminal justice system (Rawls 1988).

²⁴ Petitioner was hospitalized following an accident involving an automobile which he had apparently been driving. A police officer smelled liquor on petitioner's breath and noticed other symptoms of drunkenness at the accident scene and at the hospital, placed him under arrest, and informed him that he was entitled to counsel, that he could remain silent, and that anything he said would be used against him. At the officer's direction a physician took a blood sample from petitioner despite his refusal on advice of counsel to consent thereto. A report of the chemical analysis of the blood, which indicated intoxication, was admitted in evidence over objection at petitioner's trial for driving while intoxicated. Petitioner was convicted and the conviction was affirmed by the appellate court which rejected his claims of denial of due process, of his privilege against self-incrimination, of his right to counsel, and of his right not to be subjected to unreasonable searches and seizures.

Collecting DNA upon arrest is an extension of the post-conviction collection statutes but offers little protections for the average citizen. If anything, it is an invitation for false arrest and discriminatory profiling. Where arrestee testing has been approved by local law makers, they assert that the community is in such need of protection that the arrest of an individual, even for a misdemeanor, is enough to contravene their rights (Simoncelli and Steinhardt 2006). All of the arguments that pertain for post-conviction testing pertain doubly for arrestee testing. Unlike convicted offenders there is no diminished privacy exception. Does collecting DNA from arrestees constitute a search under the Fourth Amendment? To trigger Fourth Amendment protection, a search or seizure must be established to have occurred. For a particular intrusion to constitute a search, the target of the intrusion must have a subjective expectation of privacy in the information obtained by the government and that the expectation is "objectively reasonable" (California v. Greenwood, 486 U.S. 35, 40; 1988). Three factors considered in determining a reasonable expectation of privacy are the extent to which DNA is exposed to the public, the extent of bodily intrusion and the nature of the information obtained (Kaye 2001). In general information and property that is exposed to the public is not considered to have Fourth Amendment protection. Fingerprints, which are left on every item that we touch, are not considered protected under the Fourth Amendment because they are constantly displayed to the public (United States v. Dionisio, 410 U.S. 1, 1973; Mills, 686 F.2d 135,139 3d Cir. 1982; Coddington v. Evanko, WL 2416429 3d Cir. 2004). This rationale can be extended to DNA as we shed it throughout our daily activities and thus acquisition would not constitute a search. However, it is unlikely that the courts will apply the rationale so broadly as to include DNA, as it can potentially reveal a broad array of personal information and requires the use of specialized technology not publicly available (Maclin, 2006). Physical intrusion into the body constitutes a

search under the Fourth Amendment. The taking of blood (*Schmerber* v. *California*; a reasonable search under "special needs"), breath and urine samples (*Skinner* v. *Railway Labor Executives' Association*; involved federally mandated drug testing of railway workers implicated in accidents or violations of safety rules), have all been deemed searches. In Canada similar rulings on Charter protections have occurred, *R. v. Dyment*, 2 S.C.R. 417, 1988 at p. 428. Though DNA sampling can be accomplished through minimal invasion it is possible that it will be considered a search in the same vein as *Schmerber*, *Skinner and Dyment*).

4.5.7 Discrimination and Universality

Throughout the U.S. criminal justice system there is a disturbing element of racial disparity (Duster 2003). New Jersey state police memoranda, reports and other papers documented that the state police engaged in racial profiling over a period of ten years and the problem was so pervasive that the state entered into a consent decree with the Department of Justice that barred police officers from relying "on the race or national or ethnic origin of motorists in selecting vehicles for traffic stops... except where state troopers are on the look-out for a specific suspect ..." (McAlpin, 2000, *United States* v. *New Jersey*, 1999). A 1995 survey of Maryland police interstate stops reported that while African Americans made up only 14 percent of the people driving along the interstate they constituted 73 percent of the cars stopped and searched. Though the arrest rates were roughly equivalent, the biased stop and search criteria led to a disproportionate number of minority arrests (Higgins 1997; Duster 2004).

Canada is not immune from racial disparity within the criminal justice system, but this discrimination has tended to focus on Aboriginal Canadians and blacks (Mosher, 1998; Roberts and Doob 1997). Aboriginal Canadians and blacks are over-represented in federal penitentiaries and though the provincial jail population varies from province to province, and so does the

definition of Aboriginal, the rough estimates have Aboriginal Canadians vastly over-represented in prison admissions with the same being so for black inmates in those provinces that collect information on visible minorities (Roberts and Doob 1997). The disparity with respect to Aboriginal representation can in part be a reflection of the differences in the way justice is administered by police, the courts and correctional institutions (Griffiths and Verdun-Jones 1994; LaPrairie 1990). A summary of royal commissions and studies on police and Aboriginal peoples concluded that the likelihood of conflict and high arrest rate was increased by the mutual hostility and distrust that characterized the interaction between the two groups (Griffiths and Verdun-Jones 1994). With respect to blacks accused in Canada, limited information is available country-wide, but a large-scale study by the commission on Systematic Racism in the Ontario Criminal Justice System provides data indicating that racial disparity exists at two of three bail release stages: the police and police station but not at the showing of cause (Commission on Systematic Racism in the Ontario Criminal Justice System, 1994a, 1995; Griffiths and Verdun-Jones 1994). Further there is also a statistically significant racial bias in sentencing independent of the bail decision (Griffiths and Verdun-Jones 1994).

Taking into account the disparity in treatment of minority groups within the U.S. and Canadian criminal justice systems, not to mention the groups targeted post 9/11, this leads to concerns of unjust and unfounded over-representation of these groups within the forensic DNA databanks. Another concern is that stratifying citizens based on past criminal acts or even the suspicion of criminal behavior (arrestees) will create social and legal inequities exacerbating class distinctions (Simoncelli 2006).

4.5.8 Consent and Coercion

The consent obtained during DNA dragnets, as described above, has been widely criticized as being neither informed nor free from coercion (Cho 2004; ACLU 2003; Halbfinger 2003; Wilson 2002). Individuals who refuse are instantly suspects and many of those who contribute a sample are under the impression that their profile will be compared solely with the incident crime scene profile and that their sample and profile will subsequently be destroyed. The premise of a DNA sweep is that the community will aid the police in identifying the perpetrator of a crime in which a biological sample was retrieved. Sweeps most often occur when DNA is the main or only evidence, and the target community of which the suspect is a member is relatively small. This type of DNA collection raises several issues, the first of which is the ability of a citizen to give truly free and informed consent to an officer of the law. It is naive to believe that the police are not intimidating, particularly when they are on your doorstep asking for a buccal swab. It has been argued that it is the procedure that is intimidating and not the police themselves, but the scraping of the inside of the cheek with a cotton swab is only moderately invasive. Of concern is the unequal power differential between the police and the citizenry and the possibility that the individual does not know that he can legally refuse to donate a sample. This power differential can be in itself a form of coercion.

It has been suggested that failure to provide a sample automatically makes the citizen suspect (American Civil Liberties Union 2003; Halbfinger 2003; Wilson 2003). If this is the case then refusing to volunteer a sample is an invitation for further intrusion by the police and thus the citizen is forced to weigh the balance of harms imposed by this approach to culprit identification. In addition to the questionably free nature of consent are concerns regarding the quality and specificity of the information given to obtain consent. Often individuals are not

informed that their sample may be retained and that its use is not restricted to the case currently under investigation. In many cases the consent is not a signed written document. How is it that the voluntary submission of a DNA sample should be subject to fewer rights protections than samples obtained under probable cause?

4.6 Ethical and Legal Dimensions for Surreptitiously Acquired DNA

Throughout this section I have posed ethical questions which arise as a consequence of various methods of acquiring DNA. Each deserves more attention from a critical ethical perspective. However, the focus of the next section will be on the most troubling and least addressed method: discarded or "abandoned" DNA acquisition.

As any television viewer can report, DNA, in a quantity sufficient to yield a profile, can now be obtained from discarded items such as used coffee cups, tissue, chewing gum and in some cases even fingerprints (Balogh et al. 2003). The potential suspect is presumably unaware that a biological sample has been collected, and that a profile can be compared with the crime scene profile (Halbfinger, 2003). As noted above, both the sample and the profile may be retained indefinitely, often with the donor being completely ignorant of the collection.

