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Title of Thesis/Project/Extended Essay

Maternal Genetic Fingerprints: An Examination of the Science Underlying Mitochondrial DNA and Relevant Cases

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Abstract

In most forensic cases, it is assumed that the reader knows that the deoxyribonucleic acid (DNA) mentioned is nuclear DNA; the genetic blueprint of life that can distinguish between all individuals except identical twins. In actuality, there are two types of DNA. The other category is mitochondrial DNA (mtDNA), which can only be used to differentiate maternal relatives. This second type of DNA is extremely useful due to its high copy number, enhanced stability, and maternal inheritance. However, laboratories that perform this type of analysis must be conscientious due to the extreme sensitivity and precision of the test. Unfortunately, there are only six laboratories in the world that currently are able to perform human mtDNA analysis for law cases due to the high cost involved.

Recently there has been an increase in the number of forensic cases in which mtDNA evidence has been used. The first case in which this type of evidence obtained recognition was in the identification of the Romanov family. A new scientific development called heteroplasmcy (base insertions, deletions, or replacements) was discovered in the mtDNA sequence of Czar Nicholas II. Therefore, his brother's body needed to be exhumed in order to obtain a clear match and finally settle the identity of Czar Nicholas Romanov. Another new advancement, the supposition that the maternal genetic fingerprint may also be partly paternally inherited, has caused the scientific community to reconsider the mtDNA sequence first discovered by Anderson et al. in 1981. There have been a total of six human cases in the United States, beginning in 1996, in which mtDNA has been used to obtain convictions. In Canada, there has been only one human case, R. v. Murrin, which has involved mtDNA evidence. However, this case did not result in a conviction, event though the fingerprint
matched with over 99% accuracy, due to other mitigating factors, which may have had some impact upon the verdict of not guilty.
Acknowledgments

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Introduction

Mitochondria have an extranuclear DNA genome, the sequence of which was first reported for humans by Anderson et al. in 1981. "Even though the human mitochondrial DNA circular genome of 16,569 base pairs is much smaller than the three billion base pairs thought to comprise nuclear human DNA, mtDNA has become important in anthropological and evolutionary research as well as in forensic identification" (Butler and Levin, 1998: 158). An important difference between nuclear and mitochondrial DNA lies in the pattern of inheritance—"mitochondria come from the egg of the individual's mother with little or no contribution from the father, whereas nuclear DNA is a combination of DNA from both parents" (Butler and Levin, 1998: 158). Therefore, nuclear DNA goes through recombination with each generation, while mtDNA represents only the maternal ancestry of an individual.

Human mtDNA consists of "a coding region which contains the sequences for thirteen proteins and a non-coding region called the displacement loop (D-loop) or control region, which is approximately 1,100 base pairs in length" (Butler and Levin, 1998: 158). "The forensic value of mtDNA emerges from the sequence variation in the D-loop between individuals and its resistance to extreme environmental conditions" (Butler and Levin, 1998: 158). In addition to skin, blood, semen, and saliva, mtDNA has also been extracted from teeth, hair shafts, bone fragments, and feces, all of which fail to yield adequate results with nuclear DNA analysis. Mitochondrial DNA has also proven to be more robust than nuclear DNA in typing ancient materials "due to its higher copy number per cell and its resistance to extreme environmental conditions" (Butler and Levin, 1998: 158). Hundreds to thousands
of copies of mtDNA may exist in a single cell, making it easier to recover and sequence than the single copy of nuclear DNA per cell. Furthermore, "the haploid nature of mtDNA makes it easier to sequence than diploid nuclear DNA" (Butler and Levin, 1998: 158).

Forensically, mtDNA gained notoriety when it was used to identify the remains of the Romanov family after almost 70 years underground. "An exact sequence match of the D-loop region was made between the Tsarina, her three daughters, and the living Prince Philip of the United Kingdom, whose maternal grandmother was the Tsarina's sister" (Butler and Levin, 1998: 158). Presently, mtDNA is used in cases in which the nuclear DNA is too degraded or cannot be recovered in sufficient quantities to be analyzed. However, this type of analysis is expensive and time consuming. Therefore, only a few laboratories worldwide have the capability to carry out the necessary tests for human cases. Even though numerous studies have been performed to confirm that mtDNA fingerprinting is a reliable means of forensic identification, a number of technical challenges remain. One such concern is "contamination, especially when the samples are available in limited quantities or are old and partially degraded" (Butler and Levin, 1998: 161). Extreme care must be taken to avoid contamination. A second concern is "heteroplasmy, a condition in which a single individual has more than one mtDNA sequence" (Butler and Levin, 1998: 161). These different sequences "can exist in the same mitochondrion, in different mitochondria, or in different cells of the same individual" (Butler and Levin, 1998: 161). Although heteroplasmy has been used as an identification factor (e.g. Tsar Nicholas II and his brother exhibited the same base substitution), in most cases, "heteroplasmy makes identification more problematic because it is difficult to detect and may be more prevalent than previously reported" (Butler and Levin, 1998: 161). Therefore, some laboratories will not exclude a match based on a single
control region difference.

Mitochondrial DNA is an important tool in forensic identification as it can be used to link maternal relatives who have the exact same mtDNA. In the United States, there are presently six cases which involved mtDNA evidence since 1996. All of these cases ended with the conviction of the defendant. In Canada, only one case has been tried by the courts. This event was the *Murrin* case, in 1999, which ended with a verdict of not guilty, even though there was a 99.76% match with the maternal DNA between the hairs found in the victim's underpants and the defendant. The commonalities and differences between these cases will be examined in order to make sense of why mtDNA evidence has been accepted so readily in the United States but not in Canada. Several other important factors pertaining to the *Murrin* case in Canada will also be examined, which may have had an impact on the verdict.

There are several new opportunities in which mtDNA could be used in the future. One interesting element is the ability of scientists to obtain a mtDNA fingerprint from an ink or latent fingerprint. This would be useful in cases in which the suspect is known to the police and has a fingerprint on record but is deceased, for example. A mtDNA genetic blueprint could be matched with an unknown crime scene sample and a potential suspect could be confirmed as the actual perpetrator of the crime. In Canada, the use of fingerprints as investigative tools has been affirmed by the Supreme Court of Canada in *Beare*. However, using fingerprints for another use, such as DNA analysis, would bring *Canadian Charter of Rights and Freedoms* issues to the surface. Additionally, mtDNA analysis can be used to identify missing children by matching mtDNA with maternal relatives (e.g. aunts, grandmothers, mothers, or siblings). Presently, there are many agencies that provide
identification kits in which parents can take a child’s fingerprints, along with a recent photograph and other vital personal information, and make an identification card to be used if the child goes missing. Mitochondrial DNA information could also be added to these cards in order to provide a tentative identification if a body is found and the identity is unknown or the features are unrecognizable. This would ultimately provide closure for the family. This potential use for mtDNA will be explored.

There are four clear components that comprise this thesis. The first chapter describes the science of DNA, both nuclear and mitochondrial. The legal uses of nuclear DNA will be discussed, as well as a brief description of some important Canadian cases. Following this, the structure, function, inheritance, and evolution of mitochondria will be considered, as well as their DNA and the advantages and disadvantages of its analysis. Subsequently, a comparison between nuclear and mitochondrial DNA analysis will be performed. Lastly, the design of the mtDNA genome, the analytic approach and quality assurance of mtDNA fingerprinting, and a list of laboratories that perform mtDNA analysis will be explored. The purpose of this chapter is to familiarize the reader with the science of DNA without going into too much scientific detail, so that the rest of the thesis will be readily understood.

The second chapter deals exclusively with mtDNA casework. Four United States cases and one Canadian case will be compared and contrasted. As well, the saga of the Romanovs will be discussed as a leading example of the use of mtDNA to link unknown human remains to known maternal relatives. Additionally, the only case in which mtDNA was used by the defence will be briefly reviewed. This section of the thesis ties together the science of mtDNA with the legal decisions that have been made in both Canada and the United States, while aiming to determine why mtDNA evidence has been readily accepted.
in the United States but not in Canada.

The third chapter evaluates new developments in mtDNA analysis and future developments for the science. Specifically, cases dealing with hair, bone, and tooth analysis will be examined due to the fact that these materials can be sequenced by both nuclear and mitochondrial DNA methods but after environmental damage, mtDNA analysis gives much better results. Secondly, research pertaining to obtaining mtDNA analysis results from ink fingerprints will be discussed. This type of research will be contrasted with the Supreme Court case of Beare and possible Charter challenges will be examined. Thirdly, cases of missing children in which unknown remains have been linked to known maternal relatives will be analyzed. As well, agencies and registries that deal with missing children will be evaluated for their willingness to use mtDNA samples to associate children and relatives. Finally, the use of mtDNA to identify disaster victims will be explored. Due to the fact that mtDNA is more robust than nuclear DNA, it can be used in cases where the samples have been exposed to environmental stress (e.g. plane crashes). The information contained in this chapter is meant to bring the science of mtDNA analysis into a more realistic view by examining real life scenarios in which mtDNA evidence has been used.

The fourth chapter of this thesis investigates two new scientific developments that may impact the analysis of mtDNA—heteroplasmy and paternal contribution. The discovery of sequence heteroplasmy may cause false exclusions in the comparison of mtDNA sequences, which would impact the identification of suspects in criminal investigations, while a paternal contribution to the mtDNA genome could lead to a more positive rather than tentative identification using mtDNA. This chapter strives to contemplate new advances in the science of DNA that also impact the legal setting, but which may not have been examined.
in the above mentioned cases.

The fifth and final chapter includes conclusions that can be drawn from this thesis, as well as recommendations for the future use of mtDNA.

The decision to undertake this thesis topic stemmed from my interest in DNA, which arose from studying Biochemistry at the University of Saskatchewan. During my course work, I learned how to perform PCR analysis of nuclear DNA, which I found extremely interesting. With a little additional reading, I discovered the forensic usefulness of DNA, which piqued my interest in the subject. After I wrote my Honours' thesis on the topic of DNA databases, I was eager to extend this topic into a Master's thesis. Unfortunately, the material seemed to be exhausted after the first thesis, until further data is available, which led me to pick another related DNA topic. At that time, the Shannon Murrin case was just entering the Supreme Court in Vancouver, so it seemed natural to use the first mtDNA case in Canada as part of a thesis. The rest of the topic developed from consultations with my supervisor, Dr. Gail Anderson, and an exchange of e-mails with the Head of the RCMP Forensic Lab in Ottawa, Ontario, Dr. Ron Fourney. I believe this research is significant and noteworthy as no other material has been written on this subject that encompasses descriptions of the science underlying mtDNA, mtDNA case histories, and future developments. Research has been performed in a scientific context, but not one that examines the legal significance of mtDNA.
Chapter I: The Underlying Science of DNA

Nuclear Deoxyribonucleic Acid: The Building Block of Life

Description

The human body consists of trillions of cells. Each cell contains a nucleus, within which are 46 chromosomes, divided into 23 pairs (Campbell, 1996: 281). Along each chromosome are approximately 10,000 genes. The gene is the fundamental unit of heredity. It “instructs the body’s cells to make proteins that determine everything from hair colour to susceptibility to disease” (Saferstein, 1998: 403). Each gene is composed of DNA. DNA is a polymer containing repeating nucleotides (a sugar molecule, a phosphorus-containing molecule, and one of four nitrogen-containing bases—adenine, guanine, cytosine, and thymine). Two strands of DNA coil into a double helix, with adenine pairing with thymine through a double bond and cytosine pairing with guanine through a triple bond, to make a ribbon of DNA that has been compared to a zipper with the bases forming the teeth (Saferstein, 1998: 404).

Humans inherit nuclear DNA upon conception—half from their mother’s egg and the other half from their father’s sperm. “The fertilized egg divides so that each cell in the body replicates the DNA from the original cell, and the DNA remains the same from conception to death” (Department of Justice, 1994: 3). No two people have the same nuclear DNA, except identical twins. In 1985, “Alec Jeffreys determined that by examining certain sections of nuclear DNA, scientists could differentiate between individuals” (Department of Justice, 1994: 3). These sections of nuclear DNA are considered highly polymorphic—they differ greatly among individuals (Department of Justice, 1994: 3). Most of the chemical
combinations in nuclear DNA are common to us all, since we have so much in common with each other as humans (Department of Justice, 1994: 3). However, the polymorphic sections are not common between individuals. The forensic significance of this is that the more polymorphic sites that match, the less likely it is that a different person contributed the evidence sample (Department of Justice, 1994: 3). The other significant point is that a non-match at any of these polymorphic sites absolutely excludes the individual whose nuclear DNA profile is being compared to the nuclear DNA profile in the evidence sample (Department of Justice, 1994: 3). Canada’s nuclear DNA sampling is governed by the Criminal Code, which states that “forensic DNA evidence may be performed on any of three bodily substances: individual hairs from the person, including the root sheath; buccal swabs by swabbing the lips, tongue, and inside cheeks of the mouth to collect epithelial cells; or blood by pricking the skin surface with a sterile lancet” (Criminal Code section 487.06(1)).

Legal Uses

Since its forensic introduction to Canada in 1988-89, DNA analysis has been instrumental in securing convictions in hundreds of violent crimes. It has also helped to eliminate suspects and has led to the exoneration and release of previously convicted individuals (e.g. Guy Paul Morin and David Milgaard). DNA evidence is a powerful and compelling form of scientific evidence, “almost to the point where it takes away the decision-making role from the jury” (Forensic Science Service, 1997-98: 9). DNA evidence can be contrasted with fingerprint evidence, which has long been accepted by the Courts but for which there is far less statistical backing when compared with DNA evidence (Forensic Science Service, 1997-98: 9). DNA evidence can focus investigations, and will likely shorten trials and lead to guilty pleas. It could also deter some offenders from committing
serious offences (Solicitor General, 1996: 3).

In December 1998, Canada passed legislation to enact a National DNA Database. Through the storage of DNA data in computer databases, DNA analysis can be used to solve crimes without suspects. Forensic scientists can compare DNA profiles of biological evidence samples with a database to assist the police in detecting suspects (Solicitor General, 1996: 3). “A database would also enable unsolved earlier offences, where DNA evidence had been found but not linked with an offender, to be solved if DNA samples taken from a suspect in connection with a later offence matched the evidence found at the scene of the earlier crime” (Solicitor General, 1996: 3). A national database would also help police identify serial offenders both within and across jurisdictions (Solicitor General, 1996: 3). It will take approximately one and a half years for the database to become operational. At this time it will be interesting to see how the courts rule on privacy and security of person Charter issues that have been brought to the debate over this Bill (Lambert, 1999: 9).

Mitochondrial DNA: The Other Genetic Material

Structure and Function of Mitochondria

Mitochondria are “small, intracellular, pink, oblong shaped organelles present in the cytoplasm of nearly all eukaryotic cells” (Darley-Usmar and Schapira, 1994: 1). The number of mitochondria per cell ranges between hundreds and thousands, depending upon the cell’s level of metabolic activity (Campbell, 1996: 126). A typical human has “approximately 35 to 100 trillion cells in his/her body which contain 100-1000 mitochondria each” (Fourney, 1998: 47). Therefore, “the average human has 170,000 trillion mitochondria” (Fourney, 1998: 47). The primary function of mitochondria is to provide energy for cellular functions
Evolution of Mitochondrion

In the early 1960s, "a number of experiments showed that mitochondria could not be produced by cells de novo, but instead always arose from the division of preexisting mitochondria— they are self-replicating" (Purves, Orians, and Heller, 1995: 76). By the end of the decade, it was clear that mitochondria had their own DNA, and some researchers began to speculate that they might indeed have once been semi-independent creatures. Led by Lynn Margulis, "a number of researchers suggested that today's mitochondrion are the descendants of ancient prokaryotes (prokaryotes are molecules surrounded by a membrane and cell wall but lack characteristic eukaryotic subcellular membrane enclosed 'organelles') that took up residence within eukaryotic cells, and provided them with important biochemical benefits, notably the ability to oxidize food compounds and produce energy" (see Figure I) (Purves, Orians, and Heller, 1995: 76).

Figure I: Endosymbiotic Origin of Mitochondrial DNA
This has been called the endosymbiotic theory and it was Margulis who proposed the following idea:

Picture a time, more than a billion years ago, when only prokaryotes inhabited the Earth. Some of them got their food by absorbing it directly from the environment, others were photosynthetic, and still others fed by eating their prokaryotic neighbours. Suppose that an occasional small, photosynthetic prokaryote was ingested by a larger one but did not get digested, so it sat trapped within the larger cell. Suppose further that the smaller prokaryote survived there and that it divided at about the same rate as the larger one, so successive generations of the larger prokaryote continued to be inhabited (or infected) by the offspring of the smaller one. We would call this endosymbiosis: “living within” another cell or organism, as, for example, certain algae live within sea anemones (Purves, Orians, and Heller, 1995: 77).

**Mitochondrial Deoxyribonucleic Acid**

Each mitochondrion possesses several loops of DNA; “each loop contains 37 genes which can encode thirteen proteins involved in energy production” (Darley-Usmar and Schapira, 1994: 2). Human mitochondrial DNA (mtDNA) is made up of exactly 16,569 base pairs (see Figure II and II).
Figure II: Mitochondrial DNA Map

The Human Mitochondrial DNA
From MITOMAP http://www.gen.emory.edu/mitomap.html
This can be distinguished from nuclear DNA which has more than three billion base pairs. There are "two principal regions of variation that scientists study in mtDNA, which represents approximately 3.6% of the total mitochondrial DNA content" (Fourney, 1998: 47). A wide variety of "degenerative diseases involving the central nervous system, heart, muscle, endocrine system, kidney, and liver have been associated with systemic mtDNA mutations, either base substitutions or insertion-deletions" (Fourney, 1997: 3).

**Maternal Inheritance of mtDNA**

Since mitochondria are contained within the cytoplasm of cells, and the ovum contributes more cytoplasm to the zygote than the sperm, mitochondria are maternally inherited (Campbell, 1996: 278). The consequence is that each individual will inherit the same mitochondria from his/her mother, and descendants in the female line will share an
exact duplicate of this DNA. Therefore, due to the fact that it has been proven that mtDNA is transmitted by females only, it is possible to trace genetic variation through families (Collins, 1997).

**Significance of mtDNA**

Applications of mitochondrial DNA are many. It can be used to trace the evolution of humans; as women migrate from continent to continent, mtDNA gradually accumulates one non-pathogenic mutation after another (McCord, 1). Therefore, the type of mtDNA varies from one continent to the next and the types can be traced as well as the relatedness of the different types. The amount of variation in each location determines the age of the population, since the rate of variation is constant and has been estimated to occur once every 2000-3000 years (McCord, 1). In addition, the more variation in a population the longer it has been present in a particular location. By examining and studying the mutations and common sequences found within mtDNA from different parts of the world, we can tell, with reasonable certainty, which population is descended from any other. Several such studies designed to correlate the origin of mtDNA were carried out and all came to the same conclusion, romantically called the ‘Eve Hypothesis’ (McCord, 1). It claims that all of us are descendants from some common ancestor female (Eve, from somewhere in central Africa).

