CHAPTER FOURTEEN
The Blairsville Slaying and the
Dawn of DNA Computing

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Murder

On April 13, 2006, Blairsville dentist Dr. John Yelenic was murdered in his home, about an hour east of Pittsburgh, PA. Dr. Yelenic, who was living alone at the time, had exsanguinated onto his living room floor. On the coffee table, splattered with his blood, was the unsigned divorce document from his estranged wife, Michele. She was living with her boyfriend, Pennsylvania state trooper Kevin Foley.

John Yelenic's fingernails had DNA that tied trooper Foley to the crime, with a match statistic of 13,000. Prior to Mr. Foley's February 2008 preliminary hearing, his defense lawyer Richard Galloway said that the DNA did not rule out other suspects, because there was a one in 13,000 chance it came from someone else. Moreover, said his lawyer, DNA often identifies suspects to the exclusion of billions or trillions of others.

Computer

I was intrigued by Mr. Galloway's dismissal of the DNA match statistic. In my research as a scientist at Cybergenetics, a small Pittsburgh DNA technology firm, we had seen that computer interpretation of DNA mixtures usually preserves more identification information than does human review. Validation studies had compared our TrueAllele® computer

interpretation method with human expert review of the same DNA data, and typically showed a million-fold improvement in the match statistic. In the Foley case, that factor of a million could correct a 13,000 statistic into the billions, a level that the defense would find compelling.

Cybergenetics contacted the Pennsylvania prosecutor, Senior Deputy Attorney General Anthony Krastek, whose office is in Pittsburgh, and told him about our TrueAllele technology. When I spoke with SDAG Krastek, I explained to him how the computer could examine the DNA evidence far more carefully than could any person, and how that enhanced data scrutiny would objectively produce a more accurate match statistic. In our experience, when a person reported a DNA match in the thousands, the computer often found a statistic in the billions. SDAG Krastek arranged to send us the electronic data evidence that had been produced by the FBI laboratory in Quantico, Virginia.

I met with SDAG Krastek in Cybergenetics' Oakland office on Friday, April 11, 2008. He handed me a compact disc (CD) containing the FBI's DNA data. We discussed the case and the TrueAllele computer approach. The forensic problem was that there was a vast amount of the victim's DNA, combined with a small quantity of a second, unknown contributor.

Cybergenetics put the electronic DNA mixture data into its TrueAllele machine, asking the computer to solve the problem, and help identify the unknown contributor. The computer worked on our questions over the weekend. On Monday morning, I reviewed the results and phoned SDAG Krastek with the TrueAllele answer. The DNA under Dr. Yelenic's fingernails matched Kevin Foley with a statistic in the hundreds of billions. Further calculations would later refine this number to 189 billion.
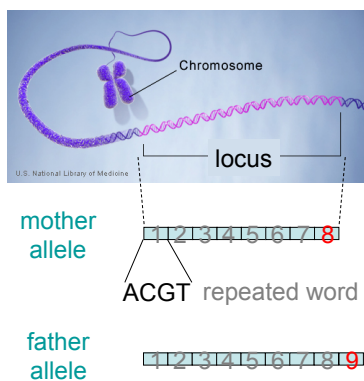
DNA

Deoxyribonucleic acid (DNA) is the genetic blueprint passed down from parent to child that encodes the human operating

system. Whereas computers represent instructions in a binary format of zeros and ones, DNA uses a four letter nucleotide alphabet of A, C, G and T. Three billion DNA letters instruct our cells on how to live, grow, reproduce and die.

