Forensic Magazine Digital Edition

30 Years of DNA Technology

Monday, 03/13/2017 - 3:18pm by Vernon J. Geberth, M.S., M.P.S.

The greatest advance in criminal investigations since fingerprints is the application of DNA technology to the criminal justice process. The proper collection and analysis of DNA can convict the guilty and exonerate the innocent. Procedural improvements have made the collection of DNA evidence more efficient and reliable. Advances in science allow forensic scientists to identify DNA samples from hair, bone, skin and tissue, and ever smaller amounts of blood and other body fluids.

Today, biological evidence retrieved from a victim or crime scene can be examined at its most fundamental level—the deoxyribonucleic acid (DNA) molecule. DNA profiling can be used to:

- Establish the probative link between evidentiary DNA and that of the possible offender's DNA
- Identify whether the DNA in question is human or non-human
- Identify the potential number of contributors to a DNA sample
- Identify the gender of the contributor to a DNA sample
- Establish the genetic relatedness of an unknown profile to known samples (paternity and maternity testing)
- If the DNA is non-human, identify the species of the DNA

1986: the beginning

The first application of DNA science to a criminal investigation was conducted in England by Dr. Alec Jeffreys, a British geneticist who developed the techniques for DNA fingerprinting and DNA profiling, which are now used worldwide in forensic science.

There had been a series of rape-murders in Leicestershire in 1983 and 1986 and the authorities had a suspect named Richard Buckland in custody because he had confessed to the crime.

In 1986, police asked Jeffreys to forensically link this suspect to the biological evidence retrieved from two female victims. Jeffreys' DNA tests exonerated their primary suspect. The British police then conducted a genetic dragnet by collecting blood samples from more than 4,000 male inhabitants from the area where the crimes had taken place. The hunt revealed that an individual named Colin Pitchfork had asked a friend to substitute a blood sample for him. Jeffreys was able to positively link Pitchfork to the series of rape-murders through DNA profiling. In 1987, Pitchfork became the first person in the world to be identified, captured and successfully prosecuted as a result of DNA evidence.

The first use of DNA profiling in America took place in Orange County, Florida during a rape case.

In 1987, Tommy Lee Andrews was convicted of rape after DNA tests matched his DNA from a blood sample with that of semen traces found in a rape victim.

I supervised the first case in New York state, People v. Castro, which was the third application of DNA technology in the United States. It was also the first case that seriously challenged a DNA profile's admissibility. The DNA evidence was a bloodstain on Castor's watch that had been analyzed for a match to the victim's.

Case example: Vilma Ponce

In February 1987, Vilma Ponce, who was eight months pregnant, and her 2-year-old daughter Natasha Otero were found slaughtered in their apartment in Bronx, New York. Vilma was laying on the living room floor. She was nude from the waist down and her maternity top was pulled up to expose her stomach and lower torso. The woman had been stabbed 61 times. Her

daughter, Natasha, had been stabbed 12 times. The fetus had also received three stabbing injuries through the womb. I was present at the scene as the commanding officer of the Bronx Homicide Task Force. During the crime scene process, we noticed bloody sneaker prints throughout the apartment, as well as droplets of blood over the sneaker prints and numerous other blood spatters. A single drop of blood found near the front door of the apartment suggested that the offender might have cut himself.

I decided to have the blood evidence taken to a private lab—Lifecodes Corporation—for analysis using a new technique called the DNA print identification test. Within 48 hours, we had developed a viable suspect, who had a fresh cut on his hand as well as dried blood on his watch. The suspect, Joseph Castro, agreed to be interviewed and provided an extensive alibi for the day of the murder. He also gave the detectives permission to have his watch examined.

This illustration depicts the victim, Vilma Ponce, who was 8 months pregnant lying on the living room floor. She was nude from the waist down and her maternity top was pulled up to expose her stomach and lower torso. She had been stabbed 61 times, with three penetrating her unborn child. Illustration: Courtesy of Medical Legal Art.

This first generation of DNA technology was RFLP multilocus type of DNA analysis, which positively identified Castro as the killer. He was arrested on March 5, 1987—one month after the murders. The DNA tests done in this case also matched another murder that had occurred one year earlier in a different building Castro was assigned to clean.

