Use of DNA Profiles in Criminal Proceedings in Alaska

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Use of DNA Profiles in Criminal Proceedings in Alaska

I. Introduction

A. Background of this Report

As part of legislation establishing a DNA registry for some convicted felons,[1] the Alaska Legislature asked the Judicial Council to:

> periodically review and distribute information relevant to the technical, legal, and scientific use of DNA profiles in criminal proceedings to: (1) judges and magistrates, (2) the Department of Law, (3) the Public Defender Agency, and (4) the office of public advocacy.[2]

While the Council did not request this assignment or receive funds with which to complete it, the Council has tried to compile information that will help Alaska practitioners.

In January and February of 1996, the Council surveyed all judges, prosecutors, public defenders and OPA defense attorneys, as well as most private criminal defense attorneys to ask what information they would find most helpful for a case involving DNA. The respondents wanted several kinds of information: analysis of state and federal law on DNA-related evidentiary issues; an overview of scientific and lab techniques for analyzing DNA; information about DNA testing done by the Alaska State Crime Lab and analysis of statistical and population genetics issues. This report discusses these topics.

B. Structure of this Report

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[1] House Bill 27 was passed in 1995.

[2] AS 22.20. 200 (1995).

Section II of this report explains the biological basis for DNA testing. Section III describes some of the more common DNA testing procedures, including RFLP and PCR-based analysis, and discusses how DNA test results are interpreted. Section IV gives the history of forensic DNA testing, including the genesis of the controversy over population substructures and other technological and scientific issues that have faced the courts. Section V summarizes legal authority on forensic DNA tests, including local statute and case law and related legal issues. The sixth section summarizes the Alaska State Crime Lab's recent DNA testing and database compilation efforts.

Note to Readers

Legal and technical issues related to the forensic use of DNA in criminal proceedings change almost weekly. Thus, a written report such as this one cannot contain the most up-to-date information. Instead, this report provides the solid background needed for attorneys and judges to understand some of the most important legal and technical issues concerning DNA in the courts. More up-to-date information is available from the experts in the field, scientific articles and fellow practitioners. The reader also should review the electronic version of this report on the Council's Internet page (http://www.ajc.state.ak.us) for hyperlinks to current information.

II. Understanding Genetic and Molecular Basis for DNA Testing

Some principles of genetics and molecular biology are necessary to understanding the two principal kinds of genetic systems used in forensic DNA typing. This section describes some basic concepts about DNA, chromosomes and genes.[3]

Each human body is composed of an enormous number of cells. Most of these cells contain a nucleus (or inner part), which in turn contains the person's genetic material (the *genome*). All the cells in every human body descend by successive divisions from a single fertilized egg.

[3] Much of the following discussion is taken from: NATIONAL RESEARCH COUNCIL, THE EVALUATION OF FORENSIC DNA EVIDENCE (prepublication copy) (1996)(hereinafter NRC II).

A. What is DNA?

Chromosomal DNA[4] is the chemical storehouse of genetic information. The DNA molecule itself resembles a twisted ladder or double helix. The molecule is composed of four chemical subunits called *bases*: guanine (G), adenine (A), thymine (T) and cytosine (C). These bases pair between strands: A on one strand with T on the other, and C with G. The sequences of *base pairs* in turn are arranged in long chains of varying lengths that form the DNA double helix. Each person has about 3.3 billion base pairs.

B. DNA Resides in the Chromosomes

Virtually the entire complement of a person's genetic material resides in the *chromosomes.*[5] A chromosome is a very thin thread of DNA, surrounded by other materials, mainly protein.[6] Chromosomes are located in the cell's nucleus.

Almost all cells in the human body contain 23 pairs of chromosomes (for a total of 46 chromosomes).[7] The two members of a chromosome pair are said to be *homologous*. One member of each homologous pair is inherited from the mother (the egg) and one from the father (the sperm).[8] Because of the way DNA in the original fertilized egg replicates itself, virtually every cell in the body has the same chromosomal make-up. An important exception is found in sperm and egg cells, which have only half as many chromosomes (a total of 23) as the rest of the body's cells. The full number, 46, is restored by fertilization.[9]

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[4] Deoxyribonucleic acid.

[5] U.S. CONGRESS, OFFICE OF TECHNOLOGY ASSESSMENT, GENETIC WITNESS: FORENSIC USES OF DNA TESTS 42 (1990)(hereinafter GENETIC WITNESS).

[6] NRC II, *supra* note 3, at O-3.

[7] More specifically, humans have 22 pairs of *autosomes* and one pair of sex chromosomes (X, Y).

[8] The mother passes on 22 autosomes and one X chromosome while the father passes on 22 autosomes and either an X or a Y chromosome. Women have two X chromosomes while men normally have an X and a Y.

[9] How can sperm cells, which contain only half of the full number of chromosomes, provide the same information as cells containing the full complement? When DNA from many sperm cells is

C. Genes and Chromosomes

Integral to the chromosomes are the genes. Structurally, a gene is "a stretch of DNA, ranging from a few thousand to tens of thousands of base pairs, that produces a specific product, usually a protein."[10] Functionally, genes are the basic units of heredity. The gene's function is determined by the order of the four kinds of bases within it. The specific base sequence acts as an encoded message, each specifying an amino acid (a protein building block).[11]

Genes are interspersed among the rest of the DNA and actually compose only a small fraction of the total DNA. Genes differ from the rest of the DNA on the chromosome only in having a specific sequence of bases which enable them to encode a specific protein. Most of the rest of the DNA has no known function.[12]

The position that the gene occupies along the chromosome (or DNA thread) is its *locus.*[13] Each chromosome contains many different loci, arranged in a specific linear order. The order is the same for every human. For example, the locus for the gene responsible for cystic fibrosis is on chromosome 7.[14] Everyone has this gene at the same position on chromosome 7, although only some people have the alteration in the gene that causes it to be defective and produce cystic fibrosis.

Genes are passed on from parents to their offspring via egg and sperm cells. During *meiosis*, the process that results in the development of sperm or egg cells, two partner chromosomes in a chromosome pair[15] line up side by side and randomly exchange parts. The result is that genes formerly located on the same chromosome can move to the homologous chromosome, and genes formerly located on homologous

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analyzed at once, collectively all the chromosomes are represented. NRC II, *supra* note 3, at O-3.