A person's DNA is packaged into 23 chromosome pairs, with one copy inherited from each parent (Figure 1). The DNA sentence at a chromosome location (or, "locus"), is called an "allele". Except for the female (X) and male (Y) chromosomes, a person has two alleles (one from each parent) at every genetic locus. A person's allele pair at a locus is called a "genotype".
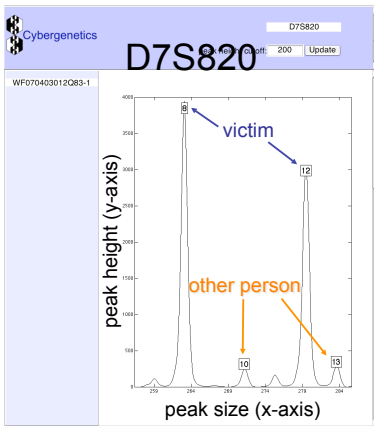
Figure 1

Human chromosomes come in pairs. The diagram shows a chromosome at different levels of magnification. The full chromosome has been unraveled to drill down to the individual DNA letters. A genetic locus used for forensic identification is made of Short (e.g., four letter) DNA words that are Tandemly Repeated (STR) to form a DNA sentence. The length of a person's DNA sentence at an STR locus is called an "allele". Two alleles (e.g., an "8" from one parent, and a "9" from the other parent) form this person's (8,9) allele pair "genotype".

Whereas functional genes are DNA sentences that code for proteins in the cellular machinery, other tracts of DNA have no known purpose. This "junk" DNA has highly variable regions with many different allele possibilities at a locus. Forensic scientists use junk DNA "loci" (plural form of locus) to identify people, since these loci have very many different allele pair possibilities—the chance that two people share the same two alleles is low.

Mixtures

A person's genotype is comprised of two alleles at a genetic locus. This allele pair shows up in the DNA data as one or two peaks (the two alleles could be the same). Peak size (x-axis) indicates the allele, while peak height (y-axis) is related to the quantity of allele present. A DNA mixture combines allele pairs from each contributor to the evidence. Nature adds up these allele pair DNA molecules in proportion to their contribution to produce a data pattern (Figure 2).

Figure 2

The crime laboratory transforms biological evidence into DNA data, the black curve shown in the figure. A DNA peak has a size (measured on the x-axis) that indicates the number of repeated words, and thus the allele value. The peak also has a height (measured on the y-axis) that reflects the amount of that allele present in the DNA evidence. This DNA mixture data is from the D7S820 (aka, "D7") locus in the Foley case fingernail evidence. The two tall peaks (8,12) correspond to the 93% major genotype contribution of victim John Yelenic. The two small peaks (10,13) correspond to a 7% minor genotype contribution from someone else. Kevin Foley's D7 genotype is (10,13), and so matches the minor evidence genotype. The *inclusion* interpretation of this data did not use Dr. Yelenic's (8,12) genotype, and so yielded relatively little match information; the *subtraction* and *addition* methods did use the victim's genotype, and were thus far more informative.

Computer interpretation of a DNA mixture is easy to understand. The computer tries out virtually every possible allele pair for the DNA contributors, adding them up in various proportions. Those genotypes (and their amounts) that better explain the data have a greater likelihood of being true.

Sophisticated methods like TrueAllele consider many other variables, and also determine the uncertainty of every variable. After many thousands of computer proposals and comparisons, a genotype is developed for every contributor at each tested genetic locus. This evidence genotype provides the probability of each allele pair. The computer-inferred genotype is completely objective, because no knowledge of any suspect is used in its determination.

In contrast, human experts tackle DNA mixture problems in a far simpler way. People cannot cope with the full complexity of all conceivable allele pairs, and the many possible combinations and sums. Nor can they explore all the feasible allele peak patterns. The analysts do not use the original peak height data, but instead declare that each allele is either present or absent. This imperfect decision is based on whether a data peak lies above or below some "threshold" value.

The examiner then asks whether a suspect's allele pair is included in the list of alleles that appear to be present in the data. This "inclusion" method discards considerable information, and can give wrong answers, but it is the most popular approach used by human experts when they examine DNA mixtures. With John Yelenic's fingernails, inclusion gave a match statistic of 13,000 to Kevin Foley.

TrueAllele's "addition" method uses peak height data, along with the fact that the victim's DNA can be present in his own fingernails. The threshold-based "inclusion" method does not use this available data.