People v. Castro was the first case that seriously challenged a DNA profile's admissibility. In a 12-week pretrial hearing, the New York Supreme Court exhaustively examined numerous issues related to the admissibility of DNA evidence. Attorney Barry Scheck, who at the time represented the Public Defender's office and is now the director of the Innocence Project, attacked the science of DNA, which resulted in a number of pretrial hearings that were required to determine whether the testing laboratory's methodology was substantially in accord with scientific standards and produced reliable results for jury consideration. The court ruled the DNA tests could be used to show that blood on Castro's watch was not his; but the tests could not be used to show that the blood was that of his victims. Regardless, the defendant was found guilty in 1989.

The important implications of this early case include:

- DNA technology was proven right despite defense efforts to discredit the science
- The defendant admitted the blood was that of the victim, as Lifecodes' tests had suggested
- The judge endorsed the basic science of DNA profiling and allowed it to be admitted for exclusion
- Procedural problems led the judge to refuse the admission of evidence that the blood matched the victim. Those problems have since been corrected and rectified.

In the 1989 case of People vs. Castro in New York the defense literally put DNA fingerprinting on trial. They were able to disclose severe insufficiencies in the technical protocols and especially in the DNA evidence interpretation. This raised doubts on the scientific and evidentiary value of forensic DNA fingerprinting. However, the science prevailed.