4.6.1 Defining Surreptitious DNA Acquisition

"Abandoned" DNA is defined by Joh as "any amount of human tissue capable of DNA analysis and separated from an individual's person inadvertently or involuntarily, but not by police coercion" (Joh, 2006). This terminology was specifically selected to emphasize the difference in intent between discarded DNA, tumors or diseased organs, and that DNA which is shed uncontrollably as a by-product of life. As we conduct our lives and interact with our environment, DNA is left inadvertently in trace amounts from hair and skin cells on the items we touch, when we talk or sneeze and as we walk. There is no interaction in which we don't leave a

trace of ourselves. This is the foundation for Locard's principle of exchange and forensic science's quest to collect, identify and individuate (Locard 1920, 1928, 1930a,b,c). As humans we cannot control the distribution of our shed particles, but legally that does not mean that we have no interest in that material.

In 2002, posing as a fictitious law firm, cold case detectives sent Mr. John Athan a letter inviting him to join a fictitious class action lawsuit concerning parking tickets. The letterhead contained the names of the "attorneys," all of whom were employed by the Seattle Police Department. Believing the letter to be true, Mr. Athan signed, dated, and returned the enclosed class action authorization form. Athan's reply was received by detectives and forwarded to the crime lab unopened. The lab obtained a DNA profile from saliva located on the flap. The DNA profile from the envelope matched a DNA profile from semen left on the body of 13-year-old Kristen Sumstad who was found inside a cardboard box, in the Magnolia neighborhood of Seattle, on November 12, 1982. Based primarily on the results of the DNA testing, the prosecuting attorney filed an information and probable cause statement to secure an arrest warrant for Mr. Athan.

The two detectives went to New Jersey, after obtaining the warrant, to arrest Athan. Pursuant to the warrant, detectives obtained a second DNA sample from Mr. Athan which matched the sample from the envelope and from Sumstad's body.

Following several pretrial motions to suppress evidence and a request for dismissal, all of which were denied, in 2004, Athan was found guilty of second degree murder and sentenced to 10 to 20 years under pre-sentencing reform act guidelines.

Mr. Athan appealed the decision to the Supreme Court of Washington in 2007. He argued that "his DNA (deoxyribonucleic acid) was collected in violation of both the United States and

Washington State Constitutions", that the actions of the detectives were illegal and unfairly prejudiced his right to a fair trial, requiring dismissal of the case under CrR 8.3(b). In the alternative, Athan argued the trial court erred in several evidentiary rulings and asked the court to remand for a new trial with instructions to exclude certain evidence (*State* v. *Athan*, 158 P.3d 27 (Wash. 2007)). The conviction was affirmed. The Court held that the collection of Mr. Athan's DNA did not violate the state or federal constitution.

Athan argued that his case involved three matters that are "private affairs"²⁵ under Washington law: one's body and bodily functions; communications with a person one believes is an attorney; and sealed correspondence intended for one's attorney. The State maintained, Athan voluntarily relinquished his DNA when he licked the envelope and mailed it to a third party and that DNA obtained from one's saliva is akin to a person's physical description, appearance, or other characteristic voluntarily exposed to the public, thus, it is not a "private affair" at all (See, e.g., State v. Carter, 151 Wn.2d 118, 126, 85 P.3d 887 (2004)). The court asserted that voluntary exposure to the public was relevant to the inquiry and could negate an asserted privacy interest (State v. McKinney, 148 Wn.2d 20, 29, 60 P.3d 46 (2002)). As such the court held that there was no inherent privacy interest in saliva, voluntarily placed on an envelope, and that the envelope, and any saliva contained on it, became the property of the recipient. The court implied that had the state used the DNA for any other purpose than identification it may have implicated a privacy interest but as the state in this case used Athan's DNA for identification purposes only, no personal information was revealed. With regards to the Fourth Amendment analysis, of the question of whether a person retains a reasonable expectation of privacy in their saliva after they

²⁵ The term "private affairs" generally means "those privacy interests which citizens of this state have held, and should be entitled to hold, safe from governmental trespass." State v. Myrick, 102 Wn.2d 506, 511, 688 P.2d 151 (1984).

lick an envelope and place it in the mail, the court expressed "[t]he analysis of DNA obtained without forcible compulsion and analyzed by the government for comparison to evidence found at a crime scene is not a search under the Fourth Amendment" (*State* v. *Athan*, 158 P.3d 27 (Wash. 2007)). The court noted that there have been no privacy challenges on the practice by police of surreptitious collection of DNA, fingerprints and footprints as evidence left in public places and as such "[t]here is no subjective expectation of privacy in discarded genetic material" (*State* v. *Athan*, 158 P.3d 27 (Wash. 2007)).

Jurisprudence has characterized DNA left on objects as "discarded" or "abandoned" DNA (*State* v. *Athan*, 158 P.3d 27 (Wash. 2007; *State* v. *Wickline*, 232 N.W. 2d 249, 253 (Neb 1989); *State* v. *Buckman*, 613 N.W. 2d 463, 474 (Neb 2000). The concept of discarded or abandoned DNA should be examined more closely. "The rules of criminal procedure appear to pose no restrictions on the police when collecting this evidence. Not only does "abandonment" affect police behavior, it raises basic questions about the changing nature of identity in the genetic age" (Joh, 2006).

4.6.1.1 Intent

It can be posited however that the argument could instead turn on the intention of the individual to discard an item. "To determine whether there has been abandonment in the Fourth Amendment sense the…court must focus on the intent of the person who is alleged to have abandoned the place or object" (*United States* v. *Thomas*, 864 F.2d 843,846 D.C. Cir 1989; *United States* v. *Anderson*, 663 F.2d 934, 938, 9th Cir.1981). How do we distinguish between intentionally discarded DNA and that which is shed, but in which we still retain an interest? We intentionally give blood to physicians for tests and discard tumors, but we do not intend to shed our DNA through skin cells and saliva. It may seem as though there is no distinction in terms of

the privacy interest at stake, but a slightly simpler analogy may illuminate the incongruity. Let us suppose that we are in the museum and our wallet slips out and is "abandoned" or lost. We report its loss to the management. Do we still have an interest in the wallet and its contents? If someone finds the wallet is it theirs to do with as they choose? Are the credit and identification cards theirs to use as they please? Even if you argue that the cards did not actually belong to the holder, but rather to the company and or the government of issue, all parties still have an interest in their rightful, continued use. The wallet was not intentionally abandoned, and one may still press a claim for the contents. It is arguably the same with shed DNA. One does not intend to shed it, but still has an interest in the information contained within. In criminal law, intention mitigates punishment and therefore the concept has some legal standing. [If an individual with HIV intentionally spits on someone they are punishable in criminal court as well as liable for damages incurred. (Brock v. State, 55S.2d 285 (Al Crim APP. 1989); Weeks v. Scott, C05.317 U.S. Fifth Circuit, Court of Appeals, No. 94-20838; June 23, 1995)]. As discussed previously, the DNA shed during the commission of a crime would be forfeit for identification purposes, but those individuals who are not suspect or those who might be suspect, but for whom evidence is lacking, should be secure in their daily lives. Thus the intention to discard or not might be more useful in legal reasoning than just equating DNA as property with interests, privacy or other, restricted to possession.

4.6.1.2 Invasion of Privacy without Intrusion

The U.S. Supreme Court has started to recognize that as a result of new technologies, privacy can be effectively invaded without physical intrusion by simply collecting what has ostensibly been discarded. The Supreme Court noted that, by recording the heat that emanated from a home, the police were in effect performing a search under the Fourth Amendment and

thus required a warrant. The collection entailed no physical entry onto the premises, just the recording of "discarded heat", equivalent to waste, but the pattern of heat revealed private information regarding the home and this private information deserved constitutional protection (*Kyllo* v. *U. S.* 533 U.S. 27, 37; 2001).