There is a middle ground—subtracting out the victim's genotype, though still not using peak heights. For a third opinion, Prosecutor Krastek brought in Dr. Robin Cotton, former scientific director of the Cellmark forensic DNA laboratory. Her "subtraction" interpretation of the Yelenic fingernail mixture evidence produced a DNA match of 23 million to trooper Foley.

Law

The scientific basis of informative DNA match statistics is the likelihood ratio (LR). The LR is a number that tells us how much more probable a match between evidence and a person is than mere coincidence. The mathematics of the LR helps ensure that this match number removes preconceptions and prejudices unrelated to the evidence.

Forensic DNA scientists test many (e.g., 10 to 15) independent genetic loci for more identification power. They multiply together the match statistics from these independent genetic loci to form a product, often quite a large number. This multiplication is called the "product rule".

In Pennsylvania, the product rule became a precedent for DNA evidence in the Commonwealth vs. Blasioli rape case. In 1996, the Superior Court held in its Blasioli decision that the product rule was generally accepted within the scientific community, and that statistical evidence derived from that method was admissible. The Pennsylvania Supreme Court affirmed that determination in 1998. Thus, new human or computer methods for calculating product rule LRs for DNA match may arise, but the foundational product rule itself is not novel under Pennsylvania case law.

Challenge

Foley's defense team challenged the TrueAllele computer interpretation. They claimed that the approach (which employs the product rule) was novel science, and thus Judge Martin should first determine its reliability before the findings could be admitted as evidence. The prosecution disagreed, maintaining that the product rule was not novel in Pennsylvania. Regardless, a pretrial hearing was held to determine (a) whether an admissibility hearing was actually needed, and (b) if it was necessary, whether TrueAllele was sufficiently reliable to allow the 189 billion DNA match statistic to be heard by the jury. Since TrueAllele had never been used or challenged in court, I began

preparing a presentation that would explain to the judge why TrueAllele was scientifically reliable.

The February 18, 2009 pretrial hearing was held on a wintry Wednesday before presiding Judge William Martin. The Indiana County Courthouse is on Philadelphia Street, adjacent to the Jimmy Stewart Museum. Judge Martin was the evidentiary gatekeeper responsible for determining whether or not TrueAllele would be admitted as scientific evidence in his trial. Sitting without a jury, the judge would hear testimony and review exhibits, presented through direct and cross-examination, and then render his admissibility decision.

Reliability

Prosecutor Krastek began by asking me about the principles of DNA mixture interpretation. I explained that, fundamentally, all interpretation methods, whether done by man or machine, operated in the same way. First, a genotype is inferred from the DNA data, by comparing hypothesized models with the data in order to determine the probabilities of each genotype explanation. Then, this evidence genotype (as a probability distribution) is compared to a reference genotype (e.g., Kevin Foley), relative to a population, to calculate a DNA match statistic called a "likelihood ratio".

- Mixture interpretation methods are all the same in principle, but differ in how much use they make of the available data.
- The FBI's *inclusion* method does not use the victim's DNA genotype. Moreover, inclusion does not use the original quantitative data, it reduces data peaks to all-or-none "allele" events.
- Dr. Cotton's *subtraction* method does use the victim genotype. However, like inclusion, her method does not use the peak heights.
- The TrueAllele computer's *addition* method makes the most use of the evidence, and considers both the

> victim genotype, as well as the quantitative height of the data peaks.
> • Using more data can preserve more identification information.

Pennsylvania applies the "Frye standard" for determining the admissibility of scientific evidence. Based on a 1923 court decision, Frye v. United States, judges use this reliability standard to assess the general acceptance of a method in a relevant scientific community.

I showed that TrueAllele is based on established science, as published in scientific journals. SDAG Krastek provided the court with a CD containing almost 50 such articles. I reviewed with the judge a bibliography that listed these articles, describing TrueAllele's scientific foundations. The topics included DNA peak heights, genotype probability, DNA computer interpretation, statistical modeling and computing, likelihood ratios, similar computer systems, and my own publications.