[10] *Id.* at O-4.

[11] *Id.*

[12] *Id.*

[13] *Id.*

[14] GENETIC WITNESS, *supra* note 5, at 42.

[15] Recall that a chromosome pair contains one maternal and one paternal chromosome. Such pairs are said to be *homologous*.

chromosomes can move to the same chromosome.[16] Genes that are very close to one another on the same chromosome might remain associated for many generations before they are separated.[17] Genes on the same chromosome are said to be *linked*; that is, they tend to be inherited together.[18] Genes residing on nonhomologous chromosomes are inherited independently, as are genes far apart on the same chromosome.[19]

D. Most Genes Are the Same for All Humans

Most loci contain the same gene in every human being, while some (like the cystic fibrosis example) can vary among individuals. It is thought that about 3 million of the total 3.3 billion base pairs differ between any two individuals.[20] In other words, most of our genetic material is the same. This fact is not surprising when we think of how many characteristics all humans share: one head, two eyes, two legs, etc.

E. Some Genes Differ Among Individuals

At each genetic locus reside two genes, one inherited from the mother and one from the father. The form of the gene inherited from the mother can differ from the one inherited from the father. An *allele* is an alternative form of a gene (for example, those producing normal and sickle cell hemoglobin). If the mother and father have different alleles, then their child will inherit one allele from the father and one from the mother. In other words, while only two genes reside at each locus, multiple different alleles are possible at each locus.[21]

An example is the gene responsible for determining blood type. Three possible variants or alleles exist for this gene: A, B, and O. Which combination of the three a person has depends on which she inherited from her parents. So if the father passed

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[16] NRC II, *supra* note 5, at 2-5.

[17] *Id.* at 2-4.

[18] *Id.*

[19] *Id.*

[20] GENETIC WITNESS, *supra* note 5, at 42.

[21] A locus for which multiple alleles exist is said to be *polymorphous*. For some loci, as many as 50-100 different alleles are possible. *Id.*

on the A allele and the mother passed on the B allele, the child is blood type AB.[22] Someone who got the O allele from one parent and the O from the other would be type O.[23] In a more generic example, if there are two possible alleles at a locus, A and a, then there are three *genotypes*: AA, Aa, and aa. The word *genotype* refers generally to the genetic makeup of an organism; however, it also can be used to describe the genetic makeup at a number of loci, from one to the total number.

DNA testing concerns itself with those loci at which genes can differ among individuals. Obviously, it would do no good to test one of the loci for which no genetic variation is possible, because the test would show a complete match for every human being. Forensic DNA tests also typically examine alleles from four or five different loci. The tests compare DNA sequences at these loci between different individuals.

III. Description of Forensic DNA Testing

As of 1995 or so, most DNA testing involved one of two basic techniques: analysis of *variable-number tandem repeats* (VNTR),[24] and polymerase chain reaction-based (PCR) methods. PCR is the newer of the two technologies.

A. VNTR Typing

Certain regions of DNA, known as *variable number tandem repeats* (VNTRs), have no known function and show great variability among individuals. These are the regions of the DNA used in VNTR typing. The VNTR loci used in forensic tests reside on different chromosomes (or sometimes very far apart on the same chromosome), so they are independently inherited.[25]

In VNTR regions, usually ranging from 500 to 10,000 nucleotide pairs, a core sequence of some 15-35 base pairs repeats many times consecutively along the

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[22] A person with two different alleles at a particular locus is said to be *heterozygous* for that particular locus.

[23] A person with the same two alleles at a locus is said to be *homozygous* for that locus.

[24] VNTR typing sometimes is referred to as restriction fragment length polymorphism (RFLP) analysis.

[25] NRC II, *supra* note 3, at 2-6.

chromosome.[26] The number of repeats varies from person to person.[27] (Sequences with different numbers of repeated units are called *alleles*, even though the word originally applied to functional genes.)[28]

Several features of VNTR regions make them useful for forensic testing and identification. First, VNTR loci have a very large number of alleles (often a hundred or more), and none of the alleles is very common, so great variation exists among individuals.[29] Second, because VNTR regions have no known function, they are less likely than functional genes to be influenced by natural selection and thus less likely to vary in the frequency with which they appear in different populations.[30] Finally, the variable number of base sequence repeats makes VNTR regions identifiable by their lengths, so they can be readily sorted by laboratory procedures.

Although details vary somewhat among labs, the basic steps of VNTR typing are uniform. In VNTR typing, the lab first excises fragments of chromosomes that begin and end with certain sequences of DNA base pairs from DNA at the crime scene and DNA from the suspect(s).[31] The tester then measures the lengths of the DNA fragments in the samples by seeing how far they move through a slab of gelatinous material when attracted by an electric charge.[32] The tester compares how far the sample fragments moved relative to how far fragments of known lengths moved.[33] The analyst then applies a "probe" or genetic marker to make visible the genetic patterns

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[26] *Id.*

[27] *Id.*

[28] *Id.*

[29] *Id.* The large number of alleles at each location translates into a large number of possible genotypes. For example, at a locus with 20 alleles, there are 20 homozygous genotypes, plus 190 heterozygous genotypes [(20)(19)/2], for a total of 210. *Id.*

[30] *Id.* at O-6. For example, genes that cause malaria resistance are more common in people of Mediterranean or African ancestry than among people living in areas where malaria is not common. *Id.*

[31] *D. Kaye, DNA Evidence: Probability, Population Genetics and the Courts*, 7 HARVARD JOURNAL OF LAW & TECHNOLOGY 101, 108 (1993).

[32] *Id.* Shorter fragments, which have lower molecular weights, move more quickly than longer fragments, which have higher molecular weights. *Id.*

[33] This process is called *electrophoresis*.

at each locus. Commonly, labs examine four or five VNTR loci for each sample.[34] The following steps compose the typical VNTR analysis:[35]

1) Isolate the DNA from the specimen to be examined;

2) cut the DNA into discrete pieces using a bacterial enzyme (called a restriction enzyme);

3) separate the different-sized DNA pieces using a process called gel electrophoresis;

4) transfer the DNA from a gel to a nylon membrane to make it easier to work with;

5) apply (or hybridize) a DNA probe to the membrane (the probe usually is radioactively labeled, although some labs are beginning to use luminescent molecules); and

6) show the location of the probe, usually by exposing the membrane to x-ray film (autoradiography), or if luminescent probes are used, to light-sensitive film.