In another recent case, the U.S. Supreme Court held that performing an additional test on urine voluntarily surrendered to medical personnel to obtain other information constituted a Fourth Amendment search. In Canada *R. v. Dyment*, 2 S.C.R. 417, 1988 a blood sample taken by a physician, while the patient was unconscious, and later turned over to a Royal Canadian Mounted Police (RCMP) officer was ruled a Charter violation under section 8. A similar garbage analogy can be made in this case for the urine that was eliminated and disseminated to third parties (*Ferguson v. City of Charleston* 532 U.S. 67, 84, 121 S. Ct. 1281, 149 L. Ed. 2d 205 (2001).²⁶ Similarly, Justice Fairhurst's dissent in *State* v. *Athan*, "the testing of Athan's saliva went beyond enhancement of natural senses and allowed law enforcement to observe Athan's DNA without his knowledge" (*State* v. *Athan*, 158 P.3d 27 (Wash. 2007)).

Shelly Colb asserts that the Supreme Court "has come to see that however easy it is to collect something (and however available that thing is for collection out in the world), the fact that private information may be gleaned from it imbues the collection itself with constitutional significance" (Colb, 2004).

²⁶ Officers obtained consensually extracted bodily fluids of pregnant patients suspected of drug abuse from hospital staff without the patients' knowledge. The United States Supreme Court explicitly rejected the practice and concluded the patients did not provide the body fluids voluntarily, reasoning that patients would not expect hospital staff to provide incriminating evidence against them even if the staff were required by law to report incriminating conduct. Id. at 78 n.13.

The whorls and ridges of fingerprints tell us little about the "private life" of the person they belong to and the same is true, at this moment in time, of a forensic DNA profile. The reality, though, is that normative practice is to retain not only the profile, but also the DNA molecules from which the profile was derived. This opens the possibility that many more genes, including those which might reveal aspects of "private life" can potentially be examined. Keep in mind that many of these samples are obtained from citizens who may be suspected of committing a crime, but who should nonetheless be presumed to be innocent. It is thus the retained DNA molecules, which has the potential to violate privacy and which would thus appear to qualify for Fourth Amendment protection. Contrary to this argument, to date Fourth Amendment rejections of post conviction testing have centered on the invasiveness of the search and the limited expectation of privacy accorded to convicted offenders (United States v. Kincade, No. 02-50380 (9th Cir. 2003)). This ruling has subsequently been vacated and 9th Circuit allows DNA testing of persons on parole with the holding relying "on a totality of the circumstances analysis to uphold compulsory DNA profiling of convicted offenders both comports with the Supreme Court's recent precedents and resolves this appeal in concert with the requirements of the Fourth Amendment" U.S. v. Kincade, 379 F.3d 813(9th Cir. 2004)). As of 2005, Wisconsin has the only state legislation requiring sample destruction after DNA analysis (Axelrad, 2005).

4.7 Ethical Issues Specific to the Forensic Use of Surreptitiously Acquired DNA

4.7.1 Autonomy and Consent

Under the Kantian conception of human dignity all persons have unconditional worth and are to be viewed as autonomous in accordance with their capacity to determine their destinies (Kant, translated by Wood 2002; Kant, translated by Gregor 1998). Though autonomy has a

myriad of definitions, the key components of autonomy for the purposes of this discussion are liberty and agency (Beauchamp and Childress 2004). Agency is understood to be the capacity for intentional action, while liberty refers to the freedom from influence and constraints. At the very least, respect for an autonomous agent grants or recognizes that person's right to hold views, to make choices, and to take actions based on personal ideals and values (Beauchamp and Childress 2004). Our decisions or choices then are paramount not only to our political lives, but also to our bodily, spiritual and psychological integrity. The right to choose is enshrined in the Constitution, the Bill of Rights and the Charter of Rights and Freedoms. The First Amendment of the U.S. Constitution, the right of freedom of speech, press and association protects expression of opinion and the right of belief in every facet of an individual's life (Pierce v. Society of Sisters 268 U.S. 510, (1925): the right to educate one's children as one chooses; 1st and 4th Amend; Meyer v. Nebraska 262 U.S. 390, (1923): the right to study the German language in private school; NAACP v. Alabama, 357 U.S. 449, 462, (1958): freedom to associate and privacy in one's associations; Griswold v. Connecticut, 381 U.S. 479, (1965): contraception for married couples). Though these rights have not been enumerated, the First Amendment has been construed to include them as necessary in making the express guarantees fully meaningful. "In other words, the First Amendment has a penumbra where privacy is protected from governmental intrusion" (Griswold v. Connecticut). The Canadian Charter of Rights and Freedoms, Section Two, covers the same expressed rights as the First Amendment. However the language provisions are enumerated in Charter Sections Sixteen through Twenty-three. Governmental intrusion is protected against by the Second, Seventh and Eighth Sections of the Charter (Canadian Charter of Rights and Freedom 1989).

Autonomy, liberty and agency are at the core of the liberal political framework and these concepts are expressed explicitly in the Canadian Constitution and Charter. Respect for autonomy with the correlative right to choose allows us to accept or reject, to authorize or decline access, to give or withhold consent. These choices, in our society, translate to consent either expressed or tacit for political, healthcare, education, justice and other systems. Consent limits what we should be subjected to on a daily basis. This right to choose implies more than a respectful attitude; it must be implemented through action.

4.7.2 Abrogated Consent

There are negative and positive obligations associated with autonomy. The negative obligation demands that actions should not be subjected to controlling constraints by others while the positive obligation "requires respectful treatment in disclosing information and fostering autonomous decision making" (Beauchamp and Childress 2004). Canvassing the neighborhood asking citizens for a DNA sample provides the individual with the opportunity to authorize or decline sample collection in compliance with respect for autonomy, provided the consent process is truly informed and free from undue influence or pressure. But as described above this is unlikely under the way sweeps are currently conducted (Chapin, 2004).

Seizing a DNA sample in a surreptitious manner ignores and demeans another's right to autonomy. It meets neither the negative nor positive obligations of autonomy. The individual is unaware and uninformed that their DNA has been collected. Agency is diminished as one has little control of shed DNA, regardless of his or her intentions, and no influence on its subsequent use. It may be argued that liberty is unaffected because the individual doesn't know that his/her DNA has been "liberated" and is therefore unlikely to act differently or be influenced or constrained by the knowledge (Kaye 2003). However the longer this practice is continued the

higher the probability that it will impact every citizen, as the demonstration of probable cause will be obsolete and citizens will no longer be free from unreasonable search and seizure. As Justice Brandeis' dissent in *Olmstead* v. *United States* asserts,

"The makers of our Constitution undertook to secure conditions favorable to the pursuit of happiness.... They sought to protect Americans in their beliefs, their thoughts, their emotions and sensations. They conferred, as against the Government, the right to be let alone - the most comprehensive of rights and the right most valued by civilized men. To protect that right, every unjustifiable intrusion by the Government upon the privacy of the individual, whatever the means employed, must be deemed a violation of the Fourth Amendment" (277 U.S. 438, 1928).

The sense that government agents have uncontrolled power to create suspects of its citizens without proven increases in security is enough to make anyone a bit paranoid, resulting in limited expression of liberty (Chapin 2004; Walker 2004; Kaye 2006). "Nonetheless, while subjecting persons who have been convicted of a crime to inclusion in a DNA database is inherently problematic, subjecting those who have never been convicted of a crime subverts our notion of a free and autonomous society and is characteristic of an authoritarian regime" (Simoncelli, 2006). There is further risk of unequal representation of samples from various minorities exacerbating racial biases and disparity within the justice system (Simonelli and Steinhardt, 2006). If, in an appeal to fairness, all citizenry were included in a national DNA facility, informed of inclusion and had influence on the security and usage of the DNA, would we still feel compelled to constrain our behavior?