A validation study is done to establish the scientific reliability of a method. I showed Judge Martin a TrueAllele validation study from a 2006 conference paper of mine. This study compared different mixture interpretation methods, and found TrueAllele to be more informative (as measured by higher DNA match statistics) than human review of the same data. We went over newer validation results (that were published later in 2009) done on 40 DNA mixtures of known composition. These results (from data unrelated to the case) could be used to predict the match statistic in the Foley case, based on the amount of DNA in the 7% minor contributor to the fingernail evidence. The results predicted that the computer's match statistic would be around a trillion, while human methods would be far less informative.

The reason why people lose information is the all-or-none "threshold" that human analysts apply to DNA data in order to simplify mixture interpretation. To explain the impact of thresholds, I showed a pure black and white (high contrast) photograph of a face; we could see that the person was a young man—and little else. I then showed the judge the

original image with all its shades of gray restored, revealing the face of a young Jimmy Stewart, as he looked in his classic film *It's a Wonderful Life*. It was visually apparent that using more of the data can retain far more information.

Indeed, that is how the forensic DNA scientific community contrasts simple inclusion with more powerful likelihood ratio methods. I showed the judge a highly influential 2006 paper written by the DNA Commission of the International Society for Forensic Genetics (ISFG). The article quotes prominent scientists as saying that the inclusion method:

- "Often robs the items of any probative value" (Dr. Bruce Weir)
- "Usually discards a lot of information compared to the correct likelihood ratio approach" (Dr. Charles Brenner)
- "Does not use as much of the information included in the data as the LR approach but, conceptually, they are equivalent" (Dr. Michael Krawczak)

The ISFG Commission's first recommendation was that "the likelihood ratio is the preferred approach to mixture interpretation".

To properly apply the Frye standard, it is important to clarify exactly who comprises the "relevant scientific community" in DNA mixture interpretation. I described the forensic scientists who largely focus on DNA inference and statistics. These researchers develop, discuss, publish, validate and assess DNA interpretation methods. In particular, they lay the scientific foundation for forensic practitioners, and may be more engaged in theory than practice.

I showed Judge Martin a 2001 scientific article about DNA mixture interpretation written by the Pennsylvania State Police laboratory in nearby Greensburg. Former laboratory director Christine Tomsey wrote that genotypes can be inferred from the data. Specifically, she approved of using a known contributor's genotype, and of considering peak height information.

A study conducted by the federal government showed that DNA mixture interpretation varies between forensic laboratories. Dr. John Butler of the National Institute of Standards and Technology (NIST) had distributed electronic data from the same DNA mixture to over 50 crime labs. The laboratory analysts' interpretations yielded DNA match statistics ranging from 31,000 (having four zeros after the one) to 213 trillion (14 zeros). This study established that the forensic science community uses different DNA mixture interpretation methods, some of which are more informative than others.

Other forensic groups use TrueAllele methods and systems. I showed Judge Martin a 2008 scientific paper by New Zealander Dr. James Curran about his computer method for resolving two person DNA mixtures. Compared with our TrueAllele approach, his equations share the same key variables, and his program finds genotype solutions in the same probabilistic way. Indeed, I pointed the judge to five different computer systems from around the world that all solve DNA mixtures.

I listed fifteen groups in government, academics and industry that had used the TrueAllele technology. For example, Cybergenetics had re-examined the victim remains DNA data from the World Trade Center disaster, in order to help identify missing people.

I showed the judge pictures of how match statistics changed at each of the 13 tested genetic loci, as the DNA mixture interpretation methods become increasingly more informative. I handed him four bar charts of inferred DNA match locus information for the Dr. Yelenic fingernail evidence.

- The *inclusion* method gave a small amount of information at each locus. Multiplying the 13 locus numbers together gave a product of 13,000.
- Dr. Robin Cotton's *subtraction* method made excellent use of Dr. Yelenic's known victim genotype at two of the loci. Considering those two larger numbers increased the match product up to 23 million.
- The TrueAllele *addition* method again showed more match information at those two loci (considering the

victim genotype), but also at several other loci (due to peak height information). This better use of the data further increased the DNA match statistic to 189 billion.
- None of these methods reached the full match strength of Kevin Foley's own genotype, which would have given a statistic of 875 trillion.