If a radioactive marker is used, the entire process for four or five probes takes several weeks.[36]

After the film is developed, the analyst must examine the images and interpret the results of the test. The film typically shows a number of parallel bands running across it, like lanes on a highway. The position of each radioactively labeled band on the membrane indicates the size of the VNTR. Because of measurement uncertainty, however, the test does not reveal the exact size of a band. The analyst must account for this uncertainty when analyzing autorads. [37]

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[34] The analyst completes the test with one probe, then washes it off and repeats the entire process with another probe targeting another VNTR locus on another chromosome. The analyst repeats the entire process for each of the multiple probes. NRC II, *supra* note 3, at 2-9.

[35] For a detailed description of a typical VNTR typing process, see Judge Steinkruger's findings of fact in *State v. Harmon.*

[36] NRC II, *supra* note 3, at 2-9. Several days are required for the radioactive probes to emit sufficient radioactivity to produce a visible band on the film. *Id.*

[37] *Id.*

Because the most common DNA tests for measuring the lengths of the VNTR fragments are not sensitive enough to distinguish between fragments that are extremely close in size, laboratories group bands of similar size into *bins*.[38] The analyst then treats the alleles within a bin as though they are a single allele.[39] The usual width of a bin is about 10% of the mean (average) size of the VNTR segment at the center of the bin.[40] After binning, the analyst compares the number and location of the various bands in the lanes on the autorad. A person whose DNA falls into the same bin is said to be *homozygous*, while a person whose DNA falls into different bins is said to be *heterozygous*. Interpretation and analysis are discussed further in section C, below.

B. Polymerase Chain Reaction (PCR) Analysis

PCR analysis can be thought of as molecular photocopying. The process results in a million or more copies of a short region of DNA (usually less than 1,000 nucleotides in length).[41] In this three-step process, the lab first heats the double-stranded DNA segments to separate them into two strands. Second, the lab hybridizes the single-stranded segments with *primers*, short DNA segments that complement and define the target sequence to be amplified. Each primer serves as the starting point for replicating the target sequence when mixed with the enzyme DNA polymerase and the four nucleotide building blocks (A, C, G and T).[42] The process copies the complement of each of the separated stands, resulting in two double-stranded DNA segments. The lab then repeats the three-step cycle, usually twenty to thirty-five times.[43]

Once the DNA is amplified, analysis proceeds the same as with VNTR methods, with a few modifications.[44] Like VNTR analysis, PCR-based methods rely on the

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[38] *Id.*

[39] *Id.*

[40] *Id.*

[41] *Id.* at 2-11. Because the process can not as yet make copies of large regions of DNA, it cannot be used on VNTRs. *Id.*

[42] *Id.*

[43] *Id.*

[44] *Id.*

principle of identifying different-sized fragments by their migration rates in an electric field.[45]

PCR-based methods are used on a number of different classes of DNA fragments and genes. One class of repeated DNA units labs analyze using PCR is *short tandem repeats* (STRs). These repeats of a few nucleotide units are very common and are distributed widely throughout the genome.[46] While STRs have fewer alleles per locus than VNTRs, a very large number of loci are potentially usable.[47]

Another PCR application uses the DQA locus.[48] Unlike VNTRs, the eight alleles at DQA code for a protein.[49] Six of the eight alleles identified at this locus can be distinguished by specific probes.[50] The DQA system can be used, along with other markers, as part of a more detailed DNA profile.[51]

A third PCR system is the Amplitype polymarker (PM). This system analyzes six loci simultaneously: DQA, LDLR (low-density lipoprotein receptor), GYPA (glycophorin A, the MN blood-groups), HBGG (hemoglobin gamma globin), D7S8 (an anonymous genetic marker on chromosome 7), and Gc (group-specific component).[52] Two or three distinguishable alleles exist at each locus.[53] However, polymarker loci vary more among races than do VNTRs.[54]

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[45] *Id.*

[46] *Id.*

[47] *Id.*

[48] *Id.*

[49] *Id.* The locus is part of the histocompatibility complex, a group of highly variable genes responsible for recognizing foreign tissue. *Id.*

[50] *Id.* at 2-13. The six alleles yield 21 possible genotypes: six homozygous and 15 heterozygous. *Id.*

[51] *Id.*

[52] *Id.*

[53] *Id.*

[54] *Id.* at 4-31.

Another PCR-based technique involves D1S80, a VNTR in which the largest allele is less than 1,000 base pairs long.[55] The locus has a sixteen-base unit repeated a variable number of times. More than 30 distinguishable alleles exist at the locus, and the size classes are discrete. The analysis is complicated, however, by insertion or deletion of a single base.[56]

Finally, another class of genetic marker is mitochondrial DNA. Unlike most DNA, which is found in the cell's nucleus, mitochondrial DNA is found outside the nucleus in the cell's mitochondria. Also unlike other DNA, mitochondrial DNA passes only from mother to child, so that all the children of one woman have identical mitochondrial DNA. Certain regions of the mitochondrial DNA are highly variable and thus have been used for forensic analysis.[57] Because each person inherits mtDNA from his or her mother, the technique cannot distinguish siblings or other maternally related relatives.

PCR-based methods differ from VNTR analysis in several respects. First, PCR-based analyses usually permit exact identification of each allele, avoiding the problem of measurement uncertainty.[58] Second, results of PCR-based analyses normally are available within about 24 hours, much sooner than VNTR results. Finally, PCR analysis is better-suited than VNTR typing for smaller samples for which very little DNA is available.[59]

PCR analysis presents at least three disadvantages not present in VNTR analysis. First, a sample that is contaminated can make multiple copies of the wrong DNA. The amplification process is so efficient that a few stray molecules of contaminating DNA could affect the band pattern enough to cause an analyst to

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[55] *Id.* at 2-13.