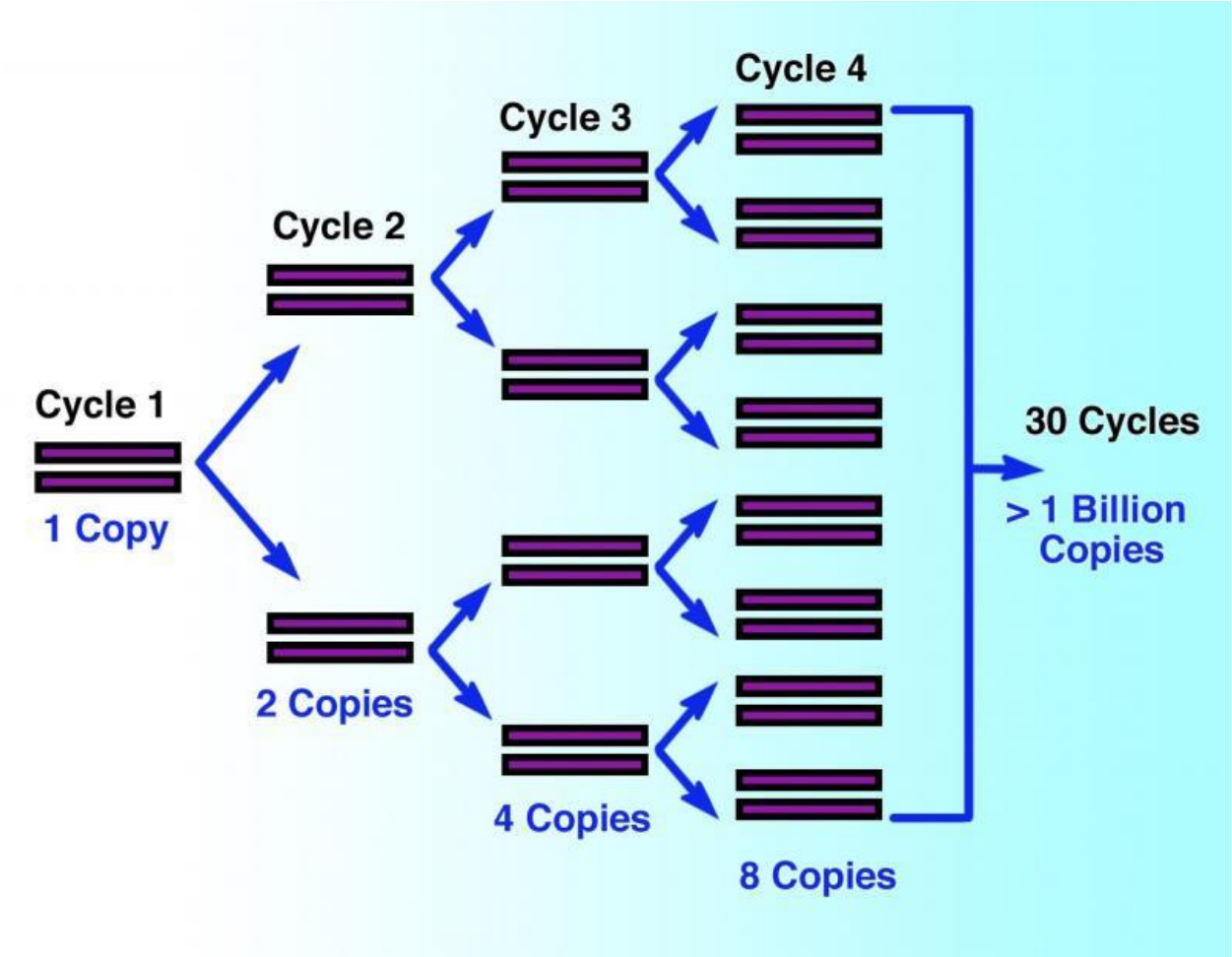
1987 into the 1990s

RFLP technology of 1986, which was the Model T Ford of DNA analysis, involved the process of identifying polymorphic regions that are unique to each individual. These variable number of tandem repeats, or VNTRs, contained fairly large repeat units with allele sizes being thousands of base pairs long.

Starting in the early 1990s, DNA fingerprinting methods based on RFLP analysis were gradually supplanted by methods based on PCR because of the improved sensitivity, speed and genotyping precision. Microsatellites, usually referred to as short tandem repeats (STRs), were found to be ideally suited for forensic applications. STR typing is more sensitive than single-

locus RFLP methods, less prone to allelic dropout than VNTR systems, and more discriminating than other PCR-based typing methods.

In 1993, Dr. Kary Mullis received a Nobel Prize for his work during the 1980s that resulted in the invention of the polymerase chain reaction (PCR), which mimicked the cell's ability to replicate DNA—enabling scientists to take small samples of DNA and essentially copy it a million fold. PCR technology has similar basic steps to RFLP—extraction, amplification and detection. Additional research identified much smaller VNTRs—which were only a few base pairs long—that, when coupled with PCR, became the foundation of STR technology.



Schematic representation of the DNA duplication process. At the completion of a 30 cycle reaction, greater than 1 billion copies are duplicated. Illustration: Courtesy of Medical Legal Art; Copyright 2005 Medical Legal Art; and Practical Homicide Investigation: Tactics, Procedures and Forensic Techniques 5th Edition CRC Press, 2015 p. 716

PCR amplification allows production of many copies of the region of DNA interest. PCR works like a "molecular xerox machine." Millions of copies of a particular sequence of DNA can be made in about 3 hours in a thermal cycler. This is great for forensic science when there is very little DNA to start with. Initially, DNA samples that were small or degraded were beyond the reach of DNA-typing techniques.

Case example: O.J. Simpson

In one of the most publicized criminal homicide cases in the country, O.J. Simpson's DNA was linked to the murder of Nicole Brown Simpson and Ron Goldman in 1994. It set off a media storm as the defense tried to undermine DNA technology. The RFLP testing indicated there was at least a 999,999 chance in a million that two samples submitted to the court matched O.J. Simpson. Polymerase chain reaction tests performed on a second drop in the trail of blood leading from the crime scene also indicated a match with O.J. Simpson's blood.

But, his lawyers contended the blood sample collection and analysis process was so careless that tampering could not be ruled out, and thus DNA test results could not be believed. The defense brought in experts to challenge police laboratory techniques and put forensic science on trial. The end result was that DNA labs become more proficient and all DNA testing that was introduced into court had to be from laboratories that were accredited.

1990: CODIS

The national DNA database known as the Combined DNA Index System (CODIS) began as a 14-state pilot study in 1990. The program expanded nationally as a result of the 1994 DNA Identification Act (Public Law 103 322), giving the FBI legal authority to establish a DNA database for the nation's criminal justice system. Today, all 50 states participate in the CODIS program, which is comprised of two main DNA indices; a forensic index that contains DNA profiles developed from crime scene-related evidence, and a convicted offender index containing DNA profiles from qualified convicted offenders.

The FBI, which funds the program, requires that all participating laboratories utilize the same 13 STR loci. CODIS is structured as a three-tier hierarchy: a local DNA index system (LDIS), a state DNA index system (SDIS) and a national DNA index system (NDIS). With this approach, each participating laboratory can manage its profiles in accordance with its legal requirements and, at the same time, compare its profiles electronically with other local and state laboratories in addition to the federal laboratory.