It was visually clear from this succession of pictures that nothing magical was going on. As each interpretation method made better use of the available DNA data, the match statistics increased.

Defense attorney Richard Galloway then conducted his cross-examination.

- He questioned how reliable DNA data could give different statistics. The answer, as I replied, resides in the different interpretation methods applied to the data.
- He asked why the computer did not use thresholds in the same way that people did. I said that since the computer accounts for data uncertainty in a more precise way using mathematics, the machine has no need for a coarser human approximation.
- Mr. Galloway also asked whether TrueAllele had ever been used before in court. I told him that no, it hadn't. The Foley case was the first appearance of TrueAllele (or, indeed, any such computer mixture interpretation) in a criminal case. But the question before the court was TrueAllele's reliability as the scientific method, not its use elsewhere.

It was now for Judge Martin to decide. If he admitted the evidence, this would be the first time in any trial that a sophisticated computer had ever been used to interpret DNA mixture evidence. An adverse ruling, on the other hand, could slow down for many years TrueAllele's adoption by the criminal justice system, as well as diminish the prosecution's case.

Admissibility

On March 2nd, President Judge Martin issued his opinion on the TrueAllele methodology. He wrote that, "it is recognized that there is more information available which more conservative approaches do not consider. Therefore, it seems logical that the scientific community would work towards including that unused data to arrive at a more accurate finding." Citing materials presented at the hearing, Judge Martin ruled that "based on a review of the evidence, the court finds that Dr. Perlin's methodology is admissible pursuant to the Frye rule and Rule 702." The TrueAllele DNA match results would be heard at the Foley trial.

Trial

The DNA evidence against Kevin Foley was presented on March 12th. Jerrilyn Conway of the FBI testified first, followed by Dr. Robin Cotton. My DNA testimony came last. I had never testified in court before.

Answering prosecutor Krastek's questions, I presented to the jury key points from the Frye hearing.

- There is really only one DNA interpretation principle – infer a genotype objectively from the data, and then match it with a reference genotype.
- Different interpretation methods simply make different use of the same data.
- Better use of the data can yield more identification information.

As Dr. Butler's 2005 NIST study had shown, huge variations in DNA mixture match statistics on the same data are expected. The numbers depend on the method used to interpret the evidence. Our National Institute of Justice (NIJ) study on 40 NIST mixture samples could be used as a calibration to predict a match statistic number, based on interpretation method and contributor DNA amount. The 189 billion TrueAllele

statistic for a match between Dr. Yelenic's fingernails and Mr. Foley was predictable, and not in any way unexpected.

Mr. Galloway's cross-examination revisited much of the same Frye hearing terrain. I explained to the jury that the different reported match statistics resulted from how different methods used the data. The defense attorney protested that, with precise methods, the same data should give the same answer.

I replied that when a scientist examines a microscope slide with the naked eye (like the weak "inclusion" method), they can only see so far. Using a magnifying glass (i.e., "subtraction" method) on the same slide, they will see more. And, with a microscope (the computer's "addition" approach) they would see even more. "The information is there," I said. "The question is what is the resolution of the instrument that you are using to make the observation."

"Are you uncomfortable with what the FBI does?" asked Mr. Galloway. "No," I replied. "But if you are a doctor trying to diagnose bacterial disease, sometimes you need a microscope. ...I would be more comfortable using a higher precision instrument to make a diagnosis that might be more informative —same slide, same data—just a more precise approach."

Verdict

On the morning of March 18th, state trooper Kevin Foley testified in his own defense. That afternoon, the prosecution and the defense made their closing arguments. "John Yelenic provided the most eloquent and poignant evidence in this case," said SDAG Krastek. "He managed to reach out and scratch his assailant," capturing the murderer's DNA under his fingernails. The jury deliberated, and that night convicted Mr. Foley of first-degree murder.