**Advantages and Disadvantages of mtDNA Analysis**

There are several advantages of mitochondrial DNA analysis over nuclear DNA analysis in forensic science. First, the large number of copies of the DNA (i.e. 100s in a cell vs. one in a nucleus) greatly increases the prospect of obtaining useful DNA results (Fourney, 1998: 50). All tissues contain mtDNA, and any sample that can be used for nuclear DNA
analysis can also be used for mtDNA analysis (Fourney, 1998: 50). As well, there are several other samples that can only be used for mtDNA analysis, such as badly damaged or aged teeth and bones. Second, the maternal inheritance of the DNA makes interpretation relatively simple—only a single copy exists (Fourney, 1998: 50). Therefore, unidentified bodies or parts can be easily linked to living relatives. Third, mtDNA can survive harsh natural and environmental abuses. It even appears that in some tissues, mitochondria are more resistant to degradation than nuclear DNA. For example, successful results have been achieved from bones buried for numerous years, badly burned, and even exposed to saltwater (Fourney, 1998: 50). According to Fourney, "old or degraded samples and even hair shafts containing little or no roots are also sources of mtDNA" (Fourney, 1998: 50). Finally, the procedure used is based on a sophisticated scientific procedure and is a very precise way of identifying variation and similarities between individuals (Fourney, 1998: 51). The method used to determine how rare or common a DNA pattern may be in the general population is based on simple observation and relies on no specific scientific molecular or population biology principles (Fourney, 1998: 51).

Nevertheless, there are also several disadvantages. First, mtDNA analysis will not provide discrimination between individuals who are related maternally. Accordingly, brothers or sisters who are biologically related have the same mtDNA pattern, as well as aunts, uncles, and grandmothers (Fourney, 1998: 51). Second, there is still the probability of a match between unrelated individuals in the general population (Fourney, 1998: 51). Third, the procedure is very sensitive to contamination and is quite expensive ($2,000 U.S. dollars per sample on average) (Fourney, 1998: 51). In addition, few laboratories are equipped to handle mtDNA casework. Fourth, the cause and frequency of spontaneous
change in an individual’s mtDNA pattern are not understood (Fourney, 1998: 51). Members of the same family have been documented to have differing patterns. This phenomenon is called heteroplasmy and may cause some concern regarding the true level of discrimination between a match (Fourney, 1998: 51). For example, “a single site variation between two mtDNA samples may be a ‘false exclusion’ suggesting that two samples have come from different individuals” (Fourney, 1997: 9). In addition, heteroplasmy has been observed between two samples of the same individual (e.g. different sequences in a sample of hair and a sample of blood). For example, a case reported by the FBI shows that mtDNA was sequenced from a woman’s sample of blood for the purpose of establishing a mtDNA database. The base at position 16,355 was originally typed as cytosine (Wilson et al., 1997: 168). However, “with the procurement of an improved technique to sequence hair, another sample from the same woman was taken, but the base at position 16,355 was typed as thymine” (Wilson et al., 1997: 168). Therefore, the sequence from the blood differed from the sequence of the hair from the exact same woman. In order to limit false positive identification, “the FBI advocates that multiple references should be obtained and carefully scrutinized for the presence of heteroplasmy at the relevant nucleotides” (Wilson et al., 1997: 170). Finally, the procedure is relatively new to the courts and may require some special expert testimony prior to legal acceptance (Fourney, 1998: 51).

**Comparison of Nuclear and Mitochondrial DNA Analysis**

Mitochondrial DNA differs from nuclear DNA in its location, its sequence, its quantity in the cell, its robustness, its lack of recombination, and its mode of inheritance (see Table I) (Isenberg and Moore, 1999: 1).
Nuclear DNA is housed within the cell’s 23 pairs of chromosomes which reside in the nucleus, while mitochondrial DNA is situated in the mitochondrion, which reside in the cell’s cytoplasm. Nuclear DNA has more bases than mtDNA (four—A, T, C, and G) but mtDNA is displayed in more copies than nuclear DNA, which is useful where the amount of DNA is limited (Isenberg and Moore, 1999: 1).

Nuclear DNA testing is more precise than mtDNA testing due to the fact that nuclear DNA is found within the nucleus of the cell and is inherited equally from both the mother and the father (Hansen, 1998: 67). Therefore, nuclear DNA is unique to every individual, except identical twins. Mitochondrial DNA, however, is inherited solely from the mother (Hansen, 1998: 67). Consequently, all the offspring of the same mother will share the same mtDNA pattern, which also holds true for several generations.

Mitochondrial DNA is more durable than nuclear DNA so "sequences can be

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**Table I: Comparison of Human Nuclear and Mitochondrial DNA**  
(Butler and Levin, 1998: 159)

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Nuclear DNA</th>
<th>Mitochondrial DNA</th>
</tr>
</thead>
<tbody>
<tr>
<td>Size</td>
<td>~3 Billion Base Pairs</td>
<td>16,569 Base Pairs</td>
</tr>
<tr>
<td>Copies per Cell</td>
<td>2</td>
<td>Can Be &gt;1,000</td>
</tr>
<tr>
<td>Structure</td>
<td>Linear; Packaged in Chromosomes</td>
<td>Circular</td>
</tr>
<tr>
<td>Inherited From</td>
<td>Father and Mother</td>
<td>Mother</td>
</tr>
<tr>
<td>Generational Recombination</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Mutation Rate</td>
<td>Low</td>
<td>At Least 5-10 Times Nuclear</td>
</tr>
<tr>
<td>Sequence</td>
<td>Being Determined by the Human Genome Project</td>
<td>Described in 1981</td>
</tr>
</tbody>
</table>

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deduced from very old, very degraded, or very small quantities of DNA” (Hansen, 1998: 67). Therefore, mtDNA analysis can be performed on bits of bone, teeth, and strands of hair without the root sheath, where nuclear DNA testing would be impossible. Since mtDNA is more sturdy, it can still be found in materials that are dozens, if not hundreds, of years old (Hansen, 1998: 67). For example, “the Armed Forces Institute of Pathology uses mtDNA to identify the bones of servicemen missing in Southeast Asia” (Hansen, 1998: 67). Mitochondrial DNA was also used to show that the remains of Jesse James were genetically linked to known maternal-line descendants (Hansen, 1998: 67). Another advantage mtDNA has over nuclear DNA is that each cell contains a significantly larger supply of mtDNA than nuclear DNA, which may be of concern with small sample sizes (Hansen, 1998: 67). Each cell contains several thousand copies of mtDNA compared with only two copies of nuclear DNA. Finally, mtDNA does not undergo recombination as nuclear DNA does. However, “the low fidelity of mtDNA polymerase and the apparent lack of mtDNA repair mechanisms have led to a higher rate of mutation in the mitochondrial DNA genome” (Wilson et al., 1993: 68). “Some regions of the mtDNA genome appear to be evolving at five to ten times the rate of single copy nuclear genes” (Wilson et al., 1993: 68). These zones of relatively high mutability are still well conserved. These regions are of forensic interest, “due to their ability to assist in the differentiation of individuals in the human population” (Wilson et al., 1993: 68). It has been estimated that “among Caucasians there is an average of one nucleotide difference every 100 bases (1%) in the most variable regions of mtDNA, although this average is higher in individuals of African descent, being approximately 2.3%” (Wilson et al., 1993: 68).

However, mtDNA is not without its drawbacks. Mitochondrial DNA analysis is
"expensive, time-consuming, and requires a highly sterile environment and a special level of expertise in order to perform" (Hansen, 1998: 67). In fact, Canadian laboratories do not even perform this type of fingerprinting. Presently, there are only six laboratories worldwide which are able to accommodate requests for mtDNA examinations. However, there are many laboratories that are able to perform mtDNA analysis on animal samples. These results have been used in numerous animal poaching cases (Davidson, 2001).

In addition, the population database against which mtDNA samples are compared only consists of about 2,000 samples, which may be too small to draw meaningful conclusions (Hansen, 1998: 67). By contrast, the population database for nuclear DNA contains millions of samples. One of the largest mitochondrial DNA databases in the world consists of approximately one thousand sequences obtained from anonymous donors. As of July 1999, the FBI Laboratory, the Armed Forces DNA Identification Laboratory, and other laboratories have collaborated to compile a mtDNA population database containing the sequences from HV1 and HV2. The database is called the SWGDAM (Scientific Working Group on DNA Analysis Methods) database and contains sequences from four main racial groups—Caucasians, Africans, Hispanics, and Asians (Isenberg and Moore, 1999: 5). The samples were obtained from “paternity-testing laboratories, blood banks, and academic groups studying ethnic populations” (Isenberg and Moore, 1999: 5). The database contains 2,426 sequences from unrelated individuals and is constantly growing (Isenberg and Moore, 1999: 5). However, it is expected that the level of discrimination with mtDNA will never reach that reported using nuclear DNA analysis.

Finally, "unlike nuclear DNA, an individual’s mtDNA sequence can vary from time to time and from tissue to tissue, and maternal relatives have been found to not always have
the same mtDNA” (Hansen, 1998: 67). This phenomenon is termed heteroplasmy. Heteroplasmy has been recently identified within the control region of the mtDNA genome. For example, heteroplasmy was observed at position 16,169 of the mtDNA control region in the remains of Tsar Nicholas II of Russia (Wilson et al., 1997: 167). The remains of the Tsar differed from the mtDNA sequence of his maternal descendants by one nucleotide of the 740 sequenced (Wilson et al., 1997: 167). However, the sequencing of the mtDNA of the Tsar’s brother, the Grand Duke of Russia Georgij Romanov, showed complete mtDNA sequence accordance, including the heteroplasmic mixture at position 16,169 (Wilson et al., 1997: 167). Therefore, the presence of heteroplasmy in both the Tsar and Georgij increased the probability that the remains were in fact those of Tsar Nicholas II (Wilson et al., 1997: 167). However, while in this case heteroplasmy assisted the forensic scientist’s interpretation of the mtDNA evidence, a discrepancy in two mtDNA sequences may lead to a false exclusion. In addition, heteroplasmy has been observed between two samples of the same individual (e.g. different sequences in a sample of hair and a sample of blood).

Proponents of mtDNA argue that “any problems have been addressed by the four years of solid research, testing, validation, and publication in peer reviewed scientific journals” (Hansen, 1998: 68). “FBI officials articulate that they always explain the differences between mitochondrial and nuclear DNA to triers of fact and always make the limitations of mtDNA testing perfectly clear” (Hansen, 1998: 68). Furthermore, many of the objections being raised about mtDNA testing today are the same ones that were made when nuclear DNA was first introduced about ten years ago.
Forensic Analysis of mtDNA

Design of mtDNA Genome and Forensic Implications

The human mtDNA genome is 16,569 bases in length and consists of two general regions—the coding region, which is "responsible for the production of various biological molecules involved in the process of energy production in the cell," and the control region, which is "responsible for the regulation of the mtDNA molecule" (Isenberg and Moore, 1999: 2). There are two main regions within the control region which are highly variable. The first is called Hypervariable Region 1 (HVI), which is 342 base pairs long, and the second is called Hypervariable Region 2 (HVII), which is 268 base pairs in length (Isenberg and Moore, 1999: 2). In forensic mtDNA casework, 610 base pairs are usually sequenced and compared with a reference DNA sequence referred to as the "Anderson Sequence" after its founder (Isenberg and Moore, 1999: 2). "Each base pair is assigned a number and deviations are recorded as the number of the position demonstrating a difference and a letter designation of the different base" (Isenberg and Moore, 1999: 2). For example, an alteration from A to G at position 263 would be recorded as 263 G (Isenberg and Moore, 1999: 2).

Two unrelated individuals will generally have six to twelve differences in their mtDNA pattern using current identification protocols (Fourney, 1998: 47). However, these two people may have the exact same mtDNA sequence. It has been reported that the more common patterns are found in greater than 4.5% of all individuals (Fourney, 1998: 48). These variations are detected using a sophisticated scientific procedure which deciphers each of the individual chemical components of the DNA molecule in a particular region of the mitochondrial chromosome (Fourney, 1998: 48). The rarity of the pattern is then determined
by comparison with a database of mtDNA sequences. If the pattern has been previously reported, the occurrence is the number of times observed over the entire number of people in the database. As the database grows, the frequency estimation will fluctuate.

**Analytic Approach to mtDNA**

The examination of mtDNA is both "rigorous and labour-intensive" (Isenberg and Moore, 1999: 2). The steps include primary visual analysis, sample preparation, DNA extraction, polymerase chain reaction (PCR) amplification, post-amplification quantification of the DNA, automated DNA sequencing, and data analysis. The first step in mtDNA analysis of hair, for example, is the microscopic comparison of the evidence with a sample population of reference hairs (Isenberg and Moore, 1999: 2). If the hair is found to be microscopically uncharacteristic when compared to the known standard, then analysis is stopped at this point. The hair is cleaned prior to sequencing to remove contaminants, which is important to ensure that the sequence obtained originates from the sample and not from exogenous human DNA (Isenberg and Moore, 1999: 3). For hair, the cleaning process includes a detergent treatment in an ultrasonic water bath. Then, the sample is placed in an extraction solution and ground with a mortar and pestle (Isenberg and Moore, 1999: 3). The third step involves DNA extraction. The solution from step two is exposed to organic chemicals that separate the DNA from other biological molecules and then centrifuged (Isenberg and Moore, 1999: 3). The DNA remains soluble in the top layer, which is then filtered and purified for amplification. The polymerase chain reaction is used to amplify the small amount of DNA. The two strands are separated and a new strand is made using an enzyme that copies the DNA molecule (Isenberg and Moore, 1999: 3). This process is
repeated until many million copies are made. The fifth step is post-amplification purification and quantification. The DNA copies are purified using filtration and then quantified using capillary electrophoresis, which compares the amount of DNA in the PCR product to a known DNA standard to determine the concentration (Isenberg and Moore, 1999: 4). Control samples are also used to demonstrate whether or not the amplification was successful. The final step is sequencing using Sanger’s Method, in which the products are separated using gel electrophoresis based on the length of the fragments (Isenberg and Moore, 1999: 4).

Mitochondrial DNA sequences are generated by computer software and edited by a DNA examiner. Since the human body contains trillions of cells, each with thousands of copies of the mtDNA genome, complete homoplasmy (e.g. the same sequence) for each of the mtDNA molecules would be unrealistic (Isenberg and Moore, 1999: 5). Therefore, heteroplasmy is expected. Heteroplasmy can be defined as “the occurrence of more than one type at a particular position in a DNA sequence” (Isenberg and Moore, 1999: 5). Heteroplasmy was first observed in mtDNA sequences in 1994 by the Forensic Science Service (FSS) (mentioned later in the Romanov family case) (Isenberg and Moore, 1999: 5). Recent improvements in detection have led to the realization that levels of heteroplasmy may not always be the same in various tissues. For example, a hair may contain mostly C at a particular position in the genome while a blood sample may contain equal amounts of C and T at the same position (Isenberg and Moore, 1999: 5).

Quality Assurance in mtDNA Testing

Since mtDNA analysis is extremely sensitive and the risk of contamination of the
samples is high, it is important that the laboratories that test mtDNA be properly accredited. For example, the FBI Laboratory is accredited by the American Society of Crime Lab Directors/Lab Accreditation Board (ASCLD/LAB) and the DNA Analysis Units follow the quality assurance guidelines established by the SWGDAM, the DNA Advisory Board (DAB), and the ASCLD/LAB (Isenberg and Moore, 1999: 6-7). Two open and external proficiency tests are performed annually for all unit personnel. As well, everyday precautions are taken by laboratory staff such as wearing gloves and lab coats, and using laminar flow or deadspace hoods and aerosol-resistant pipette tips (Isenberg and Moore, 1999: 7). Additionally, only one item of evidence is opened at a time to prevent cross-contamination, questioned samples are always analyzed before known samples, the pre-amplification process is separated from the post-amplification process, all reagents, work spaces, and instruments are subjected to daily isopropanol, 10% bleach washings, and UV radiation, and all instruments such as the thermal cycler, capillary electrophoresis machine, the sequencing instrument, water baths, thermometers, freezers, fridges, and pipettes are calibrated frequently (Isenberg and Moore, 1999: 7). Finally, blank samples are tested as well as both positive and negative controls, the amount of amplified DNA in the blanks or negative controls cannot exceed 10% or the sample will not be tested any further, mtDNA sequences from all laboratory personnel are kept on file, and each sequence is read by two different examiners (Isenberg and Moore, 1999: 7).

**Few and Far Between: mtDNA Laboratories**

Presently there are six laboratories worldwide which are able to accommodate requests for mtDNA examinations. The first and most expensive is the Forensic Science
Service Laboratory located in Birmingham, England. Their analysis fees per questioned sample are approximately $8,500 and $7,300 per reference sample. The turnaround time is between three and six months (Pearce, 1999: 1). The second laboratory is the Department of National Defence Laboratory in Washington, D.C., which has little background in forensic cases as the majority of their experience deals with identifying the war dead (Pearce, 1999: 1-2). The FBI has a laboratory in Quantico, Virginia which does not charge a fee for analysis, although they are entirely overworked (Pearce, 1999: 2). There are two private laboratories in the United States that are able to sequence mtDNA. The first, called Mitotyping Technologies, was opened in April 1999, and is located at Penn State University (Pearce, 1999: 2). Their fees are $2,500 for questioned samples and $1,500 for reference samples and the analysis takes three months to complete (Pearce, 1999: 2). The second laboratory is called LABCORP and is located in Research Triangle Park, North Carolina (Pearce, 1999: 2). Their fees are a flat $975 per sample and their turnaround time is approximately three months (Pearce, 1999: 2). Finally, there is a small laboratory in Virginia called BODE Technologies, Inc. which charges $1,500 per sample (Pearce, 1999: 2).
Chapter II: Mitochondrial DNA Casework

The Romanovs

In 1917, the Romanov imperial family was executed by the Bolsheveks ending their 300-year rule (the Czar Nicholas II, his wife, four daughters, and a son were herded into a basement and shot) (see Figure IV) (McCord, 5).

Figure IV: Romanov Family Picture (www.geocities.com/Vienna/9443/)

Supposedly, the bodies were placed in a truck to be buried in a mine shaft but the truck encountered problems and they were buried in a shallow grave instead. However, this story was never ever verified. In 1991, the putative remains were located and exhumed. The bodies showed evidence of violence with bullet holes and bayonet marks. Facial reconstruction was difficult due to extensive damage, but tentatively the remains were identified as the Czar, Alexandra-Czarina, three children, three servants, and the family
doctor (McCord, 5). However, two bodies were missing (the son and one daughter).