As of January 2017, the National DNA Index contains 12,647,876 offender profiles, over 2 million arrestee profiles and more than 700,000 forensic (casework) profiles. As of November 2016, CODIS has produced approximately 355,535 DNA database "hits" and aided nearly 340,552 investigations.

In addition to CODIS, all 50 states maintain DNA databases. The types of profiles that are included vary from state to state.

Single nucleotide polymorphisms (SNPs)

In order to make new cells, an existing cell divides itself in two. But first it copies its DNA so the new cells will each have a complete set of genetic instructions. Cells sometimes make mistakes

during the copying process—kind of like typos or mutations. These typos lead to variations in the DNA sequence at particular locations called single nucleotide polymorphisms, or SNPs.

SNPs are used to perform DNA profiling of the Y chromosome. In addition to the 13 CODIS loci, a number of laboratories have developed multiplexes of SNPs so that male DNA can be individually typed.

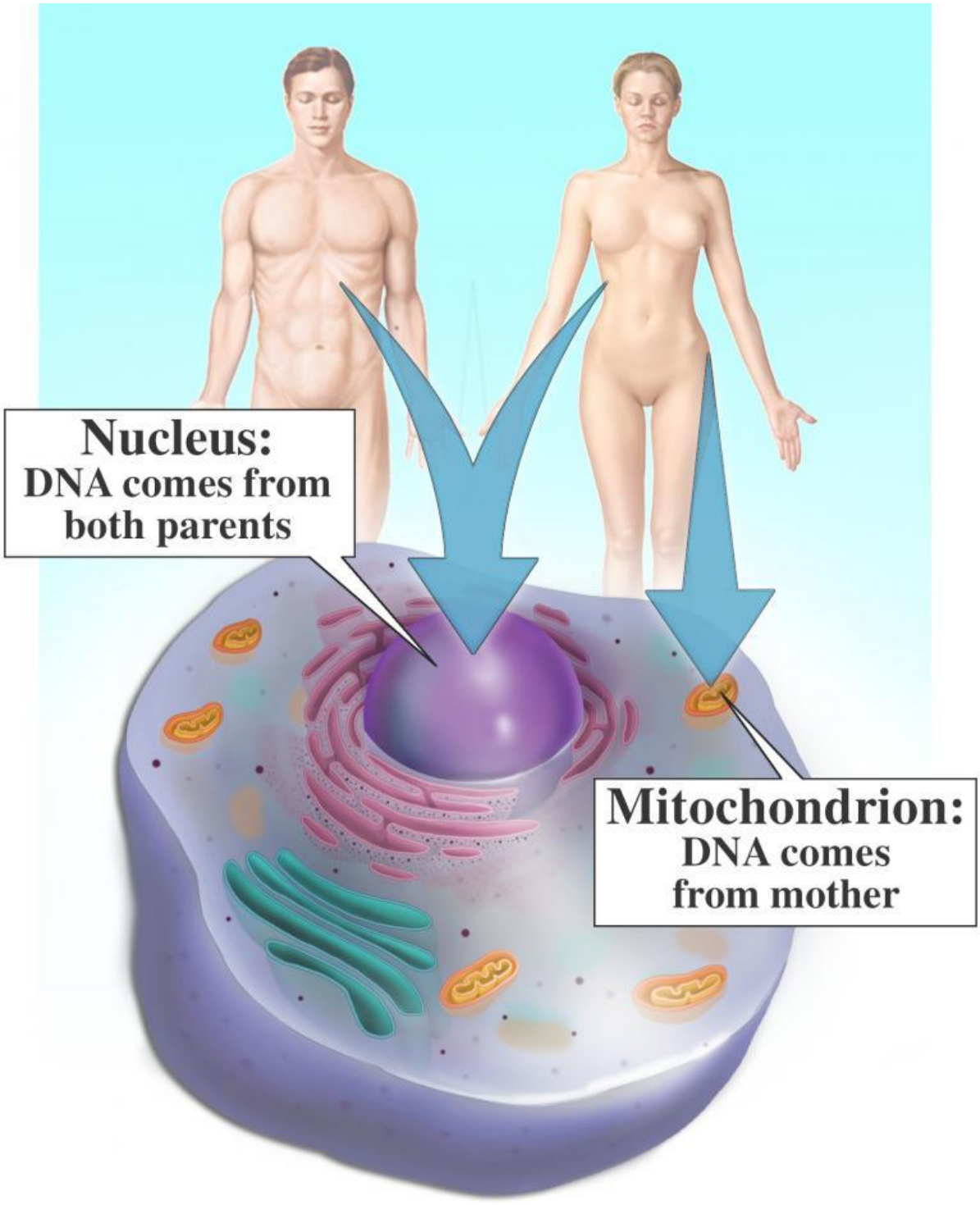
Another database: family

In addition to directly testing known samples from family members to compare to an evidence sample, an unknown sample can also be searched against DNA databases of known individuals in an effort to detect possible relatedness-based connections. This type of search, which utilizes different search parameters, has been used primarily to generate investigative leads. Proponents say it will help identify suspects who wouldn't be in the database if not arrested. Familial DNA is presently being utilized by California, Colorado, Virginia, New York, Ohio, Texas, Florida, Michigan, Utah, Wisconsin and Wyoming.

Critics of the technique argue that, in addition to violating individuals' privacy and civil rights, familial DNA testing is a form of racial profiling because the CODIS-known offender database contains a disproportionately large number of African Americans. However, it can be argued that having one's profile included in an offender database is simply a consequence of conviction, with no bias placed on demographics.

Still, due to these concerns, the FBI has resisted performing familial DNA searches.