[56] *Id.*

[57] *Id.* Mitochondrial DNA has one of the highest spontaneous mutation rates of any genome. Parts of the genome are thought to mutate quickly enough that grandmothers have a different sequence than granddaughters. For an article evaluating the validity and reliability of forensic use of sequencing mtDNA, see Wilson, DiZinno, Polanskey, Replogle & Budowle, *Validation of Mitochondrial DNA Sequencing for Forensic Casework Analysis,* 108: 2 INTERNATIONAL JOURNAL OF LEGAL MEDICINE 68-74 (1995).

[58] NRC II, *supra* note 3, at 2-13.

[59] *Id.*

declare a nonmatch when a match actually exists, or to declare a match when the evidence DNA actually does not match the suspect's DNA.[60] Second, most markers used in PCR-based typing have fewer alleles than VNTRs, and the distribution of allele frequencies is not as flat.[61] The lab therefore must analyze more loci to get the same amount of information about the likelihood that two people share a profile.[62] Finally, some of the loci used in PCR-based analysis are functional genes; some are linked to important disease-producing genes.[63] Recall that functional genes are more likely than non-functional markers to be influenced by natural selection and thus to appear with different frequencies in different segments of the population (the sickle-cell example).

C. Interpreting DNA Test Results

No matter which type of DNA testing is used, the technicians performing the test must interpret the results in some way. First, the examiner must decide whether the DNA fragments in the crime scene sample match the suspect's DNA. Second, the examiner must estimate the probity of the match; in other words, is the DNA pattern so common that it could have come from any number of people or is it so uncommon that it could have come from only a few individuals?

1. Declaring a Match. In both PCR-based and VNTR profiling, the analyst compares the location and size of the bands on the autorad to see whether any of the bands resemble each other. Labs impose two conditions for declaring a match: First, the examiner must believe that the suspect's fragments have migrated the same distance on the gel; second, computerized measurements must confirm that the difference in migration distances is less than some standard deviation of a set of independent measurements of fragments taken from one sample.[64]

2. Evaluating the Probity of the Match: Principles of Population Genetics. Evidence that the suspect's DNA matches DNA taken from the crime scene is not the end of the evaluation. Declaring a match would not be particularly probative

[60] *Id.* at 2-12.

[61] *Id.*

[62] *Id.*

[63] *Id.*

[64] *Kaye, supra* note 31, at 110.

if the suspect's DNA were so common that it was very likely to match the crime-scene DNA. The analyst should be able to estimate the chance of a match if the suspect is the source of the sample compared to the chance of a match if someone other than the suspect is the source.

To make this comparison, the examiner must estimate the relative frequency with which the incriminating DNA fragments appear in the relevant population. That frequency usually is determined by comparing the crime-scene DNA profile with some reference data set. But because available databases contain only a very small proportion of the trillions of possible profiles, the frequency of a given profile must be estimated based on the frequencies of individual alleles. Making that estimate involves assumptions about the mating structure of the population.

Obviously, populations do not mate at random.[65] Many people are more likely to choose a mate from the same geographic area, ethnic group, or religion. Furthermore, in some societies people choose mates based on physical and behavioral attributes, such as height and personality. In fact, empirical studies have shown that the population of the United States includes different population groups and subgroups with different allele frequencies.[66] Thus, estimates of the frequency with which an allele appears in the population at large must take into account slight differences among various populations.[67] The National Research Council's second report, discussed in more detail in section III(D), suggests procedures that take into account such deviations from Hardy-Weinberg proportions.[68]

A related concept is that of *linkage equilibrium*. If mating and selection were truly random, and the entire population therefore had the same allele frequencies, then an analyst could calculate the frequency of a certain genotype simply by multiplying together the frequencies of each of the individual alleles that compose the genotype.

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[65] Random mating, the process of choosing mates independently of genotype at relevant marker loci and independently of ancestry, would produce the same proportions of alleles among all the world's population. The uniform allele proportions produced by truly random mating are referred to as the *Hardy-Weinberg (HW) proportions*. NRC II, *supra* note 3, at 4-2.

[66] *Id.* at 4-11; *Kaye, supra* note 31 at 128.

[67] The FBI and other forensic labs keep separate databases for whites and blacks. The FBI keeps two separate databases for Hispanics, one for those from the eastern U.S. and another for those from the west, because of their different origins. NRC II, *supra* note 3, at 4-6.

[68] *See, id.*, at Chapter 4.

Population geneticists would say that such a population is in linkage equilibrium. But in fact we know that loci on some chromosomes tend to be inherited together[69] and thus are in *linkage disequilibrium.* What effect does that fact have on calculating the frequency with which certain genotypes appear in the population? The answer is complex and, like the concept of *Hardy-Weinburg equilibrium,* has contributed to much of the controversy concerning DNA profiling. The National Research Council's second report examined empirical data on linkage disequilibrium suggested formulae for calculating frequencies that it claims are correct to within a factor of about ten-fold in either direction.[70]

IV. History of Forensic DNA Testing

Although DNA identification evidence and technology has evolved rapidly in the past decade and continues to change almost weekly, some understanding of its history provides context for understanding the current issues. This section highlights selected events relating to use of DNA evidence in court, including the genesis of the debate over "population substructures" and statistics.

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[69] Recall that when sperm and egg cells are made, homologous chromosomes line up and randomly exchange genes. Thus, genes that are close together on the same chromosome tend to be passed on together.

[70] NRC II, *supra* note 3, at Chapter 5.

A. Beginnings

The forensic use of DNA began in England in about 1984. In 1987 the British firm of Cellmark Diagnostics first opened a branch in the United States to introduce the British technology.[71] An American firm, Lifecodes Corporation, was founded in 1982 and began forensic DNA testing in 1987.[72] DNA testing first gained national attention in the United States after it was introduced as evidence in a 1987 Florida sexual assault case.[73] At first, the new technology largely caught defense attorneys off guard, and it was used in over one hundred cases with little or no resistance.[74] The media also reported favorably on it initially. In this atmosphere, commercial laboratories moved quickly to establish themselves. However, the labs typically sought to protect proprietary information and thus did not publish their scientific methods or subject them to peer review.[75] Many used different tools that produced results which could not be compared, largely lacked quality control systems and often had not performed sufficient validation studies.[76]

Defense attorneys first seriously challenged forensic DNA tests in court in the double-murder case of *People v. Castro*.[77] The defense systematically examined the genetic analyses and successfully argued that the DNA evidence should be excluded because the prosecution could not show that the generally accepted scientific theories and methodologies were correctly applied in the case at bar. This case also involved an out-of-court meeting between defense and prosecution scientific expert witnesses after they had testified.[78] All agreed that Lifecodes had failed to use generally accepted

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[71] COLEMAN & SWENSON, DNA IN THE COURTROOM: A TRIAL WATCHER'S GUIDE 4 (1994).