In 1992, Peter Gill of the Forensic Science Service, along with Pavel Ivanov, a Russian DNA fingerprinting expert, were asked if the identity of the bodies could be confirmed using mtDNA (McCord, 5). Nuclear and mitochondrial DNA analyses were performed on the nine bone samples. It was discovered that five of the bodies were related and three were female siblings (Davies, 1996: 340). Furthermore, a sample of maternally inherited mtDNA suspected to belong to the Czar’s wife, Alexandra, matched a sample provided by her grandnephew, the Duke of Edinburgh (Prince Philip). However, finding a reference sample for the Czar was almost impossible until two distant maternal relatives came forward (Davies, 1996: 340). It was determined that the mtDNA sequences of the Czar’s two relatives were the same, although due to heteroplasmy, the estimated probability of the remains belonging to the Czar was 98.5% (Davies, 1996: 340). More proof was demanded by the Russian Orthodox Church, so in 1994, the remains of Grand Duke Georgij (Nicholas’ brother) were exhumed for comparison. The same heteroplasmy evident in the Czar’s mtDNA was exhibited in the Grand Duke’s mtDNA, which was the first time that heteroplasmy had been applied in human identification (Davies, 1996: 340). Combining the DNA sequence identity, the rarity of heteroplasmy, and the established link between Alexandria and the Duke of Edinburgh, it was calculated that the likelihood ratio for the remains’ authenticity was in excess of 100 million (Davies, 1996: 340). Unfortunately, the two bodies of the two youngest children, Alexi and Anastasia, have never been found. It is thought that the remains were burned.

The mystery surrounding Anastasia’s fate after the Russian Revolution is one of the biggest mysteries of the 20th Century (Torretta, 1997: 1). Many stories about her, and
recently the 20th Century Fox movie, entitled Anastasia, have brought the mystery alive again. Experts in DNA testing cannot agree if she is, in fact, still missing. There were rumors she escaped to the United States and changed her name to Anna Anderson. In 1995, two different samples (hair and intestine) said to have come from Anna Anderson were separately gathered and analyzed by three different laboratories (Stoneking et. al, 1995: 9). The results, all in agreement, “strongly suggests that the samples came from the same person (Anna Anderson); the contention that the sample came from another (unknown) individual seems highly unlikely” (Stoneking et. al, 1995: 9). However, the samples could not be connected with a maternal relative of the Tsarina or His Royal Highness Prince Philip (Stoneking et. al, 1995: 9). It was discovered that the real identity of Anna Anderson was actually Franziska Schanzkowska—born in approximately 1896 and lived in Pomerania (in the north of Germany adjacent to the Polish border). During the First World War, while working in a munitions factory in Berlin, she was badly injured in an explosion. Afterward, she was admitted to two different mental hospitals, but disappeared in 1920, about the same time Anna Anderson appeared and claimed to be the Royal Duchess Anastasia (Stoneking et. al, 1995: 9). This information was ascertained by matching tissue samples of Anna Anderson with blood samples of Carl Maucher, a maternal relative of Franziska Schanzkowska (Stoneking et. al, 1995: 9). Recent forensic studies by United States’ scientists suggest that the found remains do not include Anastasia and that the remains believed by some to be those of Anastasia are, in fact, her sister, Tatiana (France et al., 2000: 221). However, Russian forensic scientists, dispute this claim and argue that the remains are those of Anastasia (Nikitin, 2000: 221). Therefore, the mystery of Anastasia continues today.
United States mtDNA Cases

State v. Ware

In State v. Ware 1999 Tenn. Crim. App. Lexis 370, the defendant, Paul Ware, was indicted in 1994 for felony murder and multiple counts of rape of a child with the State filing notice of intent to seek the death penalty (Lexis at para. 1). The trier of fact found him guilty of felony murder and two counts of child rape, and he was sentenced to life without parole for the felony murder. At a subsequent sentencing hearing, the trial court imposed concurrent twenty-five year sentences for the child rape convictions and ordered that the twenty-five year sentences be served consecutively to the sentence of life without the possibility of parole (Lexis at para. 1).

State v. Council

The case of State v. Donney S. Council 335 S.C. 1; 515 S.E. 2d 508; 1999 S.C. Lexis 76, is the second case in the United States in which mtDNA evidence was used. The defendant was indicted for murder, kidnapping, administering poison, grand larceny of a vehicle, burglary, larceny, and two counts of criminal sexual conduct in the first degree, found guilty of all charges, and sentenced to life in prison (Findlaw at p.2). On October 8, 1992, Mrs. Elizabeth Gatti was found murdered in her ransacked house, under a bedspread in the basement, hogtied with white cord and layers of duct tape over her head (Findlaw at p. 3). Her clothes were ripped, the crotch of her underwear was missing, and Mrs. Gatti had been sexually assaulted (Findlaw at p. 3). On October 11, Mrs. Gatti’s car was found near the apartment complex of the defendant, who was later arrested on October 12 (Findlaw at p. 5). The defendant admitted to having sexual contact with the victim but denied having
murdered her. Evidence in this case included: “a shoe print from the victim’s house which was matched to the defendant, residue on a chair which was matched to debris on the defendant’s shoes, fingerprints from the car and items in the car belonged to the defendant, hair consistent with those found in the house which was matched to the defendant, semen from a tissue in the house which was matched to the defendant, items belonging to Mrs. Gatti were found in the defendant’s girlfriend’s apartment, the defendant forged three of Mrs. Gatti’s cheques, and Mrs. Gatti’s newspaper carrier positively identified the defendant as leaving Mrs. Gatti’s residence” (Findlaw at p. 5).

**State v. Underwood**

In *State v. Underwood* 518 S.E. 2d; 1999 N.C. App. Lexis 856, the defendant was convicted of first degree murder and first degree kidnapping (Findlaw at p. 1). On January 7, 1994, the body of Viktor Gunnarrson was found. The victim had been dead for about two weeks and had expired from a gunshot wound to the head (Findlaw at p. 1). In the defendant’s car, scratches were found inside the trunk and a footprint was discovered on the underside of the trunk lid (Findlaw at p. 2). In addition, the trunk mat was removed and hairs were found on the trunk mat, even after the carpets had been shampooed (Findlaw at p. 2). Two FBI experts, Hamlin and Dizinno, testified that the hairs were consistent with Gunnarrson’s hairs and that the DNA sequence from the hair and the known blood was identical (Findlaw at p. 2).

**State v. Scott**

In *State v. Scott* [1999] TN-QL 1182, the defendant, Randall Scott, was convicted of the rape of a child and aggravated sexual battery and sentenced to twenty-five and ten-year consecutive sentences (Quicklaw at para. 6). The crime occurred on April 17, 1995, when...
the nine-year-old victim walked to a corner store in order to buy earrings. The defendant grabbed her from behind, forced her into an alley behind a shed, pushed her to the ground, pulled her shorts and underpants down, touched her vaginal area with his finger, and then penetrated her anally with his penis (Quicklaw at para. 10). Forensic evidence found included three hairs on the victim's genital area and bodily fluids on the accused's clothing (Quicklaw at para. 18).

**United States Case Analysis**

There are three standards of admissibility governing scientific evidence in the United States—*Frye v. United States, Federal Rules of Evidence,* and *Daubert v. Merrell Dow Pharmaceuticals.* The *Frye* standard is the most rigid of the three since it requires general acceptance in a particular scientific field (*Scott at para. 54*). The *Federal Rules of Evidence* "give more discretion to the court by stating that the evidence must be relevant and reliable, and that evidence considered more probative than prejudicial will be admitted" (*Scott at para. 55*). Finally, in *Daubert,* the United States Supreme Court declared that general acceptance is only one of several factors to be considered in assessing reliability and relevance. Other factors include whether "the theory or technique has been tested, the extent of peer review and publication, a known error rate, and the existence and maintenance of controls and standards" (*Council at p. 26 and 28*). In Tennessee, which was where *Ware* was tried, new scientific evidence is generally admissible without expert testimony provided it conforms to the *Federal Rules of Evidence.* Tennessee Rule of Evidence 702—*Testimony by Experts,* states that "if scientific, technical, or other specialized knowledge will substantially assist the trier of fact to understand the evidence or to determine a fact in issue, a witness qualified as
an expert by knowledge, skill, experience, training, or education may testify in the form of an opinion or otherwise" (Scott at para. 57). This rule was interpreted in Ware to mean that the "rules necessarily require a determination as to the scientific validity or reliability of the evidence ... but there is no requirement in the rule that it be generally accepted" (Scott at para. 60). Although Ware was the first case in which mtDNA evidence was used, the science had been researched since June of 1996 by the FBI. Therefore, since this case was tried in Tennessee, there was no need for an admissibility hearing before this new scientific evidence was admitted to trial, which made it the best possible place to get mtDNA accepted by the courts so it could be used in other following cases. At the time of the trial, the database that was used to compare the samples contained a total of 742 samples (presently the database contains about two thousand samples).

At the Ware trial, it was revealed that there were certain hairs found on and inside the child’s body. Dr. King recovered, during the autopsy, a reddish hair which was stuck to the victim’s lip, a dark brown body hair which was “partly touching ... the mucosa of the rectum and partly touching the skin of the anus,” and a reddish pubic hair from the victim's pharynx (Ware at para. 20). Special Agent Chris Hopkins of the FBI Hair and Fibers Unit concluded that all hairs, the hairs from the victim’s pharynx and those from the sheet, were “consistent with originating from the defendant” (Ware at para. 26). However, he also testified that hair comparison is “not a means of personal positive identification,” and therefore he could not state conclusively whether the hairs belonged to the defendant (Ware at para. 27).

In order to substantiate the evidence, mtDNA analysis was performed. Special Agent Mark Wilson testified that the hair recovered from the victim’s throat and a hair from the bed sheet were compared with saliva from the defendant (Ware at para. 43). Analysis of the
samples determined that all three samples shared a common sequence. Testing also
determined that the sequence in the three samples did not match that of blood taken from the
victim (Ware at para. 43). On cross-examination, Wilson clarified the findings; “he explained that the two hairs shared 600 bases, while the victim’s blood sample shared 593 bases with the other samples” (Ware at para. 44). He stated that “the average number of differences between any two Caucasian individuals is approximately six” (Ware at para. 44).

While he maintained that the sample hairs were compatible with having originated from the defendant, “he also stated that the tests could not show that the sample hairs belonged to the defendant to the exclusion of all others” (Ware at para. 44). In reaching his conclusions, “Wilson did not assign a frequency rate to the results of the mtDNA tests which were performed in this case, stating instead only that the sequence had not before been observed in the FBI’s database of 742 individuals” (Ware at para. 44). Wilson testified:

All I’m saying is we have a database of a certain size, and this particular sequence has not been observed before. I am not saying that it’s a particular frequency, one over this or that, because it cannot be expressed that way because the database is not large enough at the present, in its present form, present size to be able to assign a frequency, you know, like one percent or whatever. This event would have to be observed many more times in order to assign it a frequency, so what we do is state a fact (Ware at para. 44).

In the appeal launched by the defendant, it was alleged that the trial court improperly allowed testimony regarding mtDNA that led to an unfair trial (Ware at para. 2). The trial court denied the defence’s motion to suppress the evidence because it was not scientifically reliable for use in the courtroom. In an affidavit filed by the defence, Dr. William Shields, a geneticist, stated that reports published by the FBI in the International Journal of Legal Studies to legitimate their studies does not have a sufficiently wide distribution (only 25 libraries in the United States subscribe to it) to prove that mtDNA evidence is reliable and
ready for the courtroom (Ware at para. 50). In addition, Dr. Shields contended that the sample sizes used by the FBI were inadequate and not sufficiently tested within the scientific community. He stated, “It is my opinion that mtDNA typing as proposed by the FBI, is not yet sufficiently reliable to be scientifically reliable... The major problem is that critical pieces of the validation process have yet to be done or have been done with insufficient sample sizes to be statistically reliable” (Ware at para. 46-47). Shields stated that at the time of the hearing, he was a scholar in residence at the University of Virginia law school, where he guest-lectured and co-taught classes in advanced evidence, “in particular, [concerning] the issues surrounding scientific evidence, both in toxic torts and DNA typing” (Ware at para. 47). He also stated that he had “been involved since 1990 in exploring the use of DNA typing in forensic situations” (Ware at para. 48). Unfortunately, Dr. Shields was not able to testify at the initial trial, only at the motion for a second trial.

In this case, a great deal of circumstantial evidence was introduced. Despite some evidence suggesting that the defendant may not have committed the crime, there was clearly substantial evidence presented at trial indicating that the defendant did commit the crime: most obvious is evidence presented that the defendant and the victim were discovered nude together in a room locked from the inside, although there is a question as to whether the crime was committed by another man (the boyfriend of the owner of the house) who was known to frequent the house where the victim and her mother were staying. Additionally, although the two medical experts who testified at trial differed in their estimations of the victim’s time of death, Dr. King concluded that the victim had been dead “one to two hours or less,” a time period which corresponds with the time that the defendant was present in the home (Ware at para. 33). Finally, FBI agent testified based on hair comparison that hair
found on the bed sheet where the victim slept and the hairs inside the victim’s pharynx all “were consistent with originating from the defendant” (Ware at para. 27). He declared that “two factors contributed to the decision on which hairs to type in the Ware case: the probative value of the hairs, and the suitability for microscopic comparison” (Ware at para. 45). Wilson explained,

A crucial aspect of these investigations . . . is the question of how probative the evidence may be . . . . In the Ware case, information provided to the FBI Laboratory indicated that the victim . . . was found naked in an unkempt laundry room floor. Her body was then moved by family members and medical personnel prior to an autopsy. Accordingly, the finding of extraneous hairs would be expected to be found on an unkempt laundry room floor. However, the finding of a pubic hair in the victim’s throat can be attributed to contact that is not merely casual. Moreover, the fact that the cause of death was asphyxiation adds additional probative value to the discovery of the foreign pubic hair in the throat. . . . [The] finding [of pubic hairs on the sheet] is probative because the victim was . . . asleep on the bed prior to the attack . . . (Ware at para. 45-46).

Ultimately, at trial, Wilson testified that “mtDNA is extensively studied. . . . It's very well understood and characterized” (Ware at para. 53). He also testified that mtDNA is “widely used” to “identify the remains of servicemen that have been killed in Vietnam or Korea” (Ware at para. 53). “This testimony indicates that mitochondrial DNA analysis also meets the Frye standard of being generally accepted in its field” (Ware at para. 54). However, “even assuming that the DNA evidence was improperly admitted into evidence, the court was convinced that any error caused by the admission of the evidence was harmless” (Ware at para. 54). Regardless of its accuracy or inaccuracy, Wilson’s testimony did not confer a substantial amount of comprehensible information to the jury. Without a frequency rate or some similar interpretation of the test results, the testimony did not provide a strong basis for scientific conclusion by a layperson. The only result that an individual untrained in the analysis of DNA could reach after hearing Wilson testify is that the common DNA sequence shared by the hairs tested and the defendant’s saliva had never before been
noted in the 742 individuals that comprised the FBI’s then-current database (*Ware* at para. 55). Moreover, other scientific evidence presented at trial which was more clearly interpreted for the jury (including testimony by Hopkins, the hair comparison expert), although also inconclusive, points to the defendant as the donor of the hairs at issue (*Ware* at para. 55). Thus, based upon all circumstances, the court concluded that this issue did not result in reversible error. The appeal was subsequently dismissed.

In the second mtDNA case in the United States, *Council*, two expert witnesses were called during the trial: John Ortuno, a trace evidence examiner for the FBI, who testified that pubic hairs found at the scene were consistent with the defendant, and Joseph Dizinno of the FBI (*Council* at p. 22). Dizinno testified that he had extensive training in hair and fiber analysis and mtDNA analysis. In addition, “the science of mtDNA analysis had been used since 1981, with more than 600 papers written about the technique” (*Council* at p. 23). It was characterized as a recognized methodology and a reliable science (*Council* at p. 23). In this case, mtDNA analysis was used to confirm the subjective comparison of the hairs. A database of 742 sequences, “containing 319 African-American sequences, was used to characterize the findings in which a match was found between unrelated Caucasians but no match was found between African-Americans” (*Council* at p. 25). The experts testified that “most probably” the hairs were those of the defendant (*Council* at p. 25).

The defence appealed the case based on the contention that the trial court erred in admitting the mtDNA evidence. However, this type of evidence had already been admitted in the preceding case of *Ware* without an admissibility hearing, which effectively set a precedent. However, “the defence argued that according to the standard in *Frye*, the court can only hold that scientific evidence is reliable and admissible when it has attained general
acceptance of the scientific community" (Council at p. 26). The Court of South Carolina had never adopted the Frye standard. Prior to 1990, the standard in South Carolina was the "degree to which the trier of fact must accept, on faith, the scientific hypotheses not capable of proof or disproof in court and not even generally accepted outside the courtroom (State v. Jones), which is a more liberal interpretation than Frye" (Council at p. 27). Finally, the defence maintained that they had no opportunity to rebut the scientific evidence since the results were delayed and they had only received them the night before the trial, which did not allow enough time to find an expert witness (Council at p. 27). In my opinion, this is definitely a valid point because the defence should have an adequate amount of time (not overnight) in which to gather materials to present the most effective defence possible. Therefore, the trial should have been postponed in order for the defence to find an expert witness. Even better, the defence should have had an expert witness waiting to examine the results so that the trial would only have to had been delayed for a short period of time. However, the court disagreed with the defence since they did have a chance to speak with Dizinno and to cross-examine him. As well, the defence did demonstrate flaws in the evidence presented by the prosecution and did encompass points that their own expert would have covered. Therefore, the appeal was dismissed on this point.

The third case presented is that of Underwood. In this case, "the defence argued that the mtDNA evidence presented was not scientifically reliable and the reasoning and methodology behind the science had not been properly applied to the facts of the case" (Underwood at p. 5). However, this point was already argued in both Ware and Council. This case was tried in North Carolina, and according to the North Carolina Rules of Evidence, "if scientific, technical, or other specialized knowledge will assist the trier of fact
to understand the evidence or to determine a fact in issue, a witness qualified as an expert by knowledge, skill, experience, training, or education, may testify thereto in the form of opinion” (Underwood at p. 5). Therefore, “the admissibility of opinion evidence is based on whether the witness, through study or experience, has acquired skill that is better qualified than the jury to form an opinion on the subject matter to which his testimony applies (State v. Mitchell)” (Underwood at p. 5). The expert in this case, Dizinno, is chief of the FBI DNA Analysis Unit II, holds a B.Sc. degree from Notre Dame and a D.D.S. degree from Ohio State, had previously testified on mtDNA analysis, and was formerly accepted as an expert witness (in the Council case) (Underwood at p. 6). The appellant argued that the expert was of no assistance to the jury, which the court did not accept. The court decided that the source of the hair was a crucial fact and that the evidence was relevant since it had the tendency to make a fact of consequence “more probable or less probable than it would be without the evidence” (Underwood at p. 6). However, even though the evidence could not definitively eliminate the victim, it was relevant to show it was more probable. The appellant maintained that the evidence was unreliable, and that it can only be admissible if the evidence is scientifically reliable. In order to test the reliability of mtDNA evidence, “the court again looked at testing, peer review and publication, submission to the scrutiny of the scientific community, the known or potential rate of error, and the general acceptance in a relevant scientific community” (Underwood at p. 6). The court found that the evidence was relevant since it “has any logical tendency however slight to prove the fact at issue in the case” (Underwood at p. 6).