4.7.3 Are There Circumstances in which the Public Good Outweighs the Damage to Autonomy?

Respect for autonomy and choice is not absolute. Competing moral considerations may constrain autonomous action and in some situations, individual freedom to consent is outweighed by public interests. During epidemics, the requirement to consent to treatment has been overridden on the basis of public health priority. These cases are however emergency measures

and the right to autonomous choice is restored to the individual once the crisis is over and eminent danger to others is abated. Vaccinating children is mandated as a public health initiative by public school systems that require immunizations for school attendance. Newborn screening on the other hand is a public health measure designed to identify potential harms, but it is not an emergency measure. Consent for the screening is considered to be implied with the provision of other health services and one must opt out from such measures (The National Conference of State Legislatures 2002). Although the primary harm when opting out of newborn screening is to the individual being screened, there is a secondary harm to society, as it is responsible for care of these individuals whose life outcome is impaired by failure to screen and treat in a timely fashion.

With respect to public good versus autonomy, what would be the measure of the public good achieved by seizing DNA in the absence of consent? How would we evaluate it or what criteria would we use? If crime solution and public protection are considered to be the public good, then proof of the effectiveness of DNA profiling in perpetrator detection might support the need to infringe autonomy. Could crime deterrence or prevention be considered a measure of public good (Bieber 2006)? "In preventive collection, one assembles the DNA of a large number of people without the claim that any of them did anything illegal" (Etzioni 2006). Would assembling DNA have an impact on the deterrence and prevention of crime? For prevention to be effective the entrants must know that their DNA has been collected and is accessible by law enforcement. In considering this position it is not unreasonable to argue for preventive collection without any individual suspicion however consent is paramount to brace community support. Surreptitious sampling would be contrary to the ends of deterrence and prevention and reinforce fears of the public supports.

4.7.4 Higher value in crime solution vs. autonomy infringement

What would be required to shift the balance from privacy to security or from autonomy to the public good within the criminal justice setting? Legally the state can override the more general privacy or liberty interest in controlling one's body when doing so bears a rational relationship to a legitimate state interest: fingerprinting suspects, ordering blood tests for paternity cases, detention pending psychological evaluations and identifying prison inmates through DNA. These all interfere with the right to control one's body as one sees fit. We have already covered the post conviction testing exception where it was determined that DNA acquisition did not require probable cause because the purpose of the search was for inmate identification and furthermore, probable cause had already been established for their conviction (*Jones* v. *Murray*, 962 F.2d 308 4th Cir. 1992). "Except in well-defined circumstances, a search or seizure is not reasonable unless it is accomplished pursuant to a judicial warrant issued upon probable cause" (*Skinner* v. *Railway Labor Executives' Association*, 489 U.S. at 619, 1989). Therefore in cases where a warrant can be supported there is a legitimate or reasonable state interest.

The "special needs" exception to the probable cause requirement permits government officials to conduct searches and seizures without individualized suspicion of criminality, negligence or malfeasance. Does the surreptitious acquisition of DNA fall under the "special needs' purview? Some Courts have recognized situations where "special needs, beyond the normal need for law enforcement, make the warrant and probable cause requirement impracticable". See, for example, *New Jersey* v. *T.L.O.*, 469 U.S. 325,351 (1985). This in essence provides greater flexibility to law enforcement. Justice Blackmun, in his dissent, to *Ortega* explained that "only when the practical realities of a particular situation suggest that a

government official cannot obtain a warrant based upon probable cause without sacrificing the ultimate goals to which a search would contribute, does the Court turn to a 'balancing' test to formulate a standard of reasonableness for this context" (O'Connor v. Ortega 480 U.S. 732, 1987). Unfortunately it is unclear from the conflicting case law and inconsistent logic what the precise components of the special needs analyses are and where the line between criminal law enforcement and special needs is drawn. However as DNA from a specific individual will not change over time, with the possible exception of bone marrow recipients, it is difficult to apply the "special needs" exception to DNA seizure from a mere suspect. "Regardless of whether a DNA bank should be considered beyond the general needs of law enforcement the proposition that the government's "special needs" outweigh the privacy interests of innocent persons seems beyond the pale, as a matter of Constitutional principle" (Colb, 2004). In a situation where a possible suspect has refused to give a DNA sample for exclusionary purposes, and the law enforcement agency does not have enough evidence for a probable cause warrant what would be required to infringe autonomy? Could the seriousness of the offense be enough to tip the scale? One might propose that murder, torture, rape and pedophilia are serious enough offenses to mitigate autonomy interests in favor of security based on the initial reasoning for post conviction DNA profiling.

Does the benefit from DNA profiling with respect to crime control and prevention warrant the infringement? What proof or what degree of proof would be necessary to demonstrate that this is an effective method of crime control and prevention?

An examination of the success of the National DNA Data Bank of Canada (NDDB) in its first two years may provide some assistance. The NDDB made over 452 matches between crime scenes and convicted offender samples, and more than 29 crime scene to crime scene matches.

In its second year of operation, the total number of hits increased almost ten-fold (Charron 2003). The NDDB has received only a fraction of the eligible samples (less than 50 per cent of 18,600 primary offence samples and less than 10 per cent of the 94,500 secondary offence samples). Thus the figures could be more robust.

If however we examine the success of DNA sweeps where suspicionless individuals are targeted the evidence thus far does not support such measures. A United States survey of all the reported cases, since 1990, of police requesting voluntary DNA samples from potential suspects found that the tests successfully identified an offender in only one of eighteen cases (Walker 2004). "This suggests that DNA sweeps are extremely unproductive in identifying criminal suspects" (Walker 2004). Unfortunately there is no measure of crime prevention.

Privacy, confidentiality and autonomy are at the heart of this issue.

4.7.5 Privacy and Confidentiality

Autonomy or choice as discussed above is not an enumerated Constitutional right. Instead it is believed to emanate from the Bill of Rights and the Charter of Rights and Freedoms (*Griswold* v. *Connecticut*, 1965; *R.* v. *Dyment*, 1988; *Aubry* v. *Éditions Vice-Versa Inc.*, 1998; Benn 1971, 1988). Within a legal concept autonomy has been considered with privacy (*Dagg* v. *Canada* (*Minister of Finance*), 1997). Privacy in general is a complex, multifaceted concept and has at minimum three dimensions: physical, informational and decisional (Allen 1996). Conceptually it involves several different but overlapping personal interests and encompasses informational privacy, relational privacy, decisional privacy and physical privacy (Beauchamp and Childress 2004; Annas 2004). Informational privacy concerns control over highly personal information about ourselves while relational privacy is that which allows us to determine with whom we have personal, intimate relationships. Decisional privacy is the freedom from

surveillance and the influence of others when making personal decisions and choices (Annas 2004). Physical privacy concerns the ability to exclude others from our personal things and places. Confidentiality concerns the safeguarding of these private aspects protecting them from disclosure without our authorization. Autonomy has its place within each aspect of privacy in our exercise of choice, liberty and agency to execute those choices further it is a necessary component of the pubic interest. "Private interests are offered support through the criminal law but only where the protection of those interests is itself a matter of public interest. At the forefront of such interests is that of individual autonomy" (Wilson 2002). The pursuit of criminals must be balanced by the interests of the individuals within society as well as society as a whole. Thus an attack on the core values, privacy and autonomy, of a single individual is in essence an attack on everyone both individually and collectively. "To be an autonomous person in the sense of being the effective author of our own lives requires our ownership and possession of property to be protected from interference and our personal security to be guaranteed. An efficient, ordered society must necessarily underwrite the interests of individuals in the security of their person and property" (Wilson 2002).

The acquisition of a person's DNA without his/her knowledge to be used for the state's interests does not support the personal security necessary to stabilize social order. The lack of transparency threatens the legitimacy of the state's interest. Too many governments have run rough shod over civil liberties and individual rights to the detriment of their citizens in the name of the "common good". Clandestine sampling is akin to an Orwellian pursuit of law and order rather than justice.

Recognizing the advances in DNA technology are creating new opportunities for identifying criminal suspects and exonerating convicted offenders, there have been

recommendations that the law enforcement profession, in consultation with community groups and legal experts, develop a model policy and procedures to guide in the collection and use of DNA evidence (Walker 2004; Scheck and Neufeld 2006). This is not to say, that after public debate, the collective might not decide that a universal forensic DNA bank is in its best interests.

For all of these reasons it is essential that we open the debate now and act as a community to evaluate the proper course of action through public consultation.