Appeal

Kevin Foley appealed his conviction to the Pennsylvania Superior Court. He claimed that Judge Martin erred in admitting my testimony, suggesting that TrueAllele should have failed the Frye test for "novel" scientific evidence. On March 29, 2011, appearing before an appeals court in Pittsburgh, SDAG William Stoycos referenced three bound volumes of scientific publications, hardcopy printouts of the CD articles introduced at the trial. Foley's lawyers argued that DNA mixture interpretation was unreliable, but Mr. Stoycos explained why TrueAllele was a reliable interpretation system.

Two TrueAllele validation studies had already successfully withstood the scrutiny of scientific peer-review. One study used laboratory-generated DNA samples, and found that quantitative analysis performed by TrueAllele was much more sensitive than qualitative analysis such as that performed by the FBI (Perlin & Sinelnikov, "An information gap in DNA evidence interpretation", PLoS ONE, 2009). Another paper used DNA samples from actual cases and reached similar conclusions (Perlin et al., "Validating TrueAllele DNA mixture interpretation", *Journal of Forensic Sciences*, 2011). This casework study "validated the TrueAllele genetic calculator for DNA mixture interpretation" and found that when "a victim reference was available, the computer was four and a half orders of magnitude more efficacious than human review."

In its December 28, 2011 decision, the Superior Court affirmed Judge Martin's ruling. The court noted that scientific studies of TrueAllele's reliability had been "published in peer-reviewed journals; thus, their contents were reviewed by other scholars in the field." TrueAllele may have been a new system, but the appellate court held that it was not "novel" scientific evidence. On February 15, 2012, the Superior Court published its Foley decision, establishing a statewide TrueAllele precedent throughout the Commonwealth of Pennsylvania.

On January 4, 2013, the Pennsylvania Supreme Court issued a *per curiam* Order denying Foley's petition for

allowance of appeal. As a result, the Superior Court's affirmance of Foley's judgment of sentence stands, including its determination that expert testimony regarding DNA match probability calculations in mixed sample DNA cases which relies upon the TrueAllele methodology is admissible into evidence. The intermediate appellate court's determination is binding precedent throughout the Commonwealth of Pennsylvania.

Impact

Over a hundred TrueAllele reports have been issued in criminal cases, helping both prosecution and defense. I have testified on TrueAllele DNA match statistics in state, federal, military and international trials. The Superior Court Foley precedent has accelerated the introduction of objective TrueAllele technology into Pennsylvania courts for DNA mixture evidence. I have given two continuing legal education (CLE) courses in Pittsburgh about TrueAllele and the Foley case, one at Duquesne University, and the other at the Allegheny County Courthouse for trial attorneys.

The Foley case can be instructive for defense attorneys who encounter DNA evidence that may implicate their client. The initial match statistic to Foley of 13,000 was not over-whelming. But the later computer resolution of the DNA mixture into its component genotypes changed the situation, introducing a more accurate (and much more persuasive) 189 billion statistic. By repeating to the jury over and over again the "millions", "billions" and "trillions" statistics of the different experts, the defense reinforced the large numbers. Once reliable science had established the presence of Kevin Foley's DNA under Dr. Yelenic's fingernails, Foley's adamant denial of contact with the victim became less credible. Perhaps giving some explanation of how an innocent intent had accidently escalated into a fatal consequence might have mitigated the verdict or sentence.

Commonwealth v. Foley was a landmark case in the history of DNA evidence. It was the first time that an advanced statistical computing method for interpreting DNA mixtures was ever:

1. used as evidence for a criminal case;
2. admitted into evidence after an admissibility challenge;
3. introduced as evidence in a trial;
4. upheld as reliable evidence by an appellate court; and
5. established as a statewide precedent.

Dr. John Yelenic was brutally and tragically murdered, but the trial that convicted his killer bequeathed to society a powerful truth-seeking technology for bringing criminals to justice.