In this case, the victim was not excluded and in the database the same sequence had been seen about one in ten times. It is highly unlikely that two sequences would match in
both microscopic and mtDNA sequence (Underwood at p. 6). However, “the appellant claimed that the population database, which contained more than 1,000 samples, was too small to draw meaningful conclusions, when compared with nuclear DNA analysis whose database contains millions of samples” (Underwood at p. 7). Regardless, mtDNA had been researched for more than four years, was widely accepted in evolutionary genetic studies, and had been used in court in six other states (Underwood at p. 7). Although mtDNA does not give proof of identification, it is reliable enough to warrant its admissibility into evidence (Underwood at p. 7). Therefore, the trial judge did not err and the defendant was deemed to have received a fair trial. All of these issues had been dealt with within the two previous cases. Therefore, there was essentially nothing to argue by the defence.

The fourth case in the United States to use mtDNA evidence was Scott. On appeal, the defence argued that the trial court denied the defence expert assistance (Scott at para. 8). This is the same argument that was used and failed in the previous case of Council. However, this case is more complicated due to the fact that the evidence was analyzed by two different laboratories that achieved two different sets of results. Initially, analysis on the hairs was performed by Anita Matthews, a forensic scientist employed by the Laboratory Corporation of America (LABCORP) (Scott at para. 20). Ms. Matthews concluded that “the blood on the defendant’s shorts was consistent with the victim’s DNA and that the victim could not be excluded as the donor of the blood found on the defendant’s shirt” (Scott at para. 20). Polymerase chain reaction analysis of the hairs was not consistent with that of the defendant, although it was acknowledged that LABCORP does not wash hairs before testing them and that some components of the DNA testing are considered “new and emerging technology” (Scott at para. 20).
Mitochondrial DNA analysis was also performed by the FBI Forensic Laboratory under the direction of Special Agent Mark Wilson in June, 1996. During the procedure, the hairs were washed due to the fact that if the washing is not completed, "sometimes the type that you would get at the end was actually from the scientist and not from the hair type" (Scott at para. 21). It was found that the hair taken from the rectum of the victim matched the defendant.

Therefore, the defence claimed that since two different laboratories had used this relatively new procedure, an expert was needed to interpret why two disparate results were achieved (Scott at para. 36). The trial court ruled that the appointment of a DNA expert fell in "the category of a luxurious defense, not an adequate one" (Scott at para. 39). Moreover, the trial court found that an expert was not necessary for several reasons, but mainly that the DNA was not "all that crucial" to the state's case against the defendant (there was strong circumstantial evidence and the victim had positively identified the defendant) (Scott at para. 41). The appeal court agreed with the trial court that little else could have been accomplished by an expert's guidance in recognizing weaknesses in the DNA technology and the defence did not establish a "particularized need" for an expert in preparing defence counsel for trial (Scott at para. 51). In summary, there was strong evidence of guilt without the DNA evidence and the DNA evidence presented was not entirely persuasive in view of the fact that LABCORP's testing of the hair samples was clearly erroneous (Scott at para. 51).

Finally, the defence also argued that the trial court erred by failing to conduct a pretrial hearing on the admissibility of mtDNA evidence, which was the same strategy used in the Ware case. The trial court relied on the following factors in determining whether the scientific evidence was reliable or valid: "(1) whether the scientific evidence had been tested
and the methodology with which it has been tested; (2) whether the evidence has been subjected to peer review or publication; (3) whether a potential rate of error is known; (4) whether the evidence is generally accepted in the scientific community; and (5) whether the expert’s research in the field has been conducted independent of litigation” (Scott at para. 61). The appeal court established that the trial court did not err by refusing to hold a hearing on the admissibility of mtDNA evidence based on several facts: “the DNA was relevant because it suggested that the defendant was the assailant (Scott at para. 65), Special Agent Wilson was appropriately qualified as an expert based on his certification as a mtDNA analyst, degree, and numerous publications (Scott at para. 65), and DNA evidence is statutorily regarded as trustworthy and reliable” (Scott at para. 68). As well, mtDNA evidence had been used three times previous to this case, which demonstrates the degree of admissibility the courts had afforded this type of evidence.

**State v. MacDonald: The Use of mtDNA by the Defence Team**

Mitochondrial DNA evidence has been utilized in United States courts six times since September 1996, with six convictions in these six trials. However, in all of these cases the evidence has been used by the prosecution. Mitochondrial DNA evidence will be used by the defence for the first time ever in the case of Jeffrey MacDonald (Cohen, 1999: 1). This former Army surgeon was convicted in 1979 of the killing of his wife and daughters (Cohen, 1999: 1). Hair found under the fingernails of his daughter which has been preserved over the years will be mtDNA tested in order to try to prove MacDonald’s innocence (Cohen, 1999: 6). The case was re-opened in 1999 and the results have yet to have been made public, though it is probable that mtDNA could be used to exonerate someone accused of a crime,
as nuclear DNA has been used for this very purpose many times (for example Guy Paul Morin and David Milgaard).

**R. v. Murrin: The One and Only Canadian mtDNA Case**

The first case in Canada to use mtDNA evidence is *R. v. Murrin* [1999] B.C.J. No. 2715. However, there was mention of the technique in *R. v. Feeney* [1999] B.C.J. No. 688 DRS 99-07645. In *Feeney*, the defence argued “that there was some evidence that mtDNA could be gathered by a person giving an ink fingerprint in the conventional method used by police” (*Feeney* at para 59). However, the court found that there was simply no evidentiary basis that this procedure had been used in this case, although it is possible in theory (which will be discussed later in this thesis) (*Feeney* at para 59). In *Murrin*, Shannon Murrin was charged with the first degree murder of Mindy Tran. Tran disappeared on August 17, 1994 and her body was recovered two months later in a shallow grave (*Quicklaw* at para 2). The Crown alleged that three hairs left by the killer at the crime scene contained the same mtDNA sequence as blood samples taken from Murrin (*Quicklaw* at para 3). The accused argued in the admissibility hearing that “in its present state of development, the difficulties and uncertainties render the technique [of mtDNA analysis] insufficiently reliable for consideration by the trier of fact” (*Quicklaw* at para 4). Three points were argued by the defence: “that the danger of contamination generates unreliable results, the recent discovery of a condition known as heteroplasmy provides further uncertainty, and the fundamental aspects of the science of mtDNA are still unknown” (there may be evidence of recombination and paternal inheritance) (*Quicklaw* at para 5-7).

The three hairs found in the victim’s underpants were sent to the Forensic Science
Service (FSS) Laboratory in Birmingham, England in February of 1998, in order to be analyzed due to the fact that laboratories in Canada viewed the technology as too expensive and time consuming to perform (Quicklaw at para 10). “The analysis was performed by Mr. John Bark who concluded that all three hairs had the same mtDNA sequence, the sequence matched that of the known blood samples taken from Murrin, and the sequence could not be that of the victim” (Quicklaw at para 11). Bark testified that his analysis provided “strong support” for the hypothesis that the three hairs originated from Murrin or any person maternally related to him and that the sequence found was uncommon (the sequence was observed once in a collection of 163 sequences from unrelated British Caucasians, once in a collection of 233 unrelated North American Caucasians, and twice in a larger collection of the “Miller Concordance” which consists of 4,774 sequences) (Quicklaw at para 13). “Analysis was also performed at the FBI Laboratory which concluded that 99.76% of the general population of Caucasians have sequences which differ from that found in the three hairs with 95% confidence” (Quicklaw at para 14). In order to determine if the technique of mtDNA analysis met the threshold level of reliability contemplated in Mohan, the Crown called Dr. Gillian Tully and Mr. John Bark of the FSS and Dr. Bruce Budowle and Mr. Mark Wilson of the FBI, while the defence called Dr. Donald Riley, Dr. William Shields, and Dr. Peter D’Eustachio (Quicklaw at para 16).

One of the issues in front of the court that the expert witnesses agreed upon as the most in need of analysis was the prospect of contamination, which may cause false conclusions (Quicklaw at para 90). Although “nuclear DNA testing also presents a risk of contamination, it is much less due to the mtDNA extraction process, which requires a greater number of cycles” (Quicklaw at para 92). In order to protect against contamination, “both
the FSS and FBI laboratories clean rooms and work surfaces regularly, containers are cleaned and sterilized with heat and ultraviolet light, gloves, disposable lab coats, caps, face masks, and dedicated shoes are worn (which is donned in a ‘clean room’ which has its own air supply), amplification is always carried out in a different room than extraction, a number of controls are used, and the samples are tested days apart” (Quicklaw at para 94-95). Due to these safeguards, the court found that the risk of contamination did not present a reason for rejecting the evidence of the novel scientific theory or technique (Quicklaw at para 108).

Additionally, it was initially assumed by scientists that every person has one, and only one, sequence in their mtDNA (Quicklaw at para 109). However, this is now acknowledged as false. Many observations have been made of mtDNA sequences taken from the same person that differ at one or more base positions, known as heteroplasmy (Quicklaw at para 110). There are “two types of heteroplasmy—length, when the number of bases in this string differs between mtDNA sequences taken from the same individual (more common), and sequence, when a particular base position has differing values in two mtDNA samples from the same person” (Quicklaw at para 111). The defence experts in this case spoke of the danger of “false exclusions” due to heteroplasmy (Quicklaw at para 116), although the court concluded that neither the discovery of heteroplasmy within the control region nor the alleged danger of “false exclusions” provides a reason for rejecting evidence of this novel scientific theory or technique (Quicklaw at para 121).

Lastly, “Mr. Wilson of the FBI conceded that fundamental tenets of the science are being questioned” (Quicklaw at para 122). “The assumption that there is no recombination between mitochondrial lineages and that the inheritance of mitochondria is clonal is now being questioned” (Quicklaw at para 122). Studies suggest that recombination and “paternal
leakage" are possibilities worthy of further consideration and investigation (Quicklaw at para 122).

In the cases presented above, none of the arguments of the defence were accepted on appeal; all of the defendants were found guilty of the crimes they were accused of. However, as will be seen in the circumstances surrounding the case of Shannon Murrin, this was not the case in Canada. Whether the verdict was a function of the inability of the jury to understand the science, or a question of the integrity of the police, will never be know.

United States and Canada Case Comparison

As scientific evidence becomes an increasingly valuable asset to the legal system, expert witnesses are being used to assist the legal fact-finder in understanding technical evidence. In the standard reference work, Cross on Evidence, the law's requirements of an expert (as quoted by A. Brownlie) are "that he shall give his evidence to the best of his ability on his special subject in a fair and unbiased manner, making full and frank disclosure so as to provide the court with the material necessary to enable it to come to a reasoned decision on the merits of the scientific issue" (as quoted in Gee, 1987: 310). For Smith and Wynne, "the scientific expert is an aid in factual discovery: an 'expert witness' is someone who, through specific training, knowledge, or experience is able to assist the legal system (a) in determining what facts are relevant to a particular case, and (b) by offering opinion about what the facts might mean for the reconstruction of a course of events or the outcome of a decision" (Smith and Wynne, 1989: 4).

Trial court judges have broad discretion in accepting an individual as an expert witness. According to Saferstein, "if a witness can establish to the satisfaction of a trial
judge that he or she possesses a particular skill, or has knowledge in a trade or profession that will aid the court in determining the truth of the matter at issue, that individual will be accepted as an expert witness” (Saferstein, 1998: 17). The court will usually consider knowledge acquired through experience, training, education, or a combination in sufficient grounds for qualification as an expert witness (Saferstein, 1998: 17). The competency of the witness may be established by having him or her cite educational degrees, participation in special courses, membership in professional societies, and any professional articles or books published (Saferstein, 1998: 17). Also important is the number of years of occupational experience in which the witness has engaged in areas related to the matter before court.

Unfortunately, the reliance on one person who is deemed to have an extraordinary amount of knowledge about a subject that most laypersons do not understand lends itself to inherent difficulties. Examples of these phenomena are many. Most often, this expertise is bought for a specific purpose. For example, in the O.J. Simpson case, both the defence and the prosecution located “the most authoritative scientific allies they could muster on the subject of DNA typing” (Jasanoff, 1995: 46). As well, persuasiveness, not only credentials, usually determines a witness’ worth (Jasanoff, 1995: 46). The difficult subject matter of science often leads to confusion of the jury and also the judge. Therefore, it is the duty of the expert witness to make the testimony as easy to understand as possible. This may lead the trier of fact to believe the expert with the most persuasive speaking manner, the most believable appearance, or the most professional image.

Furthermore, during a trial it is the attorney, not the expert, who determines what information is important and our judicial system demands that such determinations remain in the hands of those qualified to do so (Pipkin Jr., 1989: 108). Although the expert wishes
to be an impartial source of information, the client, the client’s attorney, and the expert’s natural competitive tendencies may pressure the expert to be a team player and help the client whenever possible (Pipkin Jr., 1989: 108). Therefore, some claim that experts have become nothing more than additional advocates for the attorneys who hire them since it is the responsibility of the attorneys to question the witnesses and see that the proper information is elicited from experts (Pipkin Jr., 1989: 107-108). Accordingly, it would be unreasonable to expect an attorney to elicit information which damages his client’s position, and the adversarial system calls for the attorney to attempt to keep out such information (Pipkin Jr., 1989: 108). The trial judge has broad discretion in determining which witnesses qualify as experts, what information or testimony will assist the jury, and whether such testimony should be admitted, which allows for a greater burden on those that oppose the evidence. Cross-examination and the use of opposing experts are supposed to reveal to the jury those witnesses whose information should be relied upon, but it is up to the attorneys to ask the proper questions so the jury can understand the principles or theories underlying key issues in the case (Pipkin Jr., 1989: 109). Therefore, questions from both sides should, in theory, clarify or correct any misinformation which is damaging (Pipkin Jr., 1989: 110). Attorneys are in a position to choose what the jury will hear, despite the fact than an incomplete picture may be presented (Pipkin Jr., 1989: 111). Attorneys also use experts to best present their theory of the case, and they obtain an expert whose viewpoint coincides with their own (Pipkin Jr., 1989: 111). Consequently, the problem of bias is related to the adversarial characteristics of the judicial process. The expert witness may feel some loyalty towards or bias in favour of his or her client. The opposing party and its attorney are usually perceived as hostile, which may produce a conscious or unconscious tendency to reveal that
information which helps the expert’s client and conceal that which might aid the opposition (Pipkin Jr., 1989:111).

Having explained the importance of expert witnesses, it is notable to mention that one of the main challenges in the Murrin case was the determination of the need for an expert. In order to ascertain the appropriateness of the expert evidence, the Supreme Court of Canada identified four criteria upon which its admission depends: relevance, necessity in assisting the trier of fact, the absence of any exclusionary rule, and a properly qualified expert (at 411, Brockman and Rose, 2001: 347). These points of law were clarified by Mr. Justice Sopinka in the case of Mohan. In Mohan, a medical doctor charged with a sexual offence wanted to call a psychiatrist to testify that he did not fit the profile of the three personality groups in which most sex offenders were found (Brockman and Rose, 2001: 347). In his ruling that the exclusion of the evidence was correct, Sopinka examined the admissibility of the evidence on the above-mentioned four criteria. He determined that “relevance is a question of law to be decided by the judge and that the evidence must be both logically and legally relevant (e.g. the evidence must be worth its cost in terms of its impact on the trial)” (Brockman and Rose, 2001: 347). “Evidence that is otherwise logically relevant may be excluded on this basis, if its probative value is overborne by its prejudicial effect, if it involves an inordinate amount of time which is not commensurate with its value or if it is misleading in the sense that its effect on the trier of fact, particularly a jury, is out of proportion to its reliability” (at 411, as quoted in Brockman and Rose, 2001: 347-348). Sopinka also emphasized the risk of expert evidence, that “Dressed up in scientific language which the jury does not easily understand and submitted through a witness of impressive antecedents, is apt to be accepted by the jury as virtually infallible and as having more weight.
than it deserves” (at 411, as quoted in Brockman and Rose, 2001: 348).

Furthermore, expert evidence must also be necessary. Justice Sopinka explained that for this criterion, the test was whether the opinion is likely to provide information “which is likely to be outside the experience and knowledge of a judge or jury” (at 413, as quoted in Brockman and Rose, 2001: 348). Consequently, the “subject matter must be such that ordinary people are unlikely to form a correct judgement about it, if unassisted by persons with special knowledge” (Brockman and Rose, 2001: 348).

The third requirement dealt with by Sopinka was that “expert evidence must also be screened, in terms of whether it is precluded by any of the exclusionary rules” (Brockman and Rose, 2001: 348). For example, “evidence that goes entirely to disposition (as in the Morin case) is inadmissible unless the accused has put his or her character in issue” (Brockman and Rose, 2001: 348). However, this type of opinion may be admissible if “either the perpetrator of the crime or the accused has distinctive behavioural characteristics such that a comparison of one with the other will be of material assistance in determining innocence or guilt” (at 423, as quoted in Brockman and Rose, 2001: 348).

“Sopinka’s fourth requirement for expert evidence is that there is a properly designated expert whose qualifications are from either study or experience” (Brockman and Rose, 2001: 349). Therefore, if the expert is proposing a new theory or technique, such as DNA analysis, “it must be subjected to special scrutiny to determine whether it meets a basic threshold of reliability and whether it is essential in the sense that the trier of fact will be unable to come to a satisfactory conclusion without the assistance of the expert” (at 414, as quoted in Brockman and Rose, 2001: 349). This mention of a novel scientific theory or technique was considered in Terceira, where “the Ontario Court of Appeal found that the
trial judge was correct in admitting DNA evidence at trial” (Brockman and Rose, 2001: 349).

In this case, it was found that the trial judge must only assess the reliability of a scientific methodology, that is, whether it “reflects a scientific theory or technique that has either gained acceptance in the scientific community, or if not accepted, is considered otherwise reliable in accordance with the methodology validating it” (at 27–8, as quoted in Brockman and Rose, 2001: 349). “Once the judge is convinced that the methodology is sufficiently reliable to be put to the jury (a threshold of reliability satisfactory to the judge) and meets the four criteria in Mohan, it is up to the jury to apply the particular science to the facts of the case before it” (Brockman and Rose, 2001: 349). Therefore, the jury will ultimately decide its validity and reliability (at 16, as quoted in Brockman and Rose, 2001: 349). It was also mentioned that concerning DNA evidence, it might also be beneficial to instruct the jury “not to be too overwhelmed by the aura of science infallibility associated with scientific evidence,” and that they should “use their common sense in their assessment of all of the evidence on the DNA issue and determine if it is reliable and valid as a piece of circumstantial evidence” (at 28, Brockman and Rose, 2001: 348).