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CHAPTER 5: CALL FOR PUBLIC CONSULTATION

5.1 Introduction

The incidence of violent crime has declined in both Canada and the United States during the period from 2002 to 2006, but many people still don't feel safe (US Department of Justice 2007; Statistics Canada 2007; Saad, 2007). Maybe this is a result of terrorist attacks around the world [47% of Americans are worried or very worried that they or a family member will become a victim of terrorism (Gallup, 2007)] or maybe the public is more pessimistic about crime as a consequence of the media attention devoted to it (Sadd 2007). While terrorism is quite different from local crime, many of the same issues of security are shared. We collectively realized how vulnerable we are to violence and how important physical security is. How we as a citizenry choose to protect ourselves is paramount precisely because we can easily destabilize democracy and the social order by failing to underwrite individual interests in the sanctity of person and property, thereby undermining the foundations upon which the social contract rests.

In the preceding chapter I discussed the importance of the social contract and the need for transparency by government toward the people it serves. In order to fulfill this requirement it is crucial for members of society to have influence on policy and give direction to those who represent them. In the context of this thesis, the important question is thus "How do we maximize the utility of DNA as a source of evidence and still protect privacy?"

Throughout history societies have grappled with the balance of security and freedom, privacy and public exposure. The society in which we live is stratified by wealth, education, power, health and criminal status. Citizens are stratified in so many ways that it is not surprising that the criminal justice system is similarly ordered. We distinguish misdemeanor crimes from assault and even graduate assault into varying degrees. That being said, DNA collections reflect

these degrees of difference as outlined in the previous chapter. DNA has the power to distinguish between individuals regardless of status and is accessible and retrievable from more and more sources. Thus it might be considered a unifying or equalizing instrument. Should we continue to stratify citizens for DNA collection by their criminal deeds or should all citizens be required to submit DNA? The problem is, as we broaden our criteria for DNA collection over more groups, how do we protect our citizens from undue intrusion and apply those criteria in a just manner?

Incursions have been made, either through dragnets or surreptitious sampling, into the lives of persons which I have argued infringes their rights and sanctity of person. This should raise concerns from all citizens. An assessment of populace sentiment toward such tactics and toward the attendant retention of the DNA sample of 'potentially suspect populations' is necessary. Truly what is required is public consultation on how the justice system should proceed in securing our safety and protecting our freedoms. The government of Canada, when establishing the national DNA bank, engaged in public consultation so as to assess public opinion on the matter (Department of Justice, Canada. 2002). Additionally, the National Research Council (NRC) made recommendations upon the introduction of DNA in forensic settings as to its use and reliability (NRC 1992; NRC 1996). Further the National Commission on the Future of DNA Evidence and the National DNA Bank Advisory Committee are still making recommendations to regulators in the US and Canada (National Institute of Justice 2002; National DNA Bank Advisory Committee 2000). The Nuffield Council on the forensic use of bioinformation: ethical issues (2008) in the United Kingdom may offer a template for future discussions. Public consultation is not novel and there are mechanisms in place by which to do so (Simoncelli and Steinhardt, 2005). Truly informed debate and public consultation cannot take

place unless the trade-offs to be made by either retaining a stratified system or opting for an allinclusive scheme are clearly stated and discussed and the ramifications assessed. In the following text all comments about banking will be based on the current general practices of collecting samples from convicted offenders. In situations when referring to arrestee testing, as is the practice in Alaska, Arizona, California, Kansas, Louisiana, New Mexico, North Dakota, Tennessee, Texas and Virginia, arrestee banking will be clearly stipulated.

5.2 A Stratified System of Sample Collection: Advantages and Challenges

5.2.1 A Stratified System of Sample Collection: Advantages

5.2.1.1 Retention of Samples Limited to the Most Violent Offenders

In the stratified system, we currently have classified the need for a criminal's DNA based on the severity of the crime. This approach was designed to target those criminals that pose the most threat and, through DNA, to track their criminal behavior. In this scenario, the DNA bank holdings will be limited to those individuals who commit and are convicted of specific crimes. Limitations on the individuals included in the national DNA bank were proposed based on several criteria. Limited inclusion based on crime severity meant limited rights infringements of only the most dangerous offenders. There was consensus that the most violent offenders be included as this would ensure a minimal level of protection while keeping costs associated with collection and processing manageable (NRC 1992; Department of Justice, Canada 2002). Further the justification for privacy incursions was legally assured with the commission of such heinous crimes as rape, sexual assault and murder. The safety of the community outweighed the privacy of a few individuals who refused to comply with social norms. According to this system, the bank would gradually increase in size with the conviction of new individuals but would be manageable, as previous offender profiles would already be in the system.

5.2.1.2 Limited Entrants Implies Limited Abuse Potential

In general, most citizens abide by the laws governing violent crime and are not likely even to be questioned or implicated in a criminal investigation. In the original conceptualization of the Canadian National DNA bank, the strictly defined groups from which a sample might be obtained limited the potential intrusion on privacy interests and was well supported by citizens at large. A corollary of this is that, with only a small proportion of citizens in the bank, the impact of any potential misuses or abuses would be minimized. These limitations, as well as the penalties for unauthorized access, (DNA Identification Act 1998, DNA Identification Act of 1994 (1994)) helped to assuage fears of government surveillance.

Subsequent to the institution of a national DNA databank, the explosion of technological innovation has lead to forensic DNA expansion resulting in an increase in the number of sources e.g.: hair, skin and buccal swabs, from which DNA can be retrieved. Further the quantities of DNA needed to generate a profile have been substantially reduced (Yu and Wallace 2007; Balogh et al. 2003; Wickenheiser 2002; Seo et al. 2002). These changes have in part prompted reevaluation of the groups from which DNA can be sampled. Expansion of the types of offenses to be included in state and national banks has been proposed.

5.2.2 Challenges Presented by a Limited Data Bank

5.2.2.1 Not Maximizing the Potential of the DNA Bank

Under the current system DNA samples from convicted murders are routinely collected and stored. Though these people may have committed serious offenses this does not automatically mean that they will repeat their offenses. Crime statistics indicate that 58.5% of offenders commit single incidents (Statistics Canada Government of Canada National Parole Board, 2007). The implicit assumption in DNA profiling is that once convicted of a crime one

will likely commit the same or a more serious crime in future. Escalation for some types of crime may confirm this belief (chronic offenders, those with five or more incidents, account for 12% of the offender population and 50.8% of the crimes), but for crimes such as murder, most offenders are not more likely to re-offend than the average citizen is to commit a crime in the first place (Statistics Canada Government of Canada National Parole Board, 2007). [The obvious exception to this generalization is gang activity, mass murder or serial murder, each of which has distinct hallmarks (Fox and Levin, 1994, 1998)]. To maintain a stratified system different offenses triggering DNA collection might be considered. According to a pilot study conducted in seven provinces and two territories, 60% of convicted offenders aged 18-25 in 1999/2000 had at least one previous conviction and 72% of repeat offenders had multiple prior convictions (Justice Monitor, 2007, Statistics Canada 2002). Using current statistics why not target those groups most likely to re-offend along with those crimes for which DNA is most likely to be retrievable?

There have been arguments that limited inclusion may not be entirely useful in maximizing the utility of DNA evidence. There are crimes in which DNA may be easily retained and effective in identifying the perpetrator but are not considered dangerous enough to warrant collection of DNA (eg., petty theft). Further there are some data that suggests a pattern of crime escalation from petty theft to breaking and entering and subsequently to assaults of varying severities (Charron 2003; Schlesinger and Revitch 1999). This pattern is so consistent as to be characterized by one of the leading criminal profilers as 'sexual burglary', starting with peeping and the removal of delicates from clotheslines, on to breaking and entering with the removal of intimate or personal belongings and subsequently escalating to sexual assault and rape (Schlesinger and Revitch 1999). Arguments of this nature are driving the expansion of the

number of eligible offenses, particularly in the United States, with the extreme position being that DNA is collected from anyone who is arrested, regardless of the nature of the offense. However, from a practical perspective, it is not clear whether mass collection is more likely to yield additional "hits" or hinder an already backlogged forensic science division (US only, DNA Backlog Elimination Act 1999; DNA Analysis Backlog Elimination Act of 2000).