Familial DNA searches are routinely performed in Europe, Australia and New Zealand. British authorities have conducted dozens of investigations using familial DNA searches since 2002.



Contribution of parents to DNA of offspring. Only the mother contributes mitochondrial DNA, of which thousands of copies can be found in a human cell. Illustration: Courtesy of Medical Legal Art; Copyright 2014 Medical Legal Art; and Practical Homicide Investigation: Tactics, Procedures and Forensic Techniques 5th Edition CRC Press, 2015 p. 704

Case example: The Grim Sleeper

The serial killer known as the "Grim Sleeper" was active in the Los Angeles area between 1985 and 1988 and was responsible for the murders of seven women and one man during that time period. An additional female victim was raped but survived. Thirteen years passed until the Grim Sleeper became active again with additional murders committed in 2002, 2003 and 2007. Along with a 911 voice recording of the killer, a common DNA profile was developed in six of the 12 cases, and additional cases were also linked by ballistics evidence. The unknown profile was searched through the CODIS system national offender and casework databases with negative results. The Grim Sleeper became the first case submitted for a familial search in California. The first search, conducted in 2008, did not produce any matches.

Eighteen months later, the search was again conducted as the convicted offender database had grown by several hundred thousand offenders by that time. During this search, a hit was generated and further testing showed that the Y-STR profile of the identified individual was an exact match to the unknown offender. A records-based search along with DNA relatedness results suggested that the individual in the database was the son of the offender. Subsequent investigation showed that the individual's father, Lonnie Franklin, Jr., had resided close to several of the murder sites during relevant time periods.

Undercover officers followed Lonnie Franklin, Jr. into a restaurant and recovered several items used by him. The DNA recovered from these items matched the DNA profile of the Grim Sleeper. Franklin, Jr. was arrested and charged with 10 counts of murder and one count of attempted murder. He has subsequently been linked to six additional homicides and awaits trial.