These four criteria were used in Murrin to decide upon the necessity of expert evidence. In Murrin, the court decided that these criterion was easily met, due to the fact that mtDNA is complicated scientific evidence, an expert is usually used in DNA trials, this was the first time this type of evidence had been used in Canada, and this type of scientific knowledge is not usually possessed by the average person in society. Therefore, the court determined that the issue of substance was “relevance” (Murrin at para. 49). “Relevance means that the evidence is so related to a fact in issue that it tends to establish it” (Murrin at para. 50). However, the evidence in this case was circumstantial evidence tending to
establish identity and is therefore clearly relevant (Murrin at para. 50). However, “expert evidence is usually dressed up in scientific language which is difficult to comprehend and suggestive of a certain degree of certainty and infallibility that the evidence may not deserve” (Murrin at para. 52). The court stated that “due to the fact that experts usually have impressive academic credentials and extensive experience may also serve to lend an air of ‘mystic infallibility’ to the evidence” (Murrin at para. 52). It was decided that the evidence (e.g. mtDNA) would not distort the fact-finding process if the limitations were made clear to the jury because mtDNA is not an actual “genetic fingerprint” (Murrin at para. 55).

In the two United States’ cases of Council and Scott, it was argued by the defence that they were not given an opportunity to properly obtain expert assistance. This type of support was considered a luxurious and unnecessary defence. This can be contrasted with the Murrin case and the importance that was attached to the expert witnesses used—four for the Crown and three for the defence.

In the previous United States cases discussed, the criterion against which mtDNA evidence was judged in order to determine if it was scientifically reliable enough to be used at trial were the Federal Rules of Evidence. It was also noted that both the Frye and Daubert decisions had been used in the past but had since been superceded by this new legislation. However, in the Murrin case, the four Daubert criteria were used. These four criterions were: “falsifiability, peer review and publication, general acceptance within the relevant academic community, and a known rate of error and the existence and maintenance of standards” (Murrin at para. 59). In regards to the first criteria, the hypothesis that a DNA sequence can be extracted and identified from human mitochondria, and “the further hypothesis that its relative frequency in the general population can be described and can serve
as convincing evidence supporting an identification, both meet the requirement of falsifiability" (Murrin at para. 68). As for the second criterion, "scientific literature on mtDNA testing and analysis can be found in journals such as Bio Techniques, The International Journal of Law and Medicine, Journal of Forensic Sciences, and Nature," to name a few (Murrin at para. 69). Therefore, the second criterion of publication and peer review has been met due to methodology and conclusions being accepted by the scientific community. The third criterion concerns widespread or general acceptance in the scientific community. The court concluded that "the evidence demonstrated that a substantial majority of scientists in the field have accepted it, although a minority of scientists, exemplified by the three defence experts, question its current validity" (Murrin at para. 70). In spite of the fact that the scientific community is not unanimous, "there is widespread and general acceptance of the reliability of mtDNA analysis in forensic casework" (Murrin at para. 70). Upon first glance, it appears as though the final criterion, that of a published rate of known error, is not met. However, "the potential sources of error are known and understood and certain fail-safe procedures have been adopted by the laboratories, including duplication of testing and proficiency testing," which ultimately satisfied the court (Murrin at para. 71).

Although contamination, heteroplasmy, and paternal inheritance were all discussed as potential sources of error in Murrin, none of these phenomena were mentioned in the four United States cases. Hence, it can be seen that the possibility that these new scientific discoveries can cause false exclusionary results is not given much weight. However, heteroplasmy was not observed in the Murrin case, the risk of contamination was deemed low due to all of the precautions exercised by both the FSS and FBI laboratories, and the issue of paternal inheritance has not yet been developed enough in order to determine just
what percentage of the mtDNA genome can be attributed to the father.

Finally, in the first United States case of Ware, mtDNA evidence was admitted to trial without the need for an admissibility hearing. Therefore, a precedent was set and for every other case that followed an admissibility hearing was not warranted. In Murrin, a lengthy admissibility hearing was conducted in order to determine if mtDNA was scientifically reliable enough to be used in a court of law. Eventually, it was determined that the evidence could be brought before a jury, but the difference between the law governing scientific evidence in the United States and Canada can be seen.

Other Contributing Factors in the Murrin Decision

There are several factors which may have contributed to the Murrin verdict. First, Mr. Murrin’s defence council told the jury that the police investigation was biased and involved “dishonest police officers” (Penticton Herald, 2000: 1). Mr. Wilson, Murrin’s lawyer, spent a great deal of time focusing on one particular aspect of the RCMP case, the beating of Murrin by three former acquaintances. In his closing statement, he reminded the jury of the testimony given by Kelowna RCMP Sgt. Beth Killaly. Killaly testified she told Sgt. Tidsbury, who has since retired from the force, she was worried that Murrin’s three friends might go too far in their planned confrontation with Murrin on January 5, 1995 (Penticton Herald, 2000: 1). The defence also suggested Tidsbury knew about and even encouraged the beating, which Tidsbury denied under oath. The three friends beat Murrin so badly he would have died if he hadn’t been driven to the hospital. Killaly said she attended a meeting of officers earlier that day about Mindy Tran’s case. Killaly testified she expressed concern to Tidsbury that Murrin might be killed by the three friends. She also testified that Tidsbury said “we could find Mr. Murrin tied to a tree” (Penticton Herald, 2000:}
1. Wilson also reminded the jury that Tidsbury denied that and other parts of Killaly’s testimony. “Why would Killaly come here and tell you lies?” Wilson said to the jury (Penticton Herald, 2000: 1).

The Crown’s theory in the case is that Murrin grabbed Mindy when she came by the house in which he was staying, killed her, and transported her in a trunk to where the body was found. The defence contends Murrin didn’t have enough time to assault the child, dispose of her body, and return home where he was seen by neighbours (Penticton Herald, 2000: 1). Wilson asserted that there were inconsistencies in testimony about where and what time Tran was seen near the accused’s home, and although investigators combed the home where they believed Tran was murdered, Wilson said there was no evidence found to indicate that the victim had been killed there (Penticton Herald, 2000: 1). With respect to the DNA evidence, Wilson suggested that the jury should look skeptically at the mtDNA evidence the Crown introduced at the trial, which they said linked Murrin to the crime.

Second, the RCMP became aware after the trial that one juror seemed to smile often at the accused, but it wasn’t until a tip that they began to examine the relationship between Murrin and the juror (Joyce, 2000: 1). Corporal Grant Learned of the RCMP said that he was called by a reporter and asked if the RCMP was aware that one juror appeared to be smiling often in Murrin’s direction during the six-month-long trial. “It seems there were a number of people who had made anecdotal observations about possible non-verbal communication,” said Learned (Joyce, 2000: 1). He said the RCMP “didn’t have anything until February 9 when we received information about this pending travel” (Joyce, 2000: 1). The travel refers to a juror, Kathy Macdonald, going to St. John’s and meeting with Murrin, which led to a police “review” of her relationship with him. Macdonald told the Vancouver Sun that she
and Murrin are working on a book together. "There's nothing going on here, but I don't want anybody making it something that there is, you know," Macdonald said (Joyce, 2000: 1). Lloyd McKenzie, a former B.C. Supreme Court justice, said there is nothing that prohibits a juror from writing a book after a trial. Dianne Martin, a law professor at Osgoode Hall in Toronto, was asked if the Crown could launch an appeal based on the notion that Macdonald had tainted the jury by flirting with Murrin in court while contemplating writing a book. "They'd need much more than that," she said. "On its own, it wouldn't be enough" (Joyce, 2000: 1).
Chapter III: New Developments in Mitochondrial DNA Analysis and Future Application of the Science

Forensic mtDNA Analysis

Hair Analysis

Shed human hairs are one of the most commonly secured biological evidence materials at crime scenes (Savolainen and Lundeberg, 1999: 77). However, simply comparing hairs microscopically cannot give a positive identification and can only rarely exclude an individual (Savolainen and Lundeberg, 1999: 77). Therefore, nuclear DNA analysis has been used to individualize hairs containing root sheaths. Unfortunately, single shed hairs and hair shafts contain only minute amounts of undegraded nuclear DNA, which can lead to unsuccessful DNA typing. Instead, mtDNA can offer a more suitable target for analysis, as it is present in more than one thousand copies per cell, thereby giving a higher probability of successful DNA typing (Savolainen and Lundeberg, 1999: 77). In the last few years, mtDNA analysis has been used in an expanding range of forensic applications, including analysis of single human hairs, teeth, fragments of bone, and saliva (Savolainen and Lundeberg, 1999: 77). A study performed by Savolainen and Lundeberg in 1999 described six cases in which sequence analysis of mtDNA derived from dog hairs were used. Mitochondrial DNA analysis was used to investigate four types of crime: murder, robbery, theft, and poaching (Savolainen and Lundeberg, 1999: 79).

In cases one and two, a female was murdered and dog hairs were found on the body. Shortly after, another female was murdered and dog hairs were also associated with the victim (Savolainen and Lundeberg, 1999: 78). Therefore, it was believed that the same
individual with the same dog committed both crimes. The dogs of seven suspects were tested but the reference samples were found not to match the evidence from the two murders (Savolainen and Lundeberg, 1999: 78). In case three, a woman was determined to be missing and believed to be murdered. Eight hairs from the dog of this female's family were found in the car of a suspect (Savolainen and Lundeberg, 1999: 78). During mtDNA analysis, it was discovered that evidence from two individuals did not match the reference standards (Savolainen and Lundeberg, 1999: 78). Case four deals with the theft of a medieval Bible and its return (Savolainen and Lundeberg, 1999: 79). Two hairs were found on the Bible and tested against dogs in two different apartments. Only one of the samples matched the found hairs (Savolainen and Lundeberg, 1999: 79). In case five, several bank robberies were performed by a group of four men. A stolen car of a family with a dog was associated with the robberies (Savolainen and Lundeberg, 1999: 79). The dog was tested and its hair was found to be consistent with the hairs taken from the car (Savolainen and Lundeberg, 1999: 79). Finally, case six involved the poaching of wolves. Two samples of hairs were taken from items of the suspects (Savolainen and Lundeberg, 1999: 79). Mitochondrial DNA testing revealed that the hairs were dog hairs and not wolf hairs (Savolainen and Lundeberg, 1999: 79).

**Bone and Tooth Analysis**

A long interval between the time of death and post-mortem examination is usually associated with the decay of soft tissues from which DNA could have been extracted, and this makes it difficult to perform identification of individuals by this method (Yamada *et al.*, 1997: 13). After soft tissues have decayed, hard tissues, for example bone and teeth, remain
as materials for use in identification. Both bone marrow and dental pulp cells are sources of DNA (Yamada et al., 1997: 13). Thick bones, particularly, contain generous amounts of marrow cells. However, in severely damaged or decomposed bodies, bones are often broken, which allow for the invasion of bacteria and the destruction of soft tissues (Yamada et al., 1997: 13). Although teeth contain fewer cells, the pulp cavity containing the cells provides a stable environment for DNA. Dental enamel protects the tooth and the pulp cavity against decay and the invasion of external bacteria and it also has been reported that the high content of hydroxyapatite in the dentine enhances the stability of DNA (Yamada et al., 1997: 13).

Research performed by Yamada et al. in 1997 describes a case in which mtDNA was extracted and compared from the victim’s bones and teeth. First, a burned, headless body was found in a parking lot in Tokyo in January 1996, and then four months later a skull was found buried in the ground near the parking lot (Yamada et al., 1997: 13). A total of 288 base pairs in the D-loop region were sequenced. The resulting sequence was found in only one out of 100 individuals in the researcher’s database (Yamada et al., 1997: 15). The sequences were consistent with another, establishing that the body and skull were those of the person, while the identity of the body was established by comparing the mtDNA sequences with that of maternal relatives (Yamada et al., 1997: 15).

In research performed by Lutz et al. in 1996, a highly charred body was found in a burnt out motor vehicle in a remote forest area. As the fire had severely destroyed the body, identification by morphological means was not possible (Lutz et al., 1996: 206). The situation, as well as the results of the autopsy, led to the assumption that the body could be that of a young woman reported missing some days before and who was assumed to have been the victim of a homicide (Lutz et al., 1996: 206). The identification of the body was
attempted with DNA from a bone sample using PCR systems and amplification of mtDNA sequences. For comparison, blood samples of the parents and a brother of the missing person were obtained (Lutz et al., 1996: 206). The entire control region was sequenced because another hypervariable region was found between positions 440 and 560. In addition, "two positions were found in the central portion of the control region which showed high frequencies of polymorphism" (Lutz et al., 1996: 207). The mother and brother contained 19 identical base variations from the Anderson sequence, while the sequence of the father shared six variable positions with the other sequences, and differed by two other base variations (Lutz et al., 1996: 207). At position 16,222, the sequences of the body parts, the brother, and the mother showed a mixture of both A and G, thus confirming the identity (Lutz et al., 1996: 207).

In a second case, several putrefied parts of an apparently adult human body (left arm, right arm, and trunk) were found in a river within a few days of each other, although it was ambiguous whether these parts belonged to the same person (Lutz et al., 1996: 206). Identification of the body parts was performed by sequencing amplified mtDNA from bone and muscle tissue. Unfortunately, blood samples of possibly related persons were not available (Lutz et al., 1996: 206). It was expected that sequences from each body part would match due to the fact that a human being is homoplasmic. However, the differences recorded in the sequenced control region from the various parts of the body found in the river are shown in Tables II and III (Lutz et al., 1996: 208). The results questioned the supposition that a human being is homoplasmic by demonstrating heteroplasmy.
### Table II: Results of mtDNA Sequencing in Case II (Lutz et al., 1996: 208)

<table>
<thead>
<tr>
<th>Position</th>
<th>And.</th>
<th>VP1</th>
<th>VP2</th>
<th>VP3</th>
<th>VP4</th>
</tr>
</thead>
<tbody>
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<td>A</td>
<td>G</td>
<td>A</td>
</tr>
<tr>
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<td>G</td>
<td>G</td>
<td>G</td>
<td>G</td>
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<td>T</td>
<td>T</td>
<td>C</td>
<td>T</td>
</tr>
<tr>
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<td>A</td>
<td>G</td>
<td>G</td>
<td>A</td>
<td>G</td>
</tr>
<tr>
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<td>A/G</td>
<td>G</td>
<td>A/G</td>
</tr>
<tr>
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<td>A</td>
<td>G</td>
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<tr>
<td>16519</td>
<td>A</td>
<td>G</td>
<td>G</td>
<td>G</td>
<td>G</td>
</tr>
</tbody>
</table>

### Table III: Results of mtDNA Sequencing in Case II (Lutz et al., 1996: 208)

<table>
<thead>
<tr>
<th>Position</th>
<th>And.</th>
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<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
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</thead>
<tbody>
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<td>T</td>
</tr>
<tr>
<td>16239</td>
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<td>G</td>
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<td>G</td>
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<tr>
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<td>G</td>
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<td>C</td>
<td>C</td>
</tr>
<tr>
<td>309a</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>G</td>
</tr>
<tr>
<td>315a</td>
<td>-</td>
<td>G</td>
<td>G</td>
<td>G</td>
<td>G</td>
<td>G</td>
</tr>
</tbody>
</table>
In research reported by Prado et al. in 1997, “unrecognizable and almost totally incinerated bones were discovered in a car crash” (Prado et al., 1997: 42). It was later ascertained that the victim was heavily insured. When mtDNA samples from the victim were compared with samples from the mother, two children, and husband of the victim, the sequences of the mother and two children were identical, but when compared with bone DNA, there were seven mismatches in 210 base pairs of a mtDNA segment (Prado et al., 1997: 42). Therefore, this was a case of attempted insurance fraud involving the murder of a homeless vagrant.

In another case reported by the same authors, a woman claimed that a rich deceased man was the father of her three children. The body was exhumed and “parts of the clavicle, mandible, and iliac bones were analyzed” (Prado et al., 1997: 42-43). Blood samples were taken from the woman and children and compared with the man. It was discovered that one child was excluded with six microsatellite mismatches, the third child was excluded with four mismatches, but the second child was in fact the child of the deceased man (Prado et al., 1997: 43).

DNA Fingerprints from Fingerprints

History of Fingerprints

The revolution in identification based on fingerprints began in Bengal in 1858, when a young Englishman working for the East India Company noticed local people using finger or palm prints as signature on contracts (Wright Mercer, 1995: 25). Different versions of this practice had been used for centuries in Asia, with the earliest reference to identifying criminals by their fingerprints occurred during the reign of Hammurabi (1792-1750 BC) in
Babylon (Wright Mercer, 1995: 25). This Englishman, named William Herschel, brought twenty years of study and a realization of its potential uses and impact to the science of fingerprinting. In 1877, Herschel wrote to the Inspector General of Indian Prisons to suggest that his system of fingerprinting be adopted to facilitate identification (Wright Mercer, 1995: 25). Unfortunately, his request was turned down.

Meanwhile, an English doctor practising in Japan had noticed fingermarks on ancient pieces of pottery (Wright Mercer, 1995: 25). Dr. Henry Faulds was the first to propose that "when bloody fingermarks or impressions on clay, glass, etc., exist, they may lead to the scientific identification of criminals" in his 1880 article in England's Nature magazine (Wright Mercer, 1995: 25). In 1888, Sir Francis Galton became interested in the science of fingerprinting and began to research and promote the science over the next seven years (Wright Mercer, 1995: 25). Galton was the first to mathematically calculate that no two people could have the same fingerprints. Argentina was the first country to adopt fingerprinting and lays claim to having the first murder case solved by fingerprint evidence alone when Francesca Rojas murdered her two sons so that she could marry her young lover in 1892 near Buenos Aires. A fingerprint classification method was developed in 1897 by Sir Edward Henry, the Inspector General of Police of Nepal and India, in order to facilitate the filing and retrieving of fingerprints (Wright Mercer, 1995: 25).

The first person to be convicted of murder on fingerprint evidence in the United States was Thomas Jennings in 1910. At this time, the United States courts had not yet ruled that fingerprints were admissible as evidence, despite their acceptance for a decade elsewhere in the world (Wright Mercer, 1995: 25). An appeal in this case was brought on the grounds that fingerprint evidence was not substantial or scientific proof of guilt. On December 21,
1911, Judge Marcus Kavanagh of the Supreme Court of Illinois stated,

When photography was first introduced it was seriously questioned whether pictures thus created could properly be introduced into evidence. But this means of proof, as well as by means of X-rays and the microscope, is now admitted without question . . . This method of identification is in such general and common use that the courts cannot refuse to take cognizance of it (Wright Mercer, 1995: 25).

The conviction was upheld and Jennings was hanged in February of 1912.