[72] *Id.*

[73] *Florida v. Andrews* (1987).

[74] COLEMAN & SWENSON, *supra* note 71, at 4.

[75] *Id.*

[76] *Id.*

[77] 545 N.Y.S.2d 985 (N.Y.Sup. 1989).

[78] COLEMAN & SWENSON, *supra* note 71, at 6.

scientific techniques in matching the blood found on the defendant's watch with that of one of the victims.[79]

B. FBI Lab Protocols

Also in 1987, the FBI and the National Institutes of Health began collaborative research to establish DNA identification techniques for the FBI.[80] The FBI established lab protocols and a standardized system used in almost all North American labs today.[81]

C. NACDL DNA Task Force

In 1989, the National Association of Criminal Defense Lawyers (NACDL) set up a DNA Task Force. Task Force members are Barry Scheck (telephone 212/406-2868) and Peter Neufeld (telephone 212/790-0368).

D. Report of National Academy of Science's National Research Council (NRC I)

In January of 1990, a fourteen-person panel began work on a study of forensic DNA for the National Academy of Science's National Research Council (NRC).[82] The report was funded by the Department of Justice. The panel released its report, DNA TECHNOLOGY IN FORENSIC SCIENCE, in 1992. The report called for standardization, mandatory accreditation and proficiency testing of lab workers. The report also proposed what turned out to be a controversial technique for calculating the statistical probability that a match between the defendant's DNA sample and the crime scene sample was attributable to chance.[83] This technique, called the *interim or modified ceiling principle*, assumed that population substructures exist, and was designed to

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[79] *Id.* Castro eventually pled guilty and so there was no review on appeal. *Id.* at 7.

[80] *Id.* at 6-7.

[81] *Id.* at 7.

[82] The National Research Council is the operating arm of the National Academy of Sciences (NAS) and the National Academy of Engineering, according to the NAS World Wide Web home page. The NRC is a private, non-profit institution that provides independent advice on science and technology issues under a Congressional charter.

[83] *See* NATIONAL RESEARCH COUNCIL, DNA TECHNOLOGY IN FORENSIC SCIENCE 82-85 (1992) (hereinafter NRC I)

yield estimates favorable to the defendant. The defense bar and some forensic scientists criticized the technique as an unscientific mix of elements combined in order to achieve consensus on the committee.[84] Others found the interim ceiling principle to be overly conservative in its incriminating power.[85]

E. Debate about Population Structures

The issue of how to estimate the population frequencies of specific DNA typing patterns has been controversial. The main questions concerned the adequacy of population databases on which frequency estimates are based (including selecting an appropriate comparison population), and the correct method for computing the estimated probability of a coincidental match of a DNA profile.

In December of 1991 the journal SCIENCE published opposing articles debating the proper way to estimate the proportion of people in the relevant population whose DNA fragments would be considered to match the fragments taken from the crime sample. Richard C. Lewontin of Harvard and Daniel L. Hartle of Washington University questioned the accepted methods of estimating the frequency with which a suspect's particular DNA profile would appear in the general population in an article entitled, *Population Genetics in Forensic DNA Typing*.[86] Lewontin and Hartle raised two issues: (1) how to define the data set to which the suspect's DNA pattern is compared; and (2) how to calculate the total probability of a random match from data from separate loci (in other words, how should data from separate VNTR loci be combined to give an overall probability of finding a given DNA profile that includes several loci).[87]

The authors first questioned whether a suspect's DNA profile should be compared to profiles in a single, homogeneous reference group. They said that common ethnic groups in the U.S. (for example, Poles and Italians) may have substantial differences in the frequencies with which multi-locus genotypes appear in their DNA

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[84] COLEMAN & SWENSON, *supra* note 71, at 11.

[85] *Id.*

[86] *Science*, 12/20/91, Vol. 254, Issue 5039, p. 1745.

[87] *Id.* at 1746.

profiles.[88] The authors cited demographic studies suggesting that members of racial groups in the U.S. do not mate randomly, and that they tend to marry within their race, ethnic group or hometown.

The authors also questioned the so-called "product" or multiplication rule for calculating probabilities across multiple VNTR loci. Under that rule, the probability of a random occurrence of a specific pattern of alleles in a DNA profile is calculated by multiplying the separate estimated probabilities of a random occurrence of each allele in the comparison population. The authors said that this rule only works if the DNA fragments are in "linkage equilibrium;" in other words, if no correlation exists between the genotypes found at different loci. In other words, the scientists questioned the standard forensic assumption that the bands in a DNA print are statistically independent.[89] They argued that some human populations (for example, the U.S. Asian population) may be "structured" so that certain common alleles would tend to occur together.[90]

Randjit Chakraborty (now at the University of Texas) and Kenneth K. Kidd of Yale defended DNA statistical analysis in an article entitled *The Utility of DNA Typing in Forensic Work*.[91] Citing principles of population genetic theory, they asserted that even a small amount of gene migration across ethnic and religious boundaries quickly homogenizes populations.[92] Also, they contested Lewontin and Hartle's idea that people tend to marry within groups, arguing that American demography for descendants of

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[88] *Id.* at 1748.

[89] At least one author subsequently has argued that "a small or moderate departure from equilibrium...may...make no meaningful difference to the match-binning frequencies...." *Kaye*, *supra* note 31, at 127. Kaye concluded that "it can be misleading to insist that 'the product rule ... can only be applied when the pairs of alleles are statistically independent....'" *Id.* The National Research Council's second report on forensic DNA evidence, released in 1996, predicted that "as population databases increase in numbers, virtually all populations will show some statistically significant departures from random mating proportions. Although statistically significant, many of the differences will be small enough to be practically unimportant." NRC II, *supra* note 3, at 1-11.