5.2.2.2 Magnifying racial disparity by over representing ethnic groups

Racial disparity exists throughout all stages of the U.S. criminal justice system (Duster 2004; Kalogeras and Mauer 2003). Beginning with arrest and proceeding through imprisonment and parole, substantial racial and ethnic disparities are present in virtually all jurisdictions in the United States. More than 60% of the people in prison are now racial and ethnic minorities (Kalogeras and Mauer 2003). For black males in their twenties, 1 in every 8 is in prison or jail on any given day (Western 2006; Kalogeras and Mauer 2003). African Americans are incarcerated at nearly six (5.6) times the rate of whites; Hispanics are incarcerated at nearly double (1.8) the rate of whites. Individual states exhibit substantial variation in the ratio of black-to-white incarceration, ranging from a high of 13.6-to-1 in Iowa to a low of 1.9-to-1 in Hawaii (Mauer and King 2007). Of the first 200 people exonerated through the use of DNA by the Innocence Project, 62% are African American, 10% are Latino and 28% are Caucasian (Innocence Project 2007). "...[T]he stigma of race and the stigma of imprisonment serves to keep alive in our public culture the subordinating social meanings that have always been associated with blackness" (Loury 2007). The additional stigma of being known to have a profile in the local, state or national database may further alienate ethnic minority individuals and communities (Nuffield Council 2008).

This is not a recent problem. A 1995 survey reported that, although the arrest rates for African Americans and Caucasians were equivalent, biased stop and search criteria lead to a disproportionate number of minority arrests by Maryland police (Higgins 1997; Duster 2004). Further evidence based on the 1991 data of Alfred Blumstein concluded that blacks comprised 57.7% of the prisoners for drug offenses, but only 40.4% of the arrestees for drug offenses (Blumstein 1993).

Canada is not immune from racial disparity within the criminal justice system, but this discrimination has tended to focus on Aboriginal Canadians as well as blacks (Mosher 1998; Roberts and Doob 1997). The commission on Systematic Racism in the Ontario Criminal Justice System in a large-scale study released data indicating racial disparity exists at two pre-trial release stages: the police when charged and police station and that there is a statistically significant racial bias in sentencing independent of the bail decision (Commission on Systematic Racism in the Ontario Criminal Justice System 1995; Griffiths and Verdun-Jones 1994; Roberts and Doob 1997).

Concerns of unjust and unfounded over-representation of minority groups within the forensic DNA databanks are justified, given the disparity in the treatment of racial groups within the U.S. and Canadian criminal justice systems, not to mention groups targeted after the World Trade Center attacks. Biases in arrest rates, conviction rates and the introduction of arrestee DNA sampling will only increase racial disparity. The expansion of DNA banks to include arrestees will certainly (continue to) stratify citizens based on past criminal acts and will create social and legal inequities exacerbating class distinctions (Rushlow 2007; Simoncelli and Steinhardt, 2005). The problem has become so abysmal as to lead one author to state that "Mass

incarceration has now become a principal vehicle for the reproduction of racial hierarchy in our

society" (Loury 2007).

"..[T]hose bearing the brunt of order enforcement belong in vastly disproportionate numbers to historically marginalized racial groups. Crime and punishment in America has a color. Its principal thesis is this: we law-abiding, middle-class Americans have made decisions about social policy and incarceration, and we benefit from those decisions, and that means from a system of suffering, rooted in state violence, meted out at our request. We had choices and we decided to be more punitive. Our society—the society we have made—creates criminogenic conditions in our sprawling urban ghettos, and then acts out rituals of punishment against them as some awful form of human sacrifice. Given our history, producing a racially defined nether caste through the ostensibly neutral application of law should be profoundly offensive to our ethical sensibilities—to the principles we proudly assert as our own. Originally published in the July/August 2007 issue of Boston Review (Loury, 2007)".

5.2.2.3 Unjust distribution of benefits and burdens

Recall from chapter 4 that distributive justice concerns what is just or right with respect to the allocation of goods among members of society. It is not only a central issue of moral and political philosophy, but an object of common-sense moral reasoning. I would argue that the current stratified system violates Rawls' formulation of distributive justice by placing greater burdens than benefits on some members of society and is therefore unjust. If we consider Rawls second principle of distributive justice with respect to a stratified DNA banking scheme, it must satisfy two conditions: (a) social inequalities attached to positions and offices under conditions of fair equality of opportunity are to be open to all; and (b) inequalities are to be to the greatest benefit of the least advantaged members of society (Rawls 1993, pp. 5-6). The principles are numbered as they were in Rawls' original *A Theory of Justice*. I will address section b of the second principle first and then return to the first section.

Initially, we must identify who the least advantaged members of society are. Are the victims of crime the least advantaged or are the criminals within the DNA bank least advantaged? A community whose individual members are rendered their due would be

considered a society guided by the principle of distributive justice. Thus it could be argued that the victims of crime have been rendered their due when the perpetrator of a crime has been imprisoned. Further, the criminal has also received his due upon incarceration. What happens after that point is a matter of human rights; "... the way societies treat those who have been deprived of their liberty is a litmus test of commitment to human rights" (The Right Hon Jack Straw MP in Coyles 2002).

In what category, advantaged or disadvantage, do we place individuals who have voluntarily given their DNA for exclusion, as compared to those from whom DNA has been collected covertly? Independent of how we define the groups, for the least advantaged members of society to receive the greatest benefit, we would have to demonstrate that a stratified system of DNA collection benefits those within the system as much as those not included. Inequalities can be just as long as they benefit the least well-off members of the group. The burdens, however, cannot be assumed by a section of the community that will not receive benefits. We cannot justify inequalities on the basis that the disadvantages of those in one position are outweighed by the greater advantages of those in another position. Currently all of the risks--loss of privacy, potential confidentiality violations, unconsented research---are assumed by those in the bank. The only benefit of being in the bank is the potential to be excluded as a suspect in other criminal investigations, but that in turn comes at the cost of having one's DNA profile repeatedly compared with multiple unidentified crime scene profiles. For those that argue that repeated searches of innocent individual's DNA is akin to negating the principle of "innocent until proven guilty" a reminder that these searches are performed en-mass and that no single individual is the target of a specific search is in order. There is one benefit that I have not mentioned and that is that society as a whole presumably benefits from increased security (either

through identification or deterrence of perpetrators of crime) however this is just the sort of thing that prompted Rawls to situate justice as fairness at an individual level as so as not to perpetuate injustice in a utilitarian fashion (Rawls Justice as Fairness 2001). Since distributive justice considers the distribution of goods among members of society at a specific time, and on that basis, determines whether the state of affairs is acceptable, it appears as if all of the burdens rest on those within the bank and are therefore unjust.

Now we must consider the first section of the second principle whereby offices and positions must be open to everyone under conditions of fair equality of opportunity, essentially on the basis of merit. The position of being in the bank for some and not for others could be viewed as an inequality of opportunity, given that being in the DNA bank is not open to all. There are those in the bank who provided DNA for exclusion, such as law enforcement personnel who had no choice or good samaritans who had limited choice due to poor informed consent, coercion or were unaware of the potential for databank inclusion. Convicted individuals within the bank presumably had the opportunity to remain outside but choose by committing a crime to be included. This choice wouldn't apply to individuals who were incarcerated before the implementation of the DNA profile bank as it was an unknown opportunity. Allowing for an opt-in system would ensure that the opportunity to be in the DNA bank is open to all.

"Injustice, then, is simply inequalities that are not to the benefit of all" (Rawls 1999). The second principle holds that an inequality is allowed only if there is reason to believe that the practice which includes or results in the inequality will work for the advantage of every party engaging in it. Every party must gain from the inequality. On a societal level can we argue that everyone benefits from identifying perpetrators under the current stratified system and that even the least advantaged member benefits?

Insert section: Brief discussion of how the current arrestee and surreptitious sampling change the equation of justice as fairness.