The Identification of Criminals Act in Canada permits the procurement of offender information at the time of charge on an indictable offence rather than waiting for conviction. This pre-conviction acquisition of information has been upheld by the Supreme Court of Canada as constitutionally justified inasmuch as there has always been a clear recognition that offenders responsible for unsolved crimes tend to flee if they know they will be identified (Express Magazine, 1998: 1).

In Re M.H. and The Queen (No. 2), the Alberta Court of Queen’s Bench held that “getting ink on one’s finger does not jeopardize one’s life, liberty, and security of person” (BCCLA, 1998: 3). Moreover, despite evidence that fingerprinting would be a stressful experience for a young offender, a young offender’s security is not infringed because he or she is caused to be emotionally upset or anxious. Therefore, “fingerprinting does not violate the guarantee of those rights found in section 7 of the Charter” (BCCLA, 1998: 3). The taking of fingerprints was also found not to be a search and seizure and, even if it was, it would be allowable since it was reasonable (BCCLA, 1998: 3).

The Supreme Court considered the civil liberties consequences of the Identification Act in R. v. Beare. The Court held that “although fingerprinting may offend the dignity of the accused, it does not unduly invade the accused’s rights” (BCCLA, 1998: 3). As well, the Court deduced that “other aspects of law enforcement and the criminal justice system,
including arrest and being charged involved other distasteful procedures and stigma which far outweighs that associated with being fingerprinted” (BCCLA, 1998: 3-4). The Court also held that “the identification procedures under the Identification Act were not arbitrary since they only apply where there are reasonable and probable grounds to believe the accused has committed an indictable offence” (BCCLA, 1998: 4). Fingerprinting is universally accepted as reliable, efficient, and minimally intrusive to the accused. “Due to the numerous and varied functions of fingerprints, the Court held that it would be inappropriate to require the police to first show on reasonable and probable grounds the necessity of fingerprinting” (BCCLA, 1998: 4).

On the question of fingerprinting violating the accused’s reasonable expectation of privacy, “the Court held that an accused charged with an indictable offence must expect a significant loss of personal privacy” (BCCLA, 1998: 4). An accused must expect that prior to his or her being taken into custody, he or she will be subjected to observation, search, and fingerprinting. Although fingerprinting may be unpleasant, it is of short duration and leaves no lasting impression. Unlike blood tests, “there is no penetration of the body and no substance is removed from the body” (BCCLA, 1998: 4). As a physical encroachment, “fingerprinting amounts to practically nothing and as a humiliation, it is insignificant compared to the publicity surrounding an indictment reported in the media” (BCCLA, 1998: 4).

However, the lower court, the Saskatchewan Court of Appeal, had considered the Act to violate the right to life, liberty, and security of person protected by the Charter (BCCLA, 1998: 4). “The Court of Appeal considered that subjecting a person to fingerprinting prior to conviction violated the elements of dignity and self-respect in the Charter; R. v. Beare”
(BCCLA, 1998: 4). The Supreme Court of Canada, however, “held that fingerprinting does not violate the Charter because any deprivation of life, liberty, and security of the person is not contrary to the principles of fundamental justice” (BCCLA, 1998: 4). The common law permits a number of other significant encroachments. For example, “a police officer can search an arrested person and take any property reasonably connected with the offence as evidence or any weapon found” (BCCLA, 1998: 4-5).

Fingerprint databases have allowed the police to spend less time compiling suspect lists and more time investigating suspects (Saferstein, 1998: 449). However, fingerprint information held in the database can only be purged if written permission is given by the arrested individual (after the charge has been dropped or the accused has been found innocent). Although effective, fingerprinting is prone to several problems such as varying quality of prints taken, difficulty in taking and successfully comparing prints, and the reality that trace evidence at crime scenes frequently is not finger impressions but blood, hair, or semen (Express Magazine, 1998: 1). While there are limitations in fingerprint comparison, comparing human cell composition or DNA is much different. The DNA unique to a person can be obtained from skin, saliva, sweat, breath, hair, blood, or any other body substance (Express Magazine, 1998: 2). This ease of acquisition of samples and the non-intrusiveness of the methods is important, as those opposed to the creation of a comprehensive offender DNA database consistently point to these “obstacles” as grounds for Charter apprehension. With just these fallacies in mind, Constable Bill Donnelly noted during his evidence before the Commons Justice Committee, “You collect more (DNA) . . . cells on your toothbrush every morning when you brush your teeth than we will ever collect in obtaining a DNA sample” (Express Magazine, 1998: 2).
Research

Research conducted by van Oorschot and Jones of the Victoria Forensic Science Centre in Australia has shown that an individual’s genetic profile can be generated from swabs taken from objects touched by hands, which will provide a new tool for crime scene investigation (van Oorschot and Jones, 1997: 767). The researchers “swabbed specific areas of hands and objects, extracted, and quantified DNA from these swabs using PCR analysis. Genetic profiles were readily obtained from swabs taken directly from the palm of a hand” (van Oorschot and Jones, 1997: 767). Dry hands and recently washed hands provided the least amount of DNA. “Swabs of regularly handled objects including a leather briefcase handle, pens, a personal locker handle, and telephone handsets produced genetic profiles that matched the users” (van Oorschot and Jones, 1997: 767). In addition, “a number of pre-cleaned objects held for approximately fifteen minutes (plastic knife handles, a mug, a glass, and new vinyl gloves) also gave the genetic profile of the holder or wearer” (van Oorschot and Jones, 1997: 767). As well, “swabs of the inside of worn condoms where no ejaculation occurred also provided the wearer’s genetic profile, which is relevant to some sexual offence investigations” (van Oorschot and Jones, 1997: 767). In order to determine the effect of multiple handlers on objects, polypropylene tubes were exchanged between individuals. “The strongest profile obtained was found to not always be the person who last held the tube, but instead depended upon the individual” (van Oorschot and Jones, 1997: 767). Finally, “hands swabbed before and after a one minute handshake revealed the transfer of DNA from one individual to another in one of the four hands tested” (van Oorschot and Jones, 1997: 767). The methods used by these two researchers have been used in their laboratory to provide evidence in attempted murder, rape, armed robbery, extortion, and drug trafficking.
cases.

**Canadian Charter of Rights and Freedoms Implications**

The possibility of obtaining a workable mtDNA fingerprint from an ink or latent fingerprint suggests that evidence could be obtained for one purpose but then used for another. Therefore, an analysis must be performed with respect to several Charter issues.

In the 1994 case of *Borden*, [1994] 3 S.C.R. 145, the court ruled that a blood sample of the accused had been improperly obtained, and that the resultant DNA evidence should be excluded from trial. The accused had voluntarily provided a blood sample for DNA fingerprinting in relation to a specific charge of sexual assault, but the police had failed to inform him that they intended to use the sample in the investigation of a prior sexual assault on an elderly woman. Since the blood sample had been obtained from the accused without his valid and informed consent, it was held to be inadmissible, and the accused was acquitted of the first offence. This situation was rectified the next year when Bill C-104 was passed (see ss. 487.04-487.09 of the *Canadian Criminal Code*). Under this legislation, a provincial court judge may issue a warrant that authorizes a peace officer to seize a bodily substance from a person for the purpose of DNA analysis. Peace officers are then able to pluck individual hairs, take a mouth swab, or collect blood droplets from a suspect without that person’s consent, provided that the sample is collected in a manner that respects both the dignity and privacy of the person, and that the force used is reasonable (Allain, 1995: 7).

However, in the case of *R. v. Arp* (1998), 129 C.C.C. (3d) 321, two women were murdered two-and-a-half years apart in the same city and under similar circumstances. The accused was arrested after the first murder and gave the investigating officers scalp and pubic hair samples when asked if he was interested in helping them eliminate him as a suspect.
The samples were to be used to determine whether any of his hair was found where the victim was discovered. Arp was released when none of his hair matched the samples taken from the victim’s coat. The officer advised him that any evidence gathered as a result of the hair sample could be used in court. During the investigation of the second murder, the accused refused to provide samples for DNA testing, but cigarette butts belonging to him were taken after his police interview and the DNA was found to match the semen sample taken from the second victim. The scalp and pubic hairs taken from the accused during the first murder investigation were also found to match the analysis of the cigarette butts and the semen. The accused was arrested and charged with the first degree murder of the second victim and then re-arrested and charged with the first degree murder of the first victim as well.

Several voir dires were held to determine the admissibility of the hair samples obtained from the appellant in 1990. The trial judge found that the central issue in determining whether the hair samples and DNA evidence derived from them was admissible was not whether the appellant's consent in 1990 was limited to the first investigation, but whether an informed and valid consent can be limited in law. In his opinion, there was no principle in law that made it unreasonable or unlawful for the police to resort to the samples already in their possession as a result of the consent given in 1990. The appellant's later refusal to provide a hair or blood sample in 1993 did not affect this conclusion. Moreover, the trial judge found that the initial consent to the taking of the hair samples was not limited either to using those samples for the purposes of a simple comparison with hairs found at the scene of the first murder or to using those samples only for the purposes of the 1990 investigation. Such a limitation would
contradict the appellant's own understanding that any information obtained from the samples could be used against him (www.lexum.umontreal.ca).

If the principles of the Borden case are applied to using an ink fingerprint for mtDNA analysis, it can be seen that the evidence obtained from the analysis would be inadmissible since the accused did not consent to using one piece of evidence for another purpose. This could be said to violate section 13 of the Charter, which states that:

A witness who testifies in any proceedings has the right not to have any incriminating evidence so given used to incriminate that witness in any other proceedings, except in a prosecution for perjury or for the giving of contradictory evidence.

However, if a DNA warrant was obtained under Bill C-104, and the judge issuing the warrant believed on reasonable grounds that the accused had committed the offence in question, the potential Charter dispute would be resolved.

As well, there is a potential section 7 violation if an ink fingerprint is used in order to procure a mtDNA fingerprint. Section 7 of the Charter states:

Everyone has the right to life, liberty, and security of person and the right not to be deprived thereof, except in accordance with the principles of fundamental justice.

The courts have consistently held that the privacy associated with the bodily integrity of a person is sacred due to the fact that much more information can be obtained from a DNA profile than from a fingerprint. However, this is not entirely true since the DNA fingerprint would only be used for identification purposes and safeguards would be imposed so that if the samples were used for purposes other than identification, a fine and/or incarceration could be administered. As well, another sample would not be needed since the ink fingerprint would be the source of the DNA profile. However, there could be an argument made that the identification could be made by procuring a DNA warrant and taking a sample from the accused. This would be possible in all cases except in those in which the suspect
was deceased. In that case, getting a normal sample would not be possible, so the only way to obtain a match would be to use the recorded ink fingerprint.

Additionally, the wording of section 7, especially the “deprivation of life, liberty, or security of the person is contrary to the principles of fundamental justice,” has received notable attention from the courts (Astroff, 1996: 224). For example, in the case of R. v. Wooley (1988), 40 C.C.C. (3d) 531, the Ontario Court of Appeal held that the right of a suspect or accused to remain silent is a well-established tenet of our legal tradition. Therefore, by compelling the suspect or accused to provide a bodily substance (or to obtain one from a pre-existing fingerprint), the state is forcing the individual to give evidence against himself or herself (Astroff, 1996: 224). This results in the individual essentially abandoning his or her right to silence, which is analogous to a Charter infringement of a most serious nature (Astroff, 1996: 225).

Missing Children

Argentina

While DNA fingerprinting is mostly publicized in criminal cases of rape and murder, it is becoming a widely used method in reuniting missing children with their parents. Disappeared or missing children from the Argentine ‘dirty war’ of the 1970s, who were kidnapped and/or adopted by the military, are now being reunited with their families through the technology of genetic fingerprinting (Strauss, 1992: 1).

In the early 1980s, geneticist Mary-Clair King of the University of California at Berkeley was approached by an Argentinian human rights group representing the remaining families of the many kidnapped/adopted children (Strauss, 1992: 1). They were looking for

Page -70-
a method of disproving forged birth certificates presented by the adoptive families. In many cases, the families were dead and the genetic tests and comparisons would have to be based on samples from the grandparents or distant relatives (Strauss, 1992: 1). Mitochondrial DNA testing was used in these cases because when only distant maternal relatives (e.g. great aunts) are alive for comparison, the genetic blueprint in the mitochondrion will match.

With the mtDNA testing, genetic trees were created and in 1984, King and her group were able to establish that there was a 99.9% chance that in many cases the adopted children were from parents murdered by the military (Strauss, 1992: 1). The kinship of 41 of the country’s 196 missing children has been established with four of these relations determined from corpses of children (Strauss, 1992: 2). The Argentine government has established the database of frozen storage of blood samples from families whose missing members have yet to be identified. These samples would be available for future comparisons, allowing the grown kidnap victims to search out their families even after there are no surviving members left (Strauss, 1992: 1).

**Agencies and Registries**

There is nothing quite so devastating as losing a child. Even worse is not to have closure in the form of a body to bury and a grave site to visit if the child is in fact found to be deceased. While many agencies, which will be mentioned below, have coordinated the creation of identification kits that provide a physical description and/or recent picture, fingerprint, and other personal information, to date there has been no agencies that have offered a mtDNA fingerprint on this identification card. If mtDNA sequences were provided, the bodies of missing children could be matched to maternal relatives, as has been done in the case of missing children in countries of war (e.g. Argentina) and missing soldiers.
The Missing Children’s Registry operates as a Canadian response centre for missing children. The agency is linked to all Canadian police and related agencies through the Canadian Police Information Centre (CPIC), U.S. police agencies through the National Crime Information Centre (NCIC), and most foreign police through Interpol (RCMP, 1995: 1). The Registry was established in August 15, 1986, when the Canadian Ministry of the Solicitor General announced a multi-faceted program to help police investigate missing children’s cases in Canada (RCMP, 1995: 1). The Registry’s objectives are to provide an investigative assistance service to all Canadian and foreign police agencies who request the services of the Registry, to assist police and searching agencies locate, recover, and return missing children and youth, to analyze and report findings collected from the Canadian Police Information Centre and Missing Children’s Registry’s database on missing children and youth, and to produce and disseminate information relevant to missing children and youth issues to police searching agencies, government, media, and the public (RCMP, 1995: 2). In April 1993, Citizenship and Immigration Canada joined the Missing Children’s Registry. Later in 1993, this joint operation officially became the “Our Missing Children” program and comprised the RCMP’s Missing Children’s Registry, Revenue Canada’s International Project Return and Citizenship, and Immigration Canada (Our Missing Children, 1999: 1).

The Missing Children Society of Canada (MCSC) is a registered, non-profit organization dedicated to the search for runaway and abducted children (MCSC, 1999: 1). MCSC provides a comprehensive investigative program, free of charge, to assist police and parents in the active, ongoing search for missing children (MCSC, 1999: 1).

N.A.M.C.A. Identification Clinics provide, free of charge, a laminated photo and a
child’s fingerprints in a handy booklet that parents receive at the time of processing (N.A.M.C.A., 1997: 1). The booklet has several pages for pertinent information including height, weight, eye colour, medical alerts, home address, home phone number, etc.

Child Quest International is a non-profit organization that utilizes many new technologies including computer photo digitizing, age enhancement to current age, on-site scanning, and poster making with world-wide distribution (Child Quest, 1999: 2). Child Quest is registered with the Attorney General’s Office and Department of Justice in Sacramento, California, and the National Center for Missing and Exploited Children in Arlington, Virginia (Child Quest, 1999: 2).

The mission of Tag-A-Kid is to provide the means that if a child becomes ‘missing,’ to deliver the tools immediately to law enforcement that will enhance the search and that when a child is ‘found,’ they can immediately be identified and their parents notified . . . always with the child’s safety foremost in the minds of the parents and Tag-A-Kid (Tag-A-Kid, 2000: 1). The Tag-A-Kid Inkless Fingerprint Identification Card is accepted by law enforcement for the classification, search, and retention of fingerprints (Tag-A-Kid, 2000: 1). It is an extremely simple and low-cost method to retain accurate information on a child. The Tag-A-Kid Inkless Fingerprint Identification Card includes space for a child’s fingerprints, a photo, physical description, and pertinent medical information (Tag-A-Kid, 2000: 1).

When the previous registries were found via the Internet, I contacted each one and inquired as to whether or not their agency would be interested in learning about mtDNA and how it can be used to identify missing children, with the ultimate goal of creating a mtDNA database in which samples would be gathered from children with one part being stored within
the database and the other housed on an identification card along with other pertinent personal information. Unfortunately, only two responses were received. The first response was from the Tag-A-Kid organization, who inquired about further material that would explain how mtDNA is used. The second response was from Child Quest International, who stated that they did not have the funds to undertake this type of research, as they were a non-profit organization. It was hoped that a better response rate would have occurred, but that was not the case. This could be attributed to the fact that most of the funding for these types of organizations arises through donations and that they are mostly non-profit.

**Missing Children's Mitochondrial DNA Database**

Unfortunately, missing children stories are real life sagas experienced by thousands of families each year with devastating consequences. Every day there are painful reminders of the missing child and holidays are especially painful. No family is immune to the possibility of such a disappearance (Anonymous, 2000: 1). In many cases DNA sampling offers the only hope of a family reunion and a program called KIDS-DNA Inc. in the United States makes this possible. In the case of a missing person, time is the most critical factor. By linking to this DNA database, a child's DNA left at the scene of a crime could lead to the possibility of a faster and safer recovery (Anonymous, 2000: 1).

KIDS-DNA Inc. was founded by medical industry professionals on the premise that no family should have to endure the agony of not being able to positively identify a lost child when found alive or dead (Anonymous, 2000: 1). Their technical staff is experienced in managing DNA testing laboratories and is supported by an advisory board composed of pathologists and other medical doctors.

The unique Collection-Pro kit sold by KIDS-DNA Inc. lets parents collect a DNA
sample. "with the precision of a professional" (for the price of $18.95). This includes a positive identification indicator that lets the user know instantaneously that DNA was collected for testing. The Collection-Pro Kit includes: one pair of gloves, two sterile buccal sample swabs, one DNA identification swab, one sample collection technique indicator, one pre-paid return envelope, one tamper proof collection seal, one fingerprint home identification kit, and easy to follow step-by-step directions (Anonymous, 2000: 1). Results are sent back to parents in the following format:

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Next, parents fill out a form to verify a match in the database. However, they first have to go through a security login and password verification and while doing a search in the database, if a match appears, the user will get the results (a name, phone number, city and state where the person was found) (Anonymous, 2000: 1). As well, gold and silver pendants and a key chain with the corresponding DNA numbers to match the genetic fingerprint on the back can be purchased from the KIDS-DNA Inc. web-site.

This same program could be possible in Canada, which would consist of a mtDNA database in order to retain samples of a child’s mtDNA in order for it to be matched with a maternal relative or with the child’s own sample in order to prove identity (of both deceased and living children). This would be helpful in cases in which a child is missing for a long time. 
period of time and the identity of the child is hard to prove because he or she has aged considerably. If this type of database was operational, a sample of the child’s hair, for example, could be sequenced and matched with the corresponding sample in the database. This is especially helpful if maternal relatives are unavailable or deceased.