[90] Recall that the "product rule" used by forensic labs to calculate genotype frequencies assumed that the individual alleles that make up a genotype can be treated as statistically independent, and their frequencies multiplied to yield an overall frequency with which the profile would be expected to appear in the general population.

[91] *Science*, 12/20/91, Vol. 254, Issue 5039, at 1735.

[92] *Id.* at 1737.

Caucasian immigrants is closer to a "melting pot" than to a rigid subdivision.[93] Second, they referred to research on blood groups and protein markers that had failed to find any significant departure from the assumptions of statistical independence generally relied on for computing genotype probabilities.[94]

By 1994, two scientists who arguably represented the range of scientific debate, Bruce Budowle (a forensic scientist at the FBI) and Eric Lander (a molecular biologist at Whitehead Institute in Cambridge, Massachusetts) published a commentary in NATURE concluding that the remaining scientific debate about forensic DNA typing was purely academic and that for practical purposes the scientific issues all had been resolved.[95] With regard to the population substructure issue, the authors wrote that:

> FBI scientists have studied more than 25 distinct subpopulations, as well as 50 separate samples from the US population. The effort has yielded a remarkable database for examining allele frequency variation among ethnic groups. Reassuringly, the observed variation is modest for the loci used in forensic analysis and random matches are quite rare.[96]

They added that while population substructure could matter in principle, its effect in practice is not significant.[97] They concluded that whatever controversy existed had been resolved by the [first] NRC report, the TWGDAM[98] guidelines, and the extensive scientific literature.[99]

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[93] *Id.*

[94] *Id.*

[95] Lander & Budowle, *DNA Fingerprinting Dispute Laid to Rest*, 371 NATURE 735-738 (10/27/94).

[96] *Id.* at 738.

[97] *Id.* They pointed out that different theories for estimating genotype frequencies produce estimates differing only by two orders of magnitude: "whether the population frequency of a typical four-locus genotype should be stated, for example, as .00010 or .0000010." *Id.*

[98] TWGDAM is the Technical Working Group on DNA Analysis Methods. It is sponsored by the FBI and the Department of Justice.

[99] Lander & Bowle, *supra* note 94, at 735.

F. NRC II

Also in 1994 the National Academy of Sciences convened a second panel (NRC II) to update its first report. The NRC released its second report, THE EVALUATION OF FORENSIC DNA EVIDENCE, in May of 1996.[100] The report concluded that the science behind DNA forensics is valid, but recommended new ways of interpreting DNA evidence to assess how likely it is that two matching samples came from different people. Specifically, the report addressed the problem that people of the same race are more likely to have a similar genetic makeup than people of different races. The report acknowledged the problem that people of the same race are more likely to have a similar genetic makeup than people of different races, but concluded that the differences were too small to be important.[101] The report advanced a new combination of formulae to calculate the likelihood that a DNA match could be coincidental, and recommended ways to protect suspects from false incrimination from improper handling of evidence.

With regard to statistical interpretation of DNA evidence, the report concluded that the interim ceiling principle endorsed by the first NRC report is no longer necessary because data bases of DNA profiles are providing an ever-clearer picture of the genetic makeup of different racial and ethnic groups.[102] The report also recommended a set of statistical calculations that takes population structure into account.[103] Specifically, it recommended that if the race of the person who left the evidence sample is known, calculations should use the data base for that person's race. The report also offered formulae to use if the race of the person who left the evidence is not known, or if the suspect comes from a segment of the population for which the data base is insufficient.

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[100] Pre-publication copies are available from the National Academy Press: 1-800-624-6242. The report costs $40 plus shipping. For more information, refer to the National Academy of Science's World Wide Web page: http://www/nas.edu. The study was funded by the National Institute of Justice, the State Justice Institute, the National Science Foundation, the National Institutes of Health, and the Department of Energy.

[101] NRC II, *supra* note 3, at 1-11.

[102] *Id.* at O-27.

[103] *Id.*

With regard to handling evidence, the report concluded that whenever feasible, forensic samples should be divided into two or more parts as soon as possible after collection, and the unused parts should be retained for an independent retest by personnel not involved in the first test and preferably in a different laboratory.[104] The report concluded that estimating the likelihood of laboratory error is unfeasible. Instead, the report recommended improving laboratory performance and accountability, and encouraging independent retests.[105]

Finally, the report recommended behavioral research to identify reasons why a potential juror might misinterpret evidence on DNA profiling. The research also should assess how well various ways of presenting expert testimony on DNA could reduce juror misunderstandings.[106]

V. Evaluating Forensic DNA Evidence in Alaska Courts

Like other state trial courts, Alaska's courts have had to grapple with the use of DNA evidence in criminal cases. Before the Alaska Court of Appeals or the Alaska Supreme Court had ruled on the admissibility of DNA evidence, the legislature passed a law admitting DNA evidence in Alaska's courts. However, the statute does not apply to cases before July 15, 1995, and it does not discuss how the evidence should be presented. Thus, issues arguably remaining for Alaska's appellate courts include whether the statute will apply to newly developed DNA testing methods, and whether and how to interpret the probity of a match to the trier of fact.

A. Admissibility of DNA Evidence

The Alaska Legislature resolved the issue of the future admissibility of DNA evidence during the first half of the 1995-96 legislative session. Under AS 12.45.035(a):

> evidence of a DNA profile is admissible to prove or disprove any relevant fact, if the court finds that the technique underlying the evidence is scientifically valid. The admission of the DNA profile does not require a finding of general

[104] *Id.* at ES-3.

[105] *Id.* at ES-4.

[106] *Id.* at ES-7.

> acceptance in the relevant scientific community of DNA profile evidence.

This statute took effect on July 11, 1995.[107]

Before the statute went into effect, however, the court of appeals already had affirmed the admission of DNA evidence in *State v. Harmon.*[108] In that case, the defendant was convicted of sexual assault and murder after a trial at which the judge admitted results of DNA testing (RFLP and PCR analysis performed by the FBI and Alaska State Crime Labs). The evidence tended to incriminate the defendant, who appealed the admissibility of the DNA evidence, among other things. In affirming the trial court's decision that the DNA evidence was properly admissible, the Alaska Court of Appeals said that courts should follow the existing *Frye*[109] test in determining the admissibility of DNA evidence.[110] The Court of Appeals did note, however, that the Alaska Supreme Court could reevaluate that standard in light of *Daubert.*[111]

The statute also amended Evidence Rule 703 to the extent that the rule would limit the admissibility of DNA profile evidence as a result of the application of the standard previously adopted by the Alaska Supreme Court in *Pulakis v. State*, that

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[107] *See* ch. 7, SLA 1995 (attached).