Ancestry determination

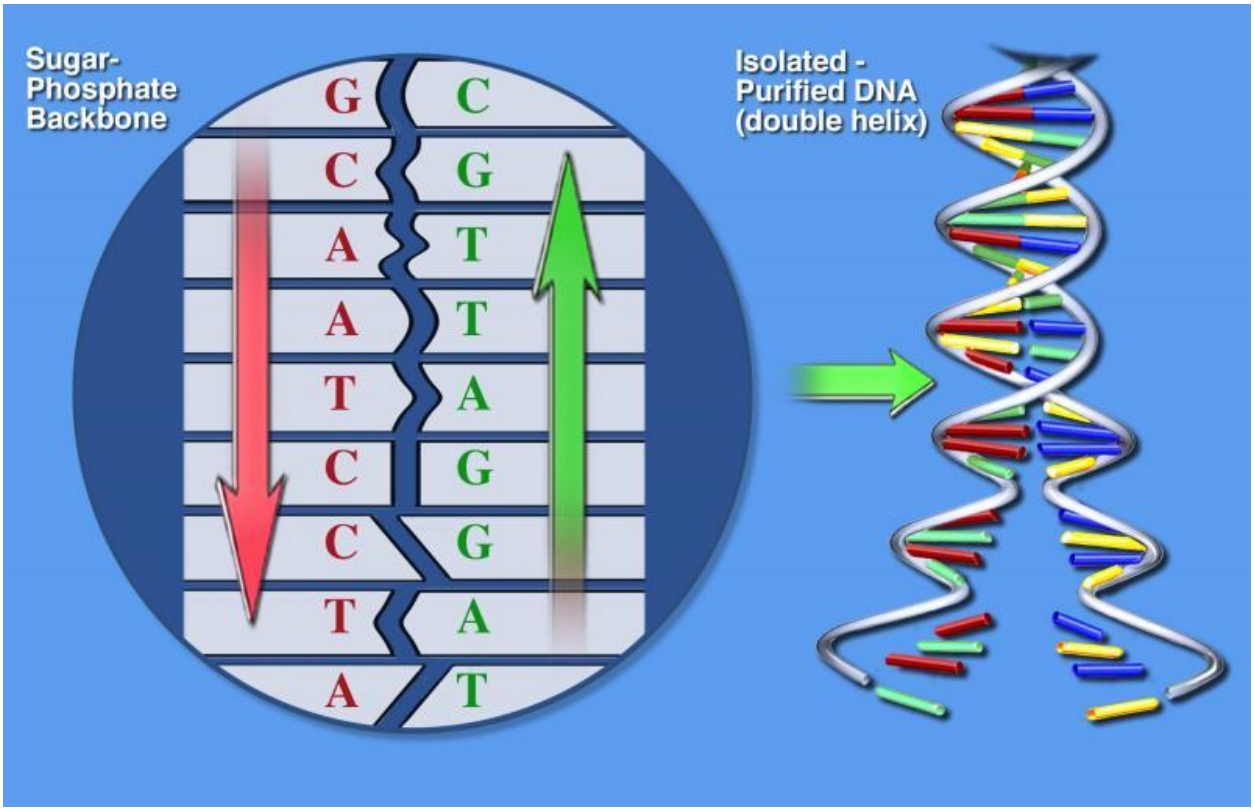
Traditional forensic DNA profiling compares DNA profiles from crime scene evidence to profiles from a known source, such as a cheek swab from a suspect. Unknown profiles can be searched in the CODIS database directly or through familial searches. However, what if all of these searches are negative? One additional type of available DNA testing is FDP (forensic DNA phenotyping)—that is, using DNA to create a genetically based description of the unknown subject's appearance, including possible ancestry.

Biographical ancestry is the heritable component of race and is based on Ancestry Informative Markers (AIMs). AIMs are genetic markers that are more common in one human population than in others. At the original time of this science it was determined that there are four main continental population groups: Sub-Saharan African (population groups with roots in the sub-saharan region of Africa); East Asian (includes Japanese, Chinese, Koreans and Pacific Islanders); Indo-European (includes Europeans, Middle Easterners and South Asians) and; Native American (those who migrated to inhabit North, South and Central America).

Case example: the Baton Rouge serial killer

Beginning in 2001, a serial rapist and murderer was active in Baton Rouge, Louisiana. The offender raped his victims and killed them by a variety of methods, including strangling, stabbing and beating. Through STR DNA profiling, the unknown subject was linked to six murders. The FBI profile was that of a white male, 25 to 35 years of age who was a blue collar worker. The profile also suggested that the unknown subject would wish to be seen as someone who was attractive and appealing to women; in reality, however, his level of sophistication in interaction with women would be low and women would describe contact with him as "awkward."

DNA evidence from the offender was submitted to DNAPrint Genomics. The results from DNAPrint indicated the unknown subject was 85% Sub-Saharan and 15% Native-American. Derrick Todd Lee was subsequently arrested for the offenses and linked to the crimes through DNA evidence matches. Lee did not fit the FBI offender profile: he was black, not white as the profile indicated. Lee was described as a charming, outgoing extrovert and had a reputation as a "ladies' man." Lee was convicted and awaiting execution on death row in Angola State Prison when he died a natural death in January 2016.