**Disaster Victim Identification**

The victims of disasters, both natural and man-made, are frequently subject to environmental insult and severe degradation. Therefore, mtDNA is often used to identify victims’ bodies due to its high copy number, greater stability, and the ability to match sequences to maternal relatives (Goodwin, Linacre, and Vanezis, 1999: 1707). For example, on February 2, 1998, Cebu Pacific flight 387 carrying 104 passengers and crew on an internal flight in the Philippines crashed into a remote mountainside, killing all on board (Goodwin, Linacre, and Vanezis, 1999: 1707). The recovery of the remains from the crash site was hampered, which led to environmental insult, as well as the mechanical and heat stress suffered during the crash (Goodwin, Linacre, and Vanezis, 1999: 1707). Despite this degradation, it was possible to successfully type 95% of the samples using mtDNA (Goodwin, Linacre, and Vanezis, 1999: 1710). When matches could be made using mtDNA, and confirmed using nuclear DNA testing, the significance of the matches was extremely high (Goodwin, Linacre, and Vanezis, 1999: 1710).

In August 1996, a Tupolev 154 aircraft with 128 passengers and thirteen crew members crashed in the mountains of Norway (Slavkin, 1996: 1024). The remains of the deceased were frozen due to extreme temperatures. Mitochondrial DNA sequences were compared from the frozen tissue with blood from relatives (e.g. mother, children, and
siblings) (Slavkin, 1996: 1024). The identity of 139 passengers and crew were positively identified (no reference points were available for the other two people). However, the identification took 22 days of round-the-clock testing.
Chapter IV: Possible Obstacles to the Widespread Use of Mitochondrial DNA in the Courts

Explanation of Heteroplasmy

According to Wilson et al., heteroplasmy appears to be an intermediate condition in the transition from one homoplasmic sequence to another (Wilson et al., 1997: 170). It has been postulated that the heteroplasmic condition in an individual could exist by one of three mechanisms: “(1) a cell contains mtDNA genomes that are homoplasmic, but different cells carry variants; (2) a cell carries two major mtDNA types, but each mitochondrion is homoplasmic; and (3) an individual mitochondrion could be heteroplasmic” (Wilson et al., 1997: 170). Wilson et al. believe that the simplest model to explain this condition is that the mitochondrion is homoplasmic and the cell is heteroplasmic (Wilson et al., 1997: 170).

These mechanisms by which mtDNA mutations arise and become fixed in mammalian maternal lineages are not yet fully understood. Hauswirth and Laipis (1982) proposed the existence of a bottleneck in mtDNA numbers during development. They suggested that, “at some stage of oogenesis or embryogenesis, selective amplification of a small number of mtDNA genomes allows a mutant genotype present initially at only very low levels to spread to fixation within a lineage in a small number of generations” (Bendall et al., 1996: 1276). In the research of Bendall et al. in 1996, “site heteroplasmy was found in the control region in four twin pairs of 180, suggesting that heteroplasmic point mutations in non-coding mtDNA may be more widespread in human populations than had been reported” (Bendall et al., 1996: 1283).
However, research performed by Baasner, Schäfer, Junge, and Madea in 1998, shows a low level of heteroplasmy, indicating that it is a very rare event or one that it is not easily detectable by direct sequencing (Baasner, Schäfer, Junge, and Madea, 1998: 177). Regardless, the actual rate and pattern of mtDNA heteroplasmy is an unsettled issue of central importance (Parsons et al., 1997: 363). A “high substitution rate raises the possibility that maternal relatives will sometimes differ at one or more base positions” (Parsons et al., 1997: 363). An extensive study was undertaken by Parsons et al. in 1997 to empirically determine the frequency with which maternal relatives differ in mtDNA sequences (Parsons et al., 1997: 363). Samples from four sources were used, “including family reference blood samples sequenced in the course of forensic casework at the Armed Forces DNA Identification Laboratory (AFDIL), blood samples from ‘Oxford’ British families, DNA from CEPH pedigree cell lines, and DNA from Old Order Amish pedigree cell lines” (Parsons et al., 1997: 363). Overall, “327 ‘generational events’ were screened and within these, ten instances of substitution were detected and the substitution rate was calculated to be one in thirty-three generations” (Parsons et al., 1997: 364).

The analysis of mtDNA heteroplasmy in dairy cattle demonstrated that “segregation from a heteroplasmic to a homoplasmic state could occur in just one generation or two, which indicates that only a few of the million or so mtDNA genomes present in the mammalian egg at the time of fertilization actually participates in further development” (Stoneking, 1996: 603). Therefore, the scientific community came to believe that mtDNA heteroplasmy was rare, most likely due to a severe bottleneck in the number of mtDNA genomes that occurs in the zygote or shortly following fertilization. To be certain, “mtDNA heteroplasmy has been observed in various species, including some mammals, but these
invariably involve heteroplasmy for the number of copies of a repeated sequence, not simply base substitutions” (Stoneking, 1996: 603). “This type of heteroplasmy probably arises due to slippage of the polymerase across the repeats during mtDNA replication, and hence may arise de novo each generation, in addition to being transmitted from mother to offspring (Stoneking, 1996: 603). Simple base substitutions are more rare but have been observed, and in humans this phenomenon is generally thought to be restricted primarily to diseases associated with mtDNA mutations. Consequently, “evolutionary geneticists have had reason to believe that heteroplasmy could be safely ignored in the analysis of mtDNA population variation” (Stoneking, 1996: 603).

In 1997, Stoneking reported that there were “several reports of heteroplasmy for base substitutions or simple length variation in apparently normal humans, which suggests that such heteroplasmy may be more common than previously thought” (Stoneking, 1996: 603). The research of Pariet et al. strengthens this case when they found a remarkable level of mtDNA sequence heteroplasmy within a species of European bats with as much variation within an individual bat as there is within the entire population— but they also provide evidence to suggest that such heteroplasmy also exists within humans, but at a reduced rate (Stoneking, 1996: 603).

In another study, differences in the sequences between mother and child samples occurred in seven cases in the form of heteroplasmic substitutions (of 154 samples). In one case, “the heteroplasmy of the maternal sequence was resolved to homoplasmy in the child” (Huhne, Pfeiffer, and Brinkmann, 1998: 28). The heteroplasmic sites were mostly pyrimidine transitions and a heteroplasmic point mutation was found in HVI in four children compared with the mother (Huhne, Pfeiffer, and Brinkmann, 1998: 28). One of these
sequences revealed heteroplasmy for T and C at position 16,189, a site with a high substitution rate. In another child, "this heteroplasmic mutation was additionally accompanied by a heteroplasmic length variation caused by at least one cytosine insertion in the poly-cytosine C-tract which produced a characteristic unclear sequence in the nucleotides following this tract" (Huhne, Pfeiffer, and Brinkmann, 1998: 28). In both cases, the corresponding mother samples were homoplasmic for T at position 16,189 with no C-insertion. Length heteroplasmy associated with a substitution at position 16,189 in the HVI C-stretch region has been described by Bendall and Sykes (1995), and is expected to be a common variant in population screening (Huhne, Pfeiffer, and Brinkmann, 1998: 28). In HVII, three heteroplasmic point mutations were found at position 310 consisting of T and C (rather common) which occurred in 16% of 100 unrelated individuals (Huhne, Pfeiffer, and Brinkmann, 1998: 28). In the fourth sample, "the sequence of the mother showed heteroplasmy for T and C at position 310 and at least two C-insertions in the HVII poly-cytosine stretch" (Huhne, Pfeiffer, and Brinkmann, 1998: 28). Similar to HVI, "this sequence alteration also results in a length heteroplasmy and blurring of bands after this tract" (Huhne, Pfeiffer, and Brinkmann, 1998: 28-29). The corresponding child sequence showed a difference of only one cytosine insertion and homoplasy for T at position 310 (Huhne, Pfeiffer, and Brinkmann, 1998: 29).

Based on these findings, a false exclusion may be made in the interpretation of the mtDNA sequence. It is not routine policy to report an exclusion based on a single base pair difference. "The significance of the evidence should be evaluated through likelihood ratio calculations that incorporate the probability that a mutation has occurred within the lineage" (Parsons et al., 1997: 366-367). Recommendations made by Butler and Levin in 1998,
maintain that larger databases are needed to estimate the frequency of a particular mtDNA type and to improve the statistics with which the significance of a match is decided and to determine how prevalent heteroplasmy is. According to these two researchers, there are four possibilities to consider: "(1) no heteroplasmy is found in either the samples or the suspect—heteroplasmy does not play a role in determining a match or an exclusion; (2) the same heteroplasmy is seen in both the samples and the suspect—if the rest of the sample matches that of the suspect, the presence of the heteroplasmy helps to confirm the match, as it did in the case of Tsar Nicholas; (3) heteroplasmy is not seen in the samples but is seen in the suspect—additional evidence would be needed to conclude either a match or an exclusion; and (4) heteroplasmy is seen in the samples but is not seen in the suspect—more evidence would be required to conclude either an exclusion or a match" (Butler and Levin, 1998: 161).

However, heteroplasmy is slowly becoming better understood by both the general scientific and forensic communities. Although the presence of heteroplasmy may complicate sequence comparisons, careful assessment of known samples, combined with an understanding of the mechanics of heteroplasmy and mtDNA population genetics, can enhance the power of a sequence match in some instances where heteroplasmy is observed. In addition, the forensic mtDNA typing community is actively tracking instances where heteroplasmy is encountered, both in controlled studies and casework (Wilson et al., 2000: 2). Specific patterns are beginning to emerge. For example, position 16,093 in HVI has been identified as a "hot spot," subject to a substantially higher rate of heteroplasmy than other positions (Wilson et al., 2000: 2).

For example, the HVI of the human mtDNA control region "was sequenced in 100 single human hair roots obtained from thirty-three unrelated individuals and two individuals
related in maternal lineage (brother and sister)” (Grzybowski, 2000: 548). Approximately two to six roots were analyzed from each individual for this experiment. Both HVI and HVII were sequenced in a forensic casework sample from the Forensic Medicine Institute in Poland (Grzybowski, 2000: 548). Throughout the direct sequencing of the HVI in thirty-five individuals, thirteen persons with heteroplasmic point mutations were discovered and twenty-four different heteroplasmic positions were distinguished (mostly T/C transitions and A/G variations). “Of the thirteen individuals in whom heteroplasmic mutations were identified, as many as seven displayed multiple heteroplasmy, with the number of heteroplasmic mutations positions ranging from two to six in a single hair root” (Grzybowski, 2000: 549). Furthermore, highly variable levels of heteroplasmy were observed, even among roots from the same individual. For example, “sequence data obtained from three separate hair roots from the same individual showed heteroplasmy at position 16,278 in only one of them, whereas two others were homoplasmic” (Grzybowski, 2000: 549). “Some positions seemed to be subject to a higher rate of heteroplasmy than other sites (e.g. 16,126, 16,294, 16,296, and 16,311) and the two individuals related maternally shared the same heteroplasmic positions” (Grzybowski, 2000: 549). As well, forensic casework samples showed “an exact match of heteroplasmic positions between evidentiary hair and one of the reference hair samples” (Grzybowski, 2000: 549).

The results of this investigation “show extremely high levels of mtDNA sequence heterogeneity occurring not only between different tissues of the same person but also within the same tissue, from hair to hair within an individual” (Grzybowski, 2000: 550). These observations strongly suggest that mtDNA heteroplasmy is more frequent in hair than in other tissues, which may result from the relative ease of its detection in this type of tissue.
(Grzybowski, 2000: 550). However, it must be kept in mind that each hair root is formed from small clusters of stem cells. In comparison, "in leukocytes, the level of heteroplasmy is the average of haplotype proportions found in great numbers of stem cells and, as such, may not be easily detectable by direct sequencing" (Grzybowski, 2000: 550). However, the presence of multiple heteroplasmy, with as many as six differences found in a single hair root, was the most interesting find of this study (Grzybowski, 2000: 550).

This type of transient mtDNA heteroplasmy has several explanations. "Since mtDNA sequences are frequently inserted into the nuclear genome, where they evolve as nuclear pseudogenes, some authors speculate that heteroplasmy could be the result of co-amplification of actual mtDNA and nucleus-embedded mtDNA sequences" (Grzybowski, 2000: 551). Presently, a single nuclear pseudogene corresponding to a human control region has been mapped to chromosome eleven. Unfortunately, "because mtDNA sequences greatly predominate in copy number over their nuclear counterparts, the co-amplification scenario is unlikely in the majority of cases" (Grzybowski, 2000: 551). In practice, "the amplification of the nuclear pseudogene together with the mtDNA control region was observed only in two instances, where specific conditions existed that caused the preferential amplification of the pseudogene" (Grzybowski, 2000: 551). The second mechanism that could be considered as responsible for the complex mutational events would be incomplete maternal transmission. Theoretically, "biparental inheritance of mtDNA may provide a good explanation for the existence of heteroplasmic mixtures at multiple positions, as there are on average eight different differences in control region sequences between two randomly selected Caucasian individuals" (Grzybowski, 2000: 551-552). However, while low levels of paternally inherited mtDNA molecules were previously detected in inter-specific mouse hybrids, there
is no evidence for the biparental mode of inheritance of mtDNA in humans (Grzybowski, 2000: 552). Furthermore, paternal contributions would be undetectable by direct sequencing of PCR-amplified DNA. Therefore, “one may also consider somatic mutations to be involved in generating so many mismatches in a particular type of tissue” (Grzybowski, 2000: 552). These would then be correlated with the stage of development of that tissue, as it was in the case of high levels of heteroplasmy observed by Jazin et al. in the human brain. Accordingly, heteroplasmy would be a natural state for particular tissue types (Grzybowski, 2000: 552).

Regardless of the fact that some mutations occurring at specific positions, identified as heteroplasmic “hot spots,” cannot be absolutely overlooked as mechanisms for generating heteroplasmy in hair, “the fact that the same heteroplasmic positions were encountered in two individuals related in maternal lineage strongly suggests that (at least) these individuals inherited heteroplasmy from their mother (Grzybowski, 2000: 552). Therefore, mutations leading to heteroplasmy would arise in the female germline. Even though there is an abundance of evidence that suggests that heteroplasmy does occur in this germline, “the molecular basis for its maintenance remains elusive” (Grzybowski, 2000: 552). In mammals, the bottleneck mechanism; that is “an event occurring during oogenesis whereby a small number or even a single mtDNA molecule is selectively sampled from a larger population for transmission and amplification, which populates the organism” (Grzybowski, 2000: 552), is presumed to be responsible for maintaining homoplasmy. According to the bottleneck theory, “heteroplasmy is an intermediate state in which new mutations are in the process of rapid shift toward homoplasmy within very few generations” (Grzybowski, 2000: 552). In spite of the bottleneck generally being proposed as the mechanism involved in segregation
of mtDNA types during oogenesis, “the possibility that a similar bottleneck might occur during postzygotic development, leading to differential segregation of mtDNA variants in various tissues” (Grzybowski, 2000: 552), cannot be excluded. Possibly the size of such a bottleneck, “varying stochastically at subsequent mitotic divisions, is also responsible for variable levels of heteroplasmy segregating differentially among individual hairs” (Grzybowski, 2000: 552).

Other than its biological meaning, extremely high levels of heteroplasmy in single hairs may have important implications for forensic casework. Because there is “no sequence homogeneity between hair and other tissues, reference hair will always be required to identify evidentiary hair samples” (Grzybowski, 2000: 552). Additionally, “due to the differential level of heteroplasmy observed from hair to hair, more reference samples should be analyzed before a final conclusion can be reached” (Grzybowski, 2000: 552). Although the occurrence of heteroplasmy makes sequence comparisons more complicated, there are some cases where heteroplasmy can increase the discriminating power of the analysis. For example, an exact match of a heteroplasmic position between one of the reference hairs obtained from the suspect and the questioned hair found at the crime scene was shown by Grzybowski in 2000. In this study, a search for the reference hair profile was conducted in the mtDNA sequence database of 200 Polish Caucasians and no match was found, which suggested that the profile was relatively rare. However, “if there was not an exact match of a heteroplasmic position between evidentiary hair and one of the reference hairs, one could only state that the suspect could not be excluded as a potential source of the evidentiary sample” (Grzybowski, 2000: 552). “Since one of the reference hairs of the suspect and the questioned sample did match at all unambiguous positions and shared heteroplasmy at the
same position, this represented simultaneous occurrence of additional unlikely events and
strengthened an association between the crime scene sample and the suspect” (Grzybowski,
2000: 552). In actuality in this case, the presence of heteroplasmmy strengthened the mtDNA
evidence (Grzybowski, 2000: 552).

**Male Contribution to the mtDNA Genome?**

For many years it has been accepted that mitochondria are inherited exclusively from
the mother. “This assumption has been used to date events in human history, including the
age of our last common female ancestor Eve” (Awadalla, Eyre-Walker, and Smith, 1999:
2524). However, mitochondria contain the enzymes necessary for homologous
recombination and there are at least two routes by which strict maternal inheritance of
mtDNA could be bypassed: “paternal mitochondria enter the egg at fertilization or there are
copies of mtDNA sequences in the nuclear genome that could be transferred back to the
mtDNA” (Awadalla, Eyre-Walker, and Smith, 1999: 2524). In a study by Awadalla, Eyre-
Walker, and Smith in 1999, recombination was detected by considering the relation between
linkage disequilibrium (LD) and distance (Awadalla, Eyre-Walker, and Smith, 1999: 2524).
As the distance between the sites increases, the effect of recombination on LD should
increase. Therefore, “recombination should then display itself as a significant decline in LD
with distance” (Awadalla, Eyre-Walker, and Smith, 1999: 2524). For this study, “five
independently collected data sets were used: 45 complete mtDNA sequences of diverse
geographical origin and four RFLP data sets spanning the whole genome from 147
individuals around the world, 86 Finnish and Swedish individuals, 153 Native Siberians, and
167 Native Americans” (Awadalla, Eyre-Walker, and Smith, 1999: 2524). The results of the
study show a decline in LD with increasing distance. This is expected "if there is genetic recombination but hard to explain in any other way as sequencing errors would tend to obscure the effect, not generate it" (Awadalla, Eyre-Walker, and Smith, 1999: 2525). The researchers speculate that there are three possible routes by which recombination could occur: "(i) paternal leakage, (ii) recombination with copies of mtDNA sequences in the nuclear genome, and (iii) recombination in heteroplasmic individuals" (Awadalla, Eyre-Walker, and Smith, 1999: 2525). However, "the last possibility is unlikely since not only would two mutations have to occur within one individual and be maintained in heteroplasmy until recombination had occurred, but all four haplotypes would need to have descendants in the sample to detect the recombination event" (Awadalla, Eyre-Walker, and Smith, 1999: 2525).