[108] 908 P.2d 434 (Ak App. 1995).

[109] *Frye v. United States*, 293 F. 1013 (D.C. Cir. 1923). The supreme court adopted the *Frye* test in Alaska in *Pulakis v. State*, 476 P.2d 474 (Alaska 1970).

[110] Because *Harmon* was tried before the effective date of the DNA admissibility statute, the court of appeals reviewed the trial judge's decision that the evidence was properly admissible. The trial judge used the two-step analysis prescribed in *Contreras v. State*, 718 P.2d 129, 135 (Alaska 1986). Under that two-step analysis, the trial judge first defined the relevant scientific community, and then determined whether general consensus about the questioned procedure existed within the scientific community. In *Harmon*, the trial judge defined the relevant scientific community as molecular biologists, DNA forensic scientists, biochemists and population geneticists. *Harmon*, 908 P.2d at 442. The judge then found a general consensus in the scientific community that DNA testing is reliable. *Id.* The trial judge rejected the analysis in *People v. Castro*, although she also concluded that the labs that tested the DNA evidence in Harmon's case followed standard, generally accepted procedures. *Id.* at 440. The court of appeals said that this finding made it unnecessary to decide whether *Castro* applied to the *Harmon* case. *Id.*

[111] *Daubert v. Merrell Dow Pharmaceuticals, Inc.,* 113 S.Ct. 2786 (1993). *Harmon*, 908 P.2d at 439 n.5. A case currently being briefed to the supreme court, *State v. Coon*, (S-6893) concerns the admissibility of novel scientific evidence (namely voice spectrogram evidence). The court was asked to decide whether *Frye* or *Daubert* should govern the admission of novel scientific evidence.

requires a finding of general acceptance of scientific evidence in the relevant scientific community as a precondition of admission of scientific evidence. Thus, the Act apparently means that trial courts need not hold *Frye* hearings and make findings of general acceptance in the relevant scientific community before admitting DNA evidence. It also may mean that DNA evidence will be admissible despite potential problems with population substructures.[112]

The statute does not distinguish between different types of DNA testing. Thus, it apparently would render the newer PCR (polymerase chain reaction) analysis admissible without *Frye* hearings,[113] and perhaps would render admissible other types of analysis currently under development.

B. Legal Framework for Evaluating Probity of DNA Evidence

Although AS 12.45.035 now governs the admissibility of DNA tests after July 11, 1995, the statute does not tell judges how the DNA profile should be presented or interpreted to the finder of fact. In *Harmon*, the trial court and the court of appeals discussed an acceptable way to report the probity of the match between the defendant's DNA and the DNA sample at the crime scene.

The experts who testified in *Harmon* calculated the probability that the match occurred by coincidence using the Modified Ceiling Principle, computed in a manner consistent with the [first] NRC Report. The trial court found that the calculations were admissible because in May of 1993 the relevant scientific community generally accepted that method for providing a statistical frequency of a random match.[114] The trial court further found admissible the statistical frequency calculations for the RFLP matches, which were computed using the Modified Ceiling Floating Bin Method.[115]

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[112] *Harmon,* 908 P.2d at 441.

[113] A case currently awaiting decision by the court of appeals, *Brodine v. State* (A-5837), also raises the issue of whether the newer PCR testing has gained sufficient acceptance in the relevant scientific community to be admissible in court. The relevance of this decision may be limited, however, because it may not be distinguishable from *Harmon*, and because future questions of the admissibility of PCR tests probably will be governed by the statute.

[114] Trial Court Opinion at paragraph 20.

[115] *Id.* at paragraph 22.

The court of appeals reviewed the trial court's decision under *Frye* and *Contreras*. It found the relevant scientific community to be human population geneticists and agreed with the trial court judge that the population statistical analyses of the PCR typing and the RFLP matching are generally accepted within the scientific community.[116]

At least two other cases currently on appeal will resolve additional statistical issues. One case awaiting decision by the court of appeals, *James Charles v. State* (A-5701), involves population substructures in Alaska. The appellant in that case claimed on appeal that the modified ceiling approach should not have been used for calculating population frequencies in his case, because the reference population was physically isolated (the case occurred in the village of Saxman). A second case awaiting decision, *Wyatt v. State* (A-5291), also involves a challenge to the modified ceiling approach. In that case, the trial court admitted DNA evidence, and the state's expert applied the modified ceiling approach to calculate population frequencies. The defense claimed on appeal that the state's expert improperly applied the modified ceiling approach.

A third case currently before the court of appeals, *Brodine v. State* (A-5837), involves the broader issue of how and whether evidence of a match should be interpreted to the finder of fact. The appellant in that case claimed that the trial judge erred by admitting evidence of a match while excluding evidence of population frequency estimates. Most courts require that the evidence of a "match" be admitted with statistical calculations estimating the probability that the match could have been a coincidence.[117] These calculations generally take the form of estimates of the probability an individual other than the defendant has DNA patterns that match at the sites examined.

C. Other Issues Affecting Probity of DNA Evidence

[116] *Harmon*, 908 P.2d at 442.

[117] The court of appeals wrote in *Harmon* "...the usefulness of DNA testing in criminal litigation depends on being able to identify what percentage of the population carries the particular gene whose presence was tested for." *Id.* at 441. The NRC I report also recommended that interpreting a DNA test include a valid scientific method for estimating the probability that a random person by chance matches the forensic sample at the sites examined. NRC I, *supra* note 83, at 9. The report said, "To say that two patterns match, without providing any scientifically valid estimate...of the frequency with which such matches might occur by chance, is meaningless." *Id.*

Assuming that AS 12.45.035(a) and case law make DNA profile evidence admissible in Alaska's courts, what evidentiary issues remain? Depending on the facts of the case, a number of technical issues could affect the weight of the evidence of a DNA profile match.[118]

1. Sample Quantity & Quality. VNTR analysis requires a suitable sample of DNA. The ultimate issue is whether problems with sample quality or quantity could cause a false positive result, or merely an inconclusive or uninterpretable result.