5.3 A Universal Bank: Advantages and Challenges

5.3.1 A Universal Bank: Advantages

Adopting an all-in arrangement has limitations as well, though it may help to ease existing discrimination and comport with distributive justice by placing all citizens on an equal footing within the forensic DNA databases. For example, mass inclusion of all individuals would be costly both in terms of financial and human resources. Further, there are those who argue that the mass influx of DNA from low-level offenders will result in backlogs and delayed identification and apprehension of violent offenders, in addition to increased error and inefficiency in the law enforcement system (Simoncelli and Krimsky 2007).

5.3.1.1 No Restrictions by Crime

Allowing all types of crime to qualify for inclusion would greatly expand the profile database and might improve efficiency in perpetrator identification. Limiting the acquisition of DNA based on the severity of the crime, as is current practice in most regions, is intuitive but fails to capture many of the recidivistic crimes for which trace DNA may be available such as theft, common assault and breaking and entering (Zamir et al. 2000, Wickenheiser, 2002, Seo et al. 2002). Based on 1999-2000 Canadian data for 18-25 year olds, 60% of convicted offenders were recidivists and property crimes accounted for 80% of the cases (Statistics Canada, 2002). Capturing and identifying this group could have a sizeable impact on crime rates.

5.3.1.2 Just Distribution

Under a universal DNA banking scheme everyone would be equal. There would be no disparity in race or gender and everyone would have a vested interest in the security of the DNA

bank. Consistent with Rawls' belief that the primary subject of justice is the basic structure of society, an all-inclusive scheme would meet his criteria for a distributively just social institution (Rawls 1999). Benefits and burdens would be such that there is equality of opportunity and the least advantaged member of society could benefit. The potential risks of loss of privacy, potential violations of confidentiality and unconsented research would be assumed by everyone in addition to the current benefits and probable benefits of increased identification of criminal perpetrators. There would however be an additional burden of cost associated with adding several hundred thousand profiles and samples to the bank both in terms of processing and administration. Further it can be argued that increasing the vested interest of society in the DNA banking system will not necessarily increase transparency of the banking organization nor will it necessarily strengthen the security of the profiling database and DNA storage facilities. It will require more than self-interest to motivate the populace, as evidenced by voter apathy in both U.S. and Canadian elections, given that elections are events in which self-interest is paramount (The Federal Election Commission and Elections Canada 2007).

5.3.1.3 Maximizes the Utility of the Database

The greater the number of persons with DNA profiles in the system, the greater the likelihood of obtaining a match between the perpetrator and the crime scene sample. Axiomatically, the probability of identifying the true perpetrator is only increased if there is DNA evidence. Of course there is a threshold beyond which a random match might occur requiring additional loci to be tested to distinguish between individual profiles and the offender profile. With the population of Canada at roughly 33,212,696 and the US at 303,824,646 (CIA World factbook Canada, United States July 2008) and the collection of 140,117 profiles as of August 2008 in the National DNA Data Bank of Canada and 6,031,000 profiles as of June 2008

in the National DNA Index System (CODIS) creating a complete database in either country will not happen quickly (National DNA Data Bank 2008, Federal Bureau of Investigation 2008).

5.3.2 A Universal Bank: Challenges

5.3.2.1 Security and Stringent Penalties for Improper Use

Admission of a DNA profile from each member of the general population would require employing a collective security approach to the database. All members would assume some responsibility for the protection, security, management and disclosure of the information based on a vested interest. The DNA banking establishment has set up a scheme where only profiles are matched in the CODIS databank. It is up to authorized personnel to contact each other to confirm the identity of the match as no names or personal information is stored with the profile. Thus it falls upon the vagaries of individual state employees to abide by each state's laws regarding access and the penalties for circumventing such laws. In addition there is considerable variability from state to state in the type of penalties, criminal or civil for various transgressions e.g.: tampering with samples or records, improper entry of samples or records into the database, improper access and use and improper disclosure of samples and records (American Society of Law Medicine and Ethics Survey of DNA Database Statutes Grid, 2006). A uniform DNA bank potentially could unify both security and sanctions of all states or provinces.

5.3.2.2 Public Concerns Mandate Public Consultation

Proposing a uniform, all inclusive, national DNA databank is not novel by any means however it is becoming a realistic endeavor as both the technology and law makers make the prospect more viable. The expansion of DNA banks is no longer a matter of if but when and public consultation is pressing. The Combined DNA Index System (CODIS) is a software program that operates local, State, and national databases of DNA profiles. CODIS uses two

main indexes Convicted Offenders and Forensic with additional indexes of Arrestee's, a Missing or Unidentified Persons, and a Missing Persons Reference. The Convicted Offender Index contains DNA profiles of individuals convicted of crimes ranging from certain misdemeanors to sexual assault and murder and the Forensic Index contains DNA profiles obtained from crime scene evidence like semen, saliva, or blood which are all used to generate investigative leads in crimes where biological evidence is recovered from a crime scene (see figure 1, Chapter 1 p.11). Since 2003, several states (18) have passed laws amending their existing DNA statutes, allowing for the inclusion of additional categories of individuals, such as "all felons, all criminals, misdemeanants, prostitutes, terrorists, those serving community sentences, immigration violators, and arrestees" [42 U.S.C. § 14135(a) (2006)]. Currently, eleven states, Alaska, Arizona, California, Kansas, Louisiana, Minnesota, New Mexico, North Dakota, Tennessee, Texas and Virginia, have legislation authorizing DNA collection from arrestees. Minnesota has decided, however, to repeal their DNA arrestee statute based on an Appeal Court ruling it an unreasonable search and seizure²⁷. The Justice for All Act (H.R. 5107), expanded CODIS by amending 42 U.S.C. 14132, 14133, 14135a, 14135e, 10 U.S.C. 1565(d) and allowing the inclusion of "persons who have been charged in an indictment or information with a crime," and "other persons whose DNA samples are collected under applicable legal authorities" in CODIS. This permits DNA profiles collected by those states that currently authorize the inclusion of

²⁷ In the Matter of the WELFARE OF C.T.L., Juvenile, which was tried before the state's Court of Appeals, a juvenile charged with aiding and abetting first-degree aggravated robbery and committing fifth-degree assault challenged the constitutionality of the Minnesota statute which allowed for the taking of a biological specimen for the purpose of DNA analysis. The court held that statutory provisions that direct law enforcement to take biological specimens from juveniles and adults who have had a probable cause determination on a charged offense but, as here, have not been convicted, violate state and federal constitutional prohibitions against unreasonable searches and seizures [In the Matter of the WELFARE OF C.T.L., Juvenile, 722 N.W.2d 484 (Minn. App. 2006)].

arrestees in their state DNA databases as well as other DNA profiles, such as those of missing persons, if "collected under applicable legal authority" to be uploaded into CODIS. In addition the statute requires that arrestee profiles in the DNA database be "promptly expunged" from CODIS by each state for individuals not convicted of the crime and for whom the charges have been dismissed or an acquittal entered. Further, with the amendment of The Violence Against Women Act 2005 (VAWA), in January 2006, the federal government is now required to collect DNA samples from persons arrested or non-US persons detained under federal authority (The Violence Against Women Act 2005). Anyone now arrested by a federal government official — for any reason whatsoever, regardless of the nature of the violation — will have their DNA sampled for the database.

Collection and retention of samples and profiles from persons using DNA dragnets and surreptitious sampling further complicates matters. Though the samples may not be uploaded to the national database they are still searchable within the state where they are held and serve as further temptation to expand CODIS without informed public debate.

Upon the disclosure, in 2006, of the government's practice of collecting phone records to investigate terrorism, more citizens disapproved than approved (Jones 2006). Fifty-seven percent of polled Americans felt that their personal privacy was violated when their telephone records were in the federal government's possession (Jones 2006). Based on the response to this incursion of privacy rights, it is likely that citizens will feel strongly about placing their DNA and DNA profile in the hands of their government. The Nuffield Council on the forensic use of bioinformation: ethical issues (2008) recommends against a population-wide DNA database as they claim the increased intrusion into privacy would only negligibly increase public safety. They conclude that it would be disproportionate to the need to control crime; be unlikely to

secure public support; and be impractical for the collection of samples from different categories of persons (such as visitors).