Analyses completed by Morris and Lightowlers in 2000 also suggest that elements of mtDNA may sometimes be inherited from the father. This premise is based on evidence that mtDNA may undergo recombination (Morris and Lightowlers, 2000: 1290). Therefore, "maternal mtDNA in the egg must cross over with homologous sequences in a different DNA molecule, of which paternal mtDNA seems the most likely candidate" (Morris and Lightowlers, 2000: 1290). Three situations in which mtDNA recombinations might occur were suggested by this research. First, "mtDNA recombination could occur between different mtDNA molecules in heteroplasmic oocytes" (Morris and Lightowlers, 2000: 1290). For this situation to arise, "oocytes would have to contain mtDNA molecules that differ at two independent sites, cross-over would have to occur, and all four combinations of the two polymorphisms would have to become fixed in a population, an unlikely outcome" (Morris and Lightowlers, 2000: 1290). Second, mtDNA recombinations might occur with
nuclear DNA. "The nuclear genome is known to contain copies of many mtDNA sequences, there being perhaps 1000 'pseudogenes' in all" (Morris and Lightowlers, 2000: 1290). Therefore, if mtDNA enters the nucleus, it could undergo recombination with these pseudogenes. However, studies involving yeast suggest that mtDNA seldom migrate from the nucleus back to mitochondria (Morris and Lightowlers, 2000: 1290). Third, recombination could occur between maternal and paternal mtDNA in the fertilized ovum (Morris and Lightowlers, 2000: 1290). Sperm mitochondria do enter the egg but are degraded within a few cell divisions, along with paternal mtDNA. Hence, inheritance of paternal mtDNA has never been shown in humans. Regardless, it is plausible “that recombination between maternal and paternal mtDNA might occur before the latter is eliminated, or that paternal mtDNA might persist in maternal mitochondria on very rare occasions” (Morris and Lightowlers, 2000: 1290). However, how paternal mitochondria enter maternal mitochondria is unknown. Mitochondria are capable of fusing with each other, but if maternal and paternal mitochondria are fused, it may be expected “that the fused organelles would be destroyed by the same pathway that degrades paternal mitochondria” (Morris and Lightowlers, 2000: 1290).

This recombination hypothesis was also studied in 1999 by Strauss. To investigate whether recombination occurs in the mtDNA genome, her team investigated DNA variations. Based on the premise that “DNA in different individuals varies at many positions, it can be said that each new mutation arises on a distinctive genetic background” (Strauss, 1999: 2436). Therefore, unless the DNA can organize itself, “the new mutation will stick with the variations already on the same chromosome as it is passed on” (Strauss, 1999: 2436). However, “recombination, which mixes up pieces of the DNA, should gradually destroy such
non-random linkages between DNA variations" (Strauss, 1999: 2436). The further apart two sites lie on the chromosomes, the faster recombination can eliminate the linkage (Strauss, 1999: 2436). Therefore, “if recombination is operating, specific variations are less likely to be found together if they are far apart on the chromosome than if they are neighbours” (Strauss, 1999: 2436). In their study, the researchers counted how often specific mutations at different sites tended to occur together and noted the distance between the mutation sites. “In four out of five human data sets and one chimpanzee set, non-random mutations at distant sites were less likely to be linked than nearby mutations—implying recombination between maternal and paternal DNA” (Strauss, 1999: 2436). This finding is crucial to the “Eve” hypothesis, that contends that a single evolutionary tree of maternal descent can be drawn. However, with recombination, there can be no single tree. Instead, different parts of the molecule have different histories (Strauss, 1999: 2436). Over time, “recombination mixes up genomes so that they become more homogenous, which makes distantly related people look more similar to each other” (Strauss, 1999: 2436).

An investigation into the possibility of the introduction of paternal mtDNA during intracytoplasmic sperm injection (ICSI) was examined in 1997, by Houshmand, Holme, Hanson, Wennerholm, and Hamberger, since in ICSI, the whole spermatozoon including head, midpiece, and tail is injected and deposited in the center of the oocyte (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 223). For this study, six babies conceived using ICSI were investigated together with their parents. “In cases I and V, a polymorphism at position 152, where the normal C was found to be substituted by a T in the other parent, was identified” (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 224). The ability to detect minuscule amounts of paternal mtDNA in predominately
maternal mtDNA was tested by the addition of different amounts of paternal DNA to maternal DNA. In the case of family I, the mother carried the polymorphism and the addition of 0.3 ng of paternal DNA to 100 ng of maternal DNA was needed to get a response, which differed significantly from the background (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 224). In the family of case V, the mtDNA polymorphism was discovered in the father, which gave a lower background signal and enabled detection of paternal mtDNA at a level of 0.2 percent (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 224). In the families of cases II, III, and IV, a polymorphic site was found in nucleotide 73. In the fathers, a G was substituted for an A at this position, which gave a detection limit at the level of 0.2 percent of paternal mtDNA (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 224). Unfortunately, the transmission of paternal mtDNA to the six children by the ICSI technique was not found this study. Recently, “paternal transmission of the mtDNA has been shown to occur in interspecific crossings, but accumulated data indicates that intraspecific mtDNA derived from sperm or other sources is efficiently eliminated at an early stage of embryonic development after normal in vivo fertilization and in different experimental situations including injection of testes and liver mitochondria into zygotes of mice” (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 225). However, the destiny of the paternal mtDNA after the ICSI procedure, when the whole sperm including midpiece and tail is placed in the center of the ovum without prior contact with the zona pellucida or oolemma, has not been investigated, but differences in basic cellular processes may occur (Houshmand, Holme, Hanson, Wennerholm, and Hamberger, 1997: 225). It has “been shown that ICSI differs from natural fertilization in terms of a changed calcium flux and delayed cortical reaction” (Houshmand,
Holme, Hanson, Wennerholm, and Hamberger, 1997: 225). The sperm mitochondria seem to be identified and obliterated from the fertilized egg at the late pronucleus phase in normal intraspecific matings of mice (Housham, Holme, Hanson, Wennerholm, and Hamberger, 1997: 226).

The ICSI study was reproduced by Danan et al. in 1999 with a larger sample size; twenty-seven neonates born after ICSI using mature spermatozoa from twenty-one infertile couples. For each couple, the mtDNA sequenced was different in the mother than in the father. The control region of the mtDNA genome was sequenced in the neonates and compared with the two parental cases and with the pattern obtained after the paternal and maternal DNAs had been mixed (Danan et al., 1999: 468). It was discovered that all of the newborns exhibited a control region pattern identical to those found in their mother. These results demonstrate the absence of paternal contribution in the mtDNA of the twenty-seven newborns studied, with a sensitivity level of approximately two percent (Danan et al., 1999: 468). Although the uniparental inheritance of mtDNA in the blood was demonstrated in this experiment, the possible existence of a biparental mtDNA population in tissues that have not yet been explored cannot be ruled out. As well, several parents did not display a homoplastic pattern in the control region—ten of the forty-two individuals (seven mothers and three fathers) contained a mixed mtDNA population (e.g. heteroplasmy). “Of the seven heteroplasmic mothers, five transmitted their different mtDNA species in equal proportion to their children” (Danan et al., 1999: 469). In one circumstance, “the proportion of the major maternal mtDNA species was increased in the newborn; in the remaining case, the major maternal species alone was found in the newborn’s mtDNA” (Danan et al., 1999: 469).

The final study that will be examined with respect to paternal inheritance of mtDNA
is one performed by St. John, Sakkas, and Barratt in 2000. Although many prior studies have highlighted the presence of maternally inherited mtDNA point mutations, few studies have investigated these mutations in spermatozoa (St. John, Sakkas, and Barratt, 2000: 190). An example is a fifty year-old male from a four-generation family that suffers from mitochondrial encephalomyopathy. "This condition was caused by a maternally inherited point mutation in the tRNA leucine gene, in which a pedigree study of this family reflected a heteroplasmic trend in the segregation of the mitochondria" (St. John, Sakkas, and Barratt, 2000: 190). Although mtDNA analysis was not performed on that patient’s spermatozoa, biochemical studies indicated that the mutation affected sperm motility (St. John, Sakkas, and Barratt, 2000: 190). The three authors of this study hypothesized “that in the testis, where the nuclear background of spermatogonia would bias replication of deleted molecules, coupled with a decrease in the number of sperm mitochondria occurring at the spermatocyte/spermatid stage, there would be preferential accumulation of deleted molecules” (St. John, Sakkas, and Barratt, 2000: 194). Conceivably, “this mechanism would also contribute to the exclusion of the paternal mitochondrial genome from being transmitted to the offspring because the embryo would favour transmission of intact mtDNA” (St. John, Sakkas, and Barratt, 2000: 195). However, the effect of these deletions is only important when a distinct loss of mitochondrial function is identified (St. John, Sakkas, and Barratt, 2000: 195).
Chapter V: Conclusion and Recommendations

Mitochondrial DNA (mtDNA) is another valuable forensic tool available to identify both victims and suspects. "The likelihood of recovering mtDNA in small and degraded samples is greater than nuclear DNA analysis due to the high copy number per cell. Mitochondria are present in hundreds to thousands in number, as compared with the nuclear complement of two per cell" (Melton, 1999: 1). Furthermore, "mtDNA is inherited from the mother only, so that when an individual is not available for a direct comparison, any maternally related individual can provide a reference sample" (Melton, 1999: 1). This is in comparison with nuclear DNA, which is both maternally and paternally inherited. Therefore, mtDNA can only give a tentative identification, as all maternal relatives will have the exact same mtDNA sequence, whereas nuclear DNA can positively discriminate between individuals (except identical twins).

A limitation of mtDNA analysis is the size of the database. At present, the database contains about 2,400 samples, compared with nuclear DNA databases which include millions of variations of samples. Due to this small size, "the current convention in the event of a match is to report the number of times the observed sequence is present in the database to provide some idea of its relative frequency" (Melton, 1999: 2). In contrast, "nuclear DNA typing provides superior discriminatory power, such that we can now approach the possibility that a typed individual has a unique profile with respect to any other person in the world" (Melton, 1999: 2). Therefore, mtDNA can never provide the resolution of individuality that nuclear DNA typing can. For this reason, according to Melton, mtDNA analysis should be reserved for cases or samples for which nuclear DNA typing is not possible; for example,
“shed hairs with no follicle, tissue, or root bulb attached; hair shaft fragments; bones or teeth which have been subjected to long periods of high acidity, high temperature, or high humidity; stain or swab material which has been previously unsuccessfully typed for nuclear markers; and tissue (skin, muscle, organ) which has been previously unsuccessfully typed for nuclear markers (Melton, 1999: 2).

The analysis of mtDNA is extremely rigorous and time consuming. “Based on statistics from laboratories performing this type of analysis, the rate of clearance is approximately one to two cases per analyst per month” (Melton, 1999: 3). Reasons for this include: “small and/or degraded samples requiring numerous rounds of PCR to obtain sufficient DNA templates for sequencing; exhaustive procedures to control for contamination; and sequencing analyses of both strands of DNA in both hypervariable regions” (Melton, 1999: 3). Furthermore, for some types of samples (e.g. hair), mtDNA analysis is more likely to consume the entire sample than nuclear DNA typing (Melton, 1999: 3). Due to these factors, “mtDNA testing should only be performed by laboratories with considerable experience in handling the difficult samples that require this type of analysis” (Melton, 1999: 3). “Experienced laboratories can extract minimal amounts of mtDNA from difficult samples, and in the event of sample failure, an inexperienced laboratory would not know whether their extractions and PCR rounds were not sensitive enough, or whether the sample lacked non-degraded DNA” (Melton, 1999: 3). Finally, contamination controls are intensified in a mtDNA laboratory (Melton, 1999: 3).

As well as in forensic examination, mtDNA is also useful in two other scientific areas. The first deals with the fact that there are numerous serious human diseases caused by deletions and mutations in coding regions of the mtDNA genome (Melton, 1999: 1).
Second, "molecular anthropologists have been examining the genetic variation in populations all over the world to track migration patterns, expansion dates, and geographic homelands of humans" (Melton, 1999: 2).

The first forensic case that certainly put mtDNA testing into the realm of the public eye was the case of the Romanov family. Heteroplasmy definitely caused a wrinkle in this case of identifying the remains of the Romanovs, including Czar Nicholas II and the Czarina. However, the heteroplasmic mutation in the sequence of the Czar and his brother led to the indisputable recognition of the skeletal remains of the family, except for the Princess Anastasia. Unfortunately, her remains have not yet been found, although one woman claimed to be the missing member of the Romanovs. However, the mtDNA sequences did not match between the known relatives of the Romanov family and the reputed Anastasia. Therefore, the mystery of Anastasia continues.

Forensically, there are four major cases in the United States and one in Canada in which mtDNA was used as evidence. In the United States, each state has its own legislation that guides the collection and use of mtDNA. Therefore, the state of Tennessee was chosen as the first state in which mtDNA evidence would be admitted because the rules of evidence were much less strict than in other states. As well, the thinking of the prosecution was that once mtDNA evidence was admitted in one state, it would make its admission in other states easier, which definitely was the case in the next three cases. However, although Ware was convicted, there was a large amount of circumstantial evidence that may have played a bigger part than the scientific evidence. In the second case, that of Council, the methodology of mtDNA sequencing and its science was generally accepted due to the Ware case. Therefore, there was a much less detailed investigation into the flaws of the science before the evidence
was admitted to court. In the third and fourth cases, Underwood and Scott, the defence was denied the use of an expert, which could be seen as a miscarriage of justice, in that the defence should have the same privileges as the prosecution. However, both cases were held under appeal. Although the defence tried to argue that in both cases the mtDNA evidence was unreliable, the court disagreed.

The only case to have been tried in Canada using mtDNA evidence was the case of Murrin in 1999. In this case, three hairs found inside the underpants of the victim were matched using mtDNA with samples from the defendant, Shannon Murrin. The defence tried to challenge three aspects of the science: contamination, heteroplasmy, and fundamental aspects such as paternal inheritance. The court held that none of these questions held any merit and the evidence was admitted. However, the mtDNA evidence did not lead directly to a conviction. The accused was acquitted, probably based on other factors such as the alleged police misconduct (the police were accused of using force to elicit the help of another citizen in order to secure a confession from Murrin), a juror that served on the jury of the case who currently has a relationship with the accused and is supposedly writing a book to account the trial, and the possibility that another individual may be responsible for the crime (an individual currently serving a sentence for sexually based crimes at a Federal Institution in British Columbia). When I first started to write this thesis, I assumed that I would be able to answer one main question, “Why has mtDNA evidence been so easily accepted in the United States but not in Canada?” Unfortunately, I am no closer to the answer than when I started. I thought that the Murrin case would help, but it only brought more questions. Instead of the mtDNA evidence, the jury was overwhelmed by the other factors mentioned above, which probably influenced their decision. Therefore, instead of a case that helped
answer my questions, Murrin turned into a case that gave more questions. It is troublesome that a case which could have been a landmark in Canadian forensic history turned out to be the exact opposite; almost a comedy of errors. If the accused would have been found guilty based on the mtDNA evidence, it may have opened the door for many other cases that used the same evidence to be tried in Canada. Instead, there have been no cases since Murrin that have involved mtDNA evidence.

Recently, there have been several new developments in the discipline of mtDNA testing. Hair is one of the most frequent types of evidence found at a crime scene. However, a positive identification was not possible until mtDNA testing. Several cases have been solved by comparing dog hairs left at crime scenes with those on the accused’s clothing, in the accused’s vehicle, and compared with the accused’s dog. As well, bone and teeth are excellent sources of mtDNA since they are usually left at crime scenes even after environmental insult, burning, and putrefaction. Numerous cases, including disaster victim identification, have been solved using the high copy number, incredible stability, and maternal inheritance of mtDNA. Examples include plane crashes in the Philippines and Norway.

Fingerprints were first used as signatures thousands of years ago. Although it has never been proven that each and every person has a unique fingerprint, statistics have allowed us to believe beyond a reasonable doubt that fingerprints are as distinctive as snowflakes. The first conviction secured using fingerprints occurred in 1910 in the U.S.. Currently, the Identification of Criminals Act in Canada allows for the seizing of fingerprints at the time of arrest. This taking of fingerprints was held not to violate section 7 of the Charter in the case of Beare since fingerprinting is not physically violating. New research
into fingerprints has found that it is possible to obtain an mtDNA profile from a latent fingerprint, ink fingerprint, or a handshake. This new use of an old piece of evidence has, however, brought forward several Charter issues. For instance, the court held that in the case of Borden, evidence gathered for the purpose of the investigation of one crime cannot be used as evidence in another crime without the consent of the accused. However, Bill C-104 has rectified this situation since now a warrant can be obtained in order to acquire a DNA sample from a suspect with reasonable and probable grounds. The courts have continued to maintain the importance of the bodily integrity of citizens and the right not to provide incriminating evidence against oneself. Therefore, Charter issues under sections 7 and 11 may be relevant if a mtDNA sample is taken from an already existing fingerprint.

During military strife in Argentina, hundreds of children were left orphaned when their parents were killed and adopted by other military personnel. Mitochondrial DNA testing has been used to re-unite many children with their maternal relatives, since several generations needed to be skipped in order to find a match. The importance conferred upon the magnitude of finding missing children is illustrated by the numerous agencies and registries have been established in Canada, the United States, and around the world. These databases are linked to CPIC, NCIC, and Interpol in order to facilitate the detection of children who are missing. However, most of these organizations are non-profit and have trouble raising funds in order to acquire new technology, such as a mtDNA database.

One organization, called KIDS-DNA Inc., has begun to use mtDNA testing as a proactive way in which to identify missing children. For a nominal fee, parents can use a kit to collect a mtDNA sample and submit it to the company for safe keeping. If the child ever is missing, the mtDNA can be used to identify the child or its remains. Ultimately, it is this
type of database that I envision for the entire world. Mitochondrial DNA would be taken from children, stored in a database, and used for identification in order to provide closure for families. However, it is not known yet how parents would react to this type of morbid evidence gathering. I am currently undertaking a study which will try to assess the willingness of parents to allow their children to be mtDNA tested. Additionally, this database would be quite expensive, with potential funds coming from governments and perhaps parents themselves.

Finally, there are two recent scientific developments that have called the reliability of mtDNA into question. The first is heteroplasmy. While the exact mechanism of heteroplasmy is not known, there have been several speculations. However, several of the same sorts of patterns have been found; the same exclusions and inclusions which has made the identification of heteroplasmic tracts easier. In order to better understand heteroplasmy, a larger database is needed. The second new development is paternal inheritance. For many years, it was assumed that mitochondria were inherited exclusively from the mother. In a study in 1999, three different routes by which paternal recombination could occur were proposed. As well, ICSI babies were investigated in order to determine if paternal DNA persists in the maternal oocyte. However, the results of this research suggested that the paternal DNA is degraded and does not persist. Therefore, there is no solid evidence that mitochondria are composed of both maternal and paternal DNA.
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