The crime sample must contain enough DNA of sufficiently high molecular weight to allow isolation of longer fragments.[119] For smaller samples, PCR analysis may be more appropriate than RFLP analysis.

How the sample was handled also could affect test results. Facts to watch for include: opportunities for lab mislabeling; sources of possible lab contamination (of particular concern with PCR amplification, because even a small fragment from a foreign source may be amplified many times); and deviations from the lab's written procedures.

Contamination is an important issue for PCR (polymerase chain reaction) analysis. Because this analysis makes multiple copies of a small sample fragment, it is important that the sample not be contaminated. Variations in length, sequence difference, or contamination with non-DNA material can cause differential amplification in an evidence sample.[120] Also, the individual loci used in current PCR-based tests are less polymorphic than VNTR loci; therefore, the multilocus genotype frequencies from PCR-based tests typically are not as small as those in VNTR typing.[121]

2. Quality Control and Quality Assurance. Regular lab proficiency testing and auditing of laboratory operations are recommended by the National

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[118] Most of the potential issues discussed below are taken from the Federal Judicial Center's REFERENCE MANUAL ON SCIENTIFIC EVIDENCE (1994)(hereinafter FJC REFERENCE MANUAL).

[119] *Id.* at 287.

[120] GENETIC WITNESS, *supra* note 5, at 70.

[121] NRC II, *supra* note 3, at 6-11.

Research Council.[122] Proficiency tests are run on specimens submitted to the lab in the same form as evidence samples. Audits independently review lab operations to determine whether the lab is performing according to a defined standard.[123] Are proficiency tests internal or external? Does the error rate include false negatives (incorrect exclusions) and inconclusive results, or just false positives (incorrect inclusions)?

A legal issue (which arose in the *Simpson* case) was whether to factor the lab error rate into any DNA profile frequencies or to report it out separately. As discussed in section III, above, the second NRC report concluded that proficiency test results should not be combined with the estimated frequency of an incriminating profile to yield the probability that a lab would report that the DNA from a person selected at random contains the incriminating profile. The report reasoned that auditing and proficiency testing cannot be expected to give a meaningful estimate of the probability that a particular lab had made such an error in a specific case.[124]

3. Validity of Test. Validity is the probability that a test will correctly identify true matches and true non-matches.[125] The important issue for the courts is the probability that the lab will incorrectly declare a match (i.e., what is the lab's false positive error rate?).

4. Calling it a Match. The question is whether distinct bands in the suspect sample appear to be in the same position as bands in the crime sample. If the bands do not appear to match, the question is whether the test excludes the suspect/defendant as the source of the crime sample, or if the test is inconclusive because of some problem with lab procedures (insufficient quantities, partially degraded samples, using too much sample).

Inconsistencies in gel composition or variations in the electric field can cause a gel to "smile," or create contortions in the lanes of DNA. Foreign matter in the DNA sample (from the restriction enzymes or the original forensic material, for instance)

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[122] *See id.* at 3-1.

[123] *Id.* at 3-4.

[124] *Id.* at ES-4.

[125] GENETIC WITNESS, *supra* note 5, at 7.

or impurities in the gel can diminish the distinctness of banding patterns and produce artifacts that can be misleading or misinterpreted.[126]

Labs use several measurement standards to determine that the bands are similar enough to declare a match. One scholar noted some confusion as to what rule different laboratories actually use.[127] Whatever the exact numbers, however, the lab normally defines the match window as "the range within which two bands can be declared to match."[128] The range is defined by "observed differences seen in repeated measurements of DNA fragments of the same length."[129]

5. Estimating the Frequency of a Profile. This subject was discussed earlier, in section IIIC; however, some of Alaska's populations may present unique challenges for estimating allele and genotype frequencies. For some groups, including several American Indian and Inuit tribes, there are insufficient data to estimate frequencies reliably, and even the overall average might be unreliable.[130] The NRC concluded in its second report that pending development of more extensive databases, the analyst could use allele frequencies from other closely related groups.[131] The report offers a special formula to use when calculating the profile freqency.[132]

VI. Alaska Scientific Crime Detection Laboratory

The Alaska lab performs both PCR and RFLP testing. The lab has used polymarkers since May of 1995. Since January of 1996, the lab has been able to produce a seven-marker profile, meaning that it can test genes at seven different loci. The lab follows the TWGDAM standards for analysis of DNA samples.[133]

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[126] *Id.* at 115.

[127] *Kaye*, *supra* note 31, at 110 n.45.

[128] *Id.* at 111.

[129] *Id.*

[130] NRC II, *supra* note 3, at 4-37.

[131] *Id.* at 4-35.

[132] *Id.* at 4-37.

[133] One of the lab's staff is a member of TWGDAM.

Much of the lab's work is performed at the investigative stage of a criminal case, as opposed to trial work. In fact, the Alaska lab excludes approximately 40% of the suspect samples sent to it for analysis. Staff estimate that they performed approximately 120 analyses in 1994, and that they had testified in court about a dozen times by the end of 1995.

Crime lab personnel are trained by private labs such as Cellmark, and by the FBI. Proficiency testing is performed according to the standards of the National Society of Crime Lab Directors.

The crime lab has been randomly sampling people and typing their blood since 1989. The lab has collected over two hundred samples of individuals from the Bethel and Barrow areas, and this work is continuing. Staff recently published an article summarizing the results of their study in the JOURNAL OF FORENSIC SCIENCE.[134] According to lab personnel, preliminary results suggest that although there may be slightly less variability among Alaska Natives than among Caucasians, enough variability exists to make DNA tests probative.

VII. Conclusion

DNA profiling technologies will continue to change and the legal issues presented also will change. Although the legislature has decided the admissibility of DNA evidence in Alaska's courts, a number of issues about the use of DNA evidence remain undecided. This report has tried to address a few of them; no doubt readers can identify many more. The Council encourages readers to share their suggestions and comments on how to make future versions of this report more useful.

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[134] Volume 41, No. 3, pp. 478-84 (1996).