5.3.2.3 Government's Historical Treatment of Criminals and Subversives:

Historically governments do not have a good record with respect to humane treatment of criminals and subversives (Amnesty International 2008; Rummel 1994; Iacovetta, Perin and Principe 2000). Though the U.S. and Canada have somewhat better recent records there are still prominent examples of mistreatment. The rounding up of Japanese American citizens into internment camps during the second world war, the McCarthy hearings, prison chain gangs, the detainees at Guantanimo and the wire tapping of "suspect citizens" homes. Canadian examples include similar treatment of Japanese and Ukrainian Canadians during the war and ongoing aboriginal disputes. It is against this backdrop that concerns of potential governmental abuse rest. The maintenance of the DNA databases reinforces the power and efficacy of the State in matters of security, but may affect the vulnerability of the citizens, namely on safeguards of its rights, freedoms and guarantees. Under these circumstances the collection, conservation, use and circulation of the data become ethical issues, blurring the line between procedural and philosophical issues. Security not just of the person, but of the sample and ultimately the profile and the access to that information, is vital to the continued expansion of DNA databases. The results of DNA profiling and searches are particularly charged due to the permanent nature of the consequences and the potential dangers of a match by coincidence, kinship, contamination and other error either typographical or laboratory (Teichroeb 2004a; Teichroeb 2004b; Seattle Post Intelligencer Staff 2004; Penacino, Sala and Corach 2003; Smith 2007; Saltzman & Ellement 2007).

A universal bank could be worse than the present system because more people could suffer unjustly unless the collection, maintenance and management of data are subject to strict principles of transparency and independence and to high standards of quality and oversight (Maier 2003).

5.3.2.4 Fear of Government Surveillance and Secondary Uses

Gradual expansion without public consultation may jeopardize the existing social contract and open the justice system to attack for lack of transparency and violations of the public trust and previously guaranteed rights. The rapid expansion of the UK DNA Bank and its notoriety as the largest DNA database in the world (4 million samples) has evoked outcries of government surveillance (Bloxham A. 2008; Bird S. 2007; Woolf M, Goodchild S. 2006). Failure of the government to fully inform and consult the public, and legislate practices has aroused legitimate concerns (GeneWatch UK 2007; GeneWatch UK 2006; House of Commons 2005; Staley 2005; Williams, Johnson and Martin 2004). Perpetual retention of all profiles and DNA samples (innocent and guilty) along with research agendas and inadequate public disclosure has made the citizens wary if not fearful of government surveillance (GeneWatch UK 2008).

The ever expanding DNA databases in the U.S. and Canada along with the retention of DNA samples are a legitimate cause for concern. In the U.S. samples are retained by the state facility from which the profile was generated. The patchwork regulations governing secondary uses, varies from state to state, from highly permissive to entirely restrictive. The only federal intervention is federal funding guidelines that regulate research with this population. The Canadian National DNA Data Bank (NDDB) is composed of six RCMP regional laboratories, the Center of Forensic Science (Toronto) and the Laboratoire Science Justice Medico Legale

(Montreal) all of whom contribute DNA profiles and samples to the NDDB (National DNA Data Bank Advisory Committee, 2006). Understanding the need for epidemiological information of allele frequencies among different populations and taking into account that a "match" precipitates a warrant request for an additional sample; why continue to maintain samples with identifying information? Why maintain convicted offender samples at all when the resources can go toward maintaining and preserving crime scene samples and ensuring that more crime scenes are properly and completely analyzed and processed for trace evidence?

The presence of the DNA sample in government custody is a temptation for use and even the smallest amount of DNA can be amplified to accumulate enough information to be intrusive. Public consultation would allow for open debate on other issues such as secondary uses, as allowed in some jurisdictions, and adequate protections to discourage abuse and reduce human error.

5.4 Summary and Conclusion: Call for Public Consultation

Well conceived communication and consultation promotes public understanding of legislation, policies, programs and services and provides a critical insight into public attitudes and expectations. Further, good communication inspires commitment, enhances organizational cohesiveness, and promotes the development and implementation of legislation. Failure to be explicit about use and transparent about motives leads to suspicion and erosion of the relationship between the citizenry and the state. Similar lessons have been learned in research and medicine (Levitt and Weldon 2005; NIH News. 2005). Engagement with the public in order to solicit their views as an integrated part of the decision-making process should be the priority of legislators and law enforcement interested in mandating the expansion of local and National DNA Banks.

Ensuring that administrative and policy processes are open and accessible, are respectful of the public's right to be involved, and are responsive to the public need for information is key. Public consultation is fundamental to acquiring public understanding, and support, for the actions of the Criminal Justice System. The time to act is now. The Nuffield Council on Bioethics report on the forensic use of bioinformation: ethical issues could provide a template for discussion.

Canada has a strong history of public consultation through Royal Commission inquiries and this type of body can not only collect information from knowledgeable parties i.e. federal and provincial law enforcement, federal agency officials, academics and research scholars, scientists, crime laboratory officials, leadership of professional standard setting organizations, defense attorneys etc... but also formulate a survey or questionnaire to assess the opinions of the larger populace on the sacrifices that they are willing to make to ensure public safety and buttress the Criminal Justice System. The advances in electronic communication allow for wide spread stakeholder interaction. In addition open hearings could be held as a venue for position papers from both interested individuals and interest groups. The United States has recognized a similar need to unify forensic science evidence and Congress charged the National Academy of sciences to perform a similar assessment of forensic science, legal and diverse scientific communities to help formulate recommendations to establish The National Institute of Forensic Science and guide the future of the forensic science enterprise.

Consensus must be reached on surreptitious sampling and notification; guidelines and transparency of research protocols with forensic samples; resource allocation (maintaining identified samples versus expanding crime scene sample collection); and developing a roadmap for the expansion of DNA Bank holdings. As has been the case with ethical issues in the use of

molecular genetics for clinical testing, Canada should take the lead in developing the guidelines for ethical use of DNA in forensic testing.

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CHAPTER 6: CONCLUSION

While DNA profiling rests on a stronger scientific foundation than many other forensic disciplines and has come to dominate the field, as well as, popular media, its unchecked expansion holds threats to our social core. Although the field of ethics has made attempts to cope with the plethora of available genetic information, especially in clinical applications, there has been modest attention to emerging ethical issues in the forensic domain. Legal scholarship has drawn attention to some aspects of the emerging issues, with particular relevance to the challenges faced in court and those regarding individual liberties.

In particular, contemporary science has allowed us to access information far beyond what was imagined two decades ago, such that trace DNA can be obtained trivially from any individual. As a consequence, the scope and composition of existing DNA banks far exceeds the legislative mandate.

An evaluation of current practices in DNA banking revealed adequate attention to obtaining informed consent, though the concept of informed consent in genetic banking may not be sufficient to protect individuals' interests. The need for a re-examination of access to public health samples with attention to local population interests and regard to distributive justice was highlighted. Further, the development of standardized guidelines for banking practices and uniform quality assessment measures is crucial.

A comparison of current forensic and genomic markers revealed similar concordance and discordance rates with a modest performance advantage towards the forensic markers. The results support performing multiple runs to ensure reliability and further investigation to determine the optimum number of repeat runs. SNP genotyping is inexpensive, easy to automate, have high-throughput, make efficient use of samples and have reduced error rates as

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compared to STRs. Moreover, they are more abundant, more robust, have a lower mutation rate and though their lower polymorphism information content requires more alleles to be interrogated, this can be easily accomplished with recent technological advances. As the investment in SNP technology increases and multiplex panels become more refined, SNPs will likely compliment STRs in forensic DNA profiling.

A significant ethical issue arises from the forensic practice of surreptitious DNA sampling. This lack of transparency violates the autonomy of ordinary citizens, threatens the legitimacy of the State's interest by failing to underwrite the welfare of its citizens in securing their person and property. This practice may exacerbate existing issues of discrimination and universality, infringe privacy and confidentiality through coercion and failure to obtain meaningful consent. Public consultation is required to develop guidelines for socially agreed upon methods of DNA sampling, the utilization of forensic samples in research, resource allocation and the extent to which a universal DNA bank is appropriate.