DNA in its native helical form. Illustration: Courtesy of Medical Legal Art; Copyright 2014 Medical Legal Art; and Practical Homicide Investigation: Tactics, Procedures and Forensic Techniques 5th Edition CRC Press, 2015 p. 701

2000s: modern roots

The STR class of polymorphisms has become the backbone of modern forensic testing. Short tandem repeats loci are polymorphic genetic markers that are well distributed throughout the human genome. The advantage of STR technology is that the small size of STR loci improves the chance of obtaining a result.

Fluorescent detectors used in this type of analysis identify 13 different loci that can be analyzed simultaneously. These 13 loci form the basis of CODIS.

"John Doe" warrants

In addition to facilitating cold hits, DNA database legislation has given rise to a new type of arrest warrant termed "John Doe," or DNA warrants, because the warrant is issued not for a named person, but for a genetic code identified as part of a criminal investigation for which no suspect has been identified and no database match has yet been found. In September 1999, a district attorney in Wisconsin became one of the first prosecutors to obtain a warrant and file criminal charges against a man identified in the warrant solely by his DNA profile. The primary purpose of these warrants is to toll the statute of limitations in cases of violent crimes. Many states have successfully convicted offenders based on John Doe warrants and several, such as Wisconsin, have passed legislation legalizing their use.

Case example: BTK—Dennis Radar

The BTK killer had claimed credit for seven murders that occurred between 1974 and 1977. Ultimately, BTK was responsible for 10 murders; however, authorities would not know about the additional homicides until his arrest in 2005.

The BTK murders had seemingly ended in 1977 with the murder of Nancy Fox. The killer had communicated with authorities and the news media during these events calling himself "BTK," which he advised stood for "Bind them, Torture them, Kill them." BTK appeared to have gone dormant in 1979. The total BTK investigation continued however, and lasted for over 30 years—starting in January 1974 and finally culminating in his arrest and conviction in August 2005.

Dennis Radar had been linked to each of the murders through STR/PCR DNA technology. The Kansas Bureau of Investigation and the Wichita, Kansas police had their own "blooding" case as 4,000 men were requested to submit their DNA for comparison. Anyone who worked on the case, reported on the case or was even remotely involved with any of the victims was screened through this process.

The present and future

DNA from a crime scene can now be used to provide police with a composite profile of a suspect using Parabon Snapshot DNA Phenotyping. Snapshot can provide a kinship inference—determine the familial relationship between any two individuals, even distantly related.

DNA phenotyping uses SNPs to identify genomic ancestry. A person's precise ancestry can be determined on a global level. There are 7 principle populations in the world: Africa; Middle East; Europe; Central Asia; East Asia; Native American; and Oceana.

Computerized DNA interpretation

Cybergenetics' TrueAllele computer analysis of complex DNA overcomes the human limitations of "inconclusive" methods. TrueAllele objectively examines low-level, degraded, touch and mixed DNA samples to calculate match association. The software compares evidence with evidence, and separates relatives from mixtures. Crime scene and serial crime analysis can help reveal criminal activity.

Touch DNA

The touch DNA method was named for the fact that it analyzes skin cells left behind when assailants touch victims, weapons or anything else at a crime scene. Humans shed tens of thousands of skin cells each day. These cells are transferred to every surface our skin contacts, i.e. gun grips, eating utensils, steering wheels, etc. If a perpetrator deposits a sufficient number of skin cells on an item at the scene, there may be touch DNA. Touch DNA is not low copy number DNA, which allows a very small amount of DNA to be analyzed, from as little as 5 to 20 cells. The small amount of starting DNA in LCN samples requires many more cycles of amplification.

Conclusion

Millions of forensic DNA tests have been conducted in the United States and around the world. In a major advance, the analysis of DNA has evolved from a laborious process taking weeks or even months to a procedure that can be completed in a matter of days.

DNA technology is constantly evolving though new applications and innovations. Forensic scientists are combining advances in miniaturization and microchip technologies with well-established techniques of forensic DNA analysis. The fusion of these technologies could revolutionize DNA typing.

The utility and power of DNA as a tool to convict criminals or exonerate suspects has been greatly supported by careful legal reviews and stringent quality assurance guidelines that have been developed over the last 30 years.

Vernon J. Geberth is a retired Lieutenant-Commander with the New York City Police Department, and the author of "Practical Homicide Investigations."