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ISHI INTERNATIONAL SYMPOSIUM ON HUMAN IDENTIFICATION

PALM SPRINGS, CA | SEPT. 23–26, 2019

APRIL 2019

The ISHI Report

News from the World of DNA Forensics

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The ISHI Report

Letter from the Editor

Welcome to the first issue of The ISHI Report. This publication was envisioned as a vehicle to bring stories to the forensic community on topics of interest to those engaged in the important work of DNA typing for human identification. Consider the magazine a supplement to the experience of attending the actual International Symposium on Human Identification where you can hear about the latest trends in forensic science, meet others who share your passion for forensics and advance your scientific knowledge.

The inaugural issue will introduce you to several people who have impacted the field including Bill Linton, the President and CEO of Promega who was responsible for organizing the very first ISHI in 1989. You'll also meet Robin Cotton, whose testimony in courtroom hearings, including the OJ Simpson trial helped establish the reliability of DNA evidence. Finally, you'll learn more about our keynote speaker, Paul Holes who will discuss his role in helping to identity the Golden State Killer, a serial predator who terrorized California residents for decades before his arrest last year.

Genetic genealogy is shaping up to be a powerful tool to solve cold cases when all other leads have been exhausted. This issue will include an overview of the process and examples of its successful application. Use of Y-STRs is now an important part of criminal cases. Lutz Roewer, who helped to establish and maintain the YHRD database provides an update on the database with advice on how to use it.

Nearly everyone can improve their leadership and coaching skills. As scientists, softer skills like managing people can be overlooked. Former laboratory director turned management coach, John Collins will describe easy steps that anyone can take to be a more effective leader. If you like what you read, consider attending John's workshop at ISHI 30 scheduled for Sunday, September 23.

We look forward to hearing your feedback on the first issue of The ISHI Report. Look for quarterly updates with the next issue scheduled for July. Of course, let us know if you have a topic to suggest or better yet, volunteer to contribute an article.

Warm Wishes,

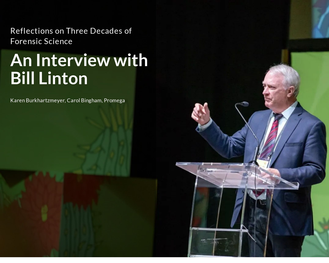
The Editorial Board

Contact ISHI

Reflections on Three Decades of Forensic Science

An Interview with Bill Linton

Karen Burkhartzmeyer, Carol Bingham, Promega



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DNA typing was in its infancy. Random Fragment Length Polymorphism represented the state of the art in forensic science and the world-wide web was years in the future when the first International Symposium on Human Identification was held in 1989.

Few at that meeting could have predicted that the arguably unglamorous job of the forensic scientist would achieve such a degree of mainstream awareness and appeal. Fueled in part by the widespread popularity of television shows like CSI, Forensic Files, and NCIS among others, forensic science has been embraced in mainstream culture and is now recognized as the most powerful tool that law enforcement has at its disposal.

But decades before the field fascinated Hollywood, many of the real-life heroes of forensics gathered over the longest-running and largest conference in the world focusing on DNA typing for human identification. For the past 29 years, the International Symposium on Human Identification (ISHI) has brought together forensic professionals from around the world to discuss and debate issues that are important and sometimes controversial in the field, to share new ideas and to collaborate to advance forensic science.

The first ISHI was convened in Madison, Wisconsin, in 1989. Why Madison? The city is where the biotech company Promega Corporation got its start 40 years ago. Among the most promising products the company began manufacturing in the late 1980s were VNTR and RFLP probes that could be used in the emerging field of DNA forensics.

Promega founder and CEO, Bill Linton sat down with us to discuss why he organized the first symposium, how the industry has evolved and his thoughts on the future of forensics.

How did the first ISHI come about? What made you decide to organize an international symposium on human identification?

The first ISHI was held at a time when there was very little commercial activity. Very little of the market had evolved because DNA typing hadn't really been established as a validated forensic method. As a result, we knew that having identified the VNTR (variable number tandem repeat) technology would lead to an evolutionary process and an educational process to get this technology into the market. The concept of a forum was really to bring together some of the early thinkers, the people who had recognized that this could be an important aspect of human identity, and bring them together and just exchange ideas, share ideas (have some speakers), but more importantly have people come together.

And being aspirational, in terms of, "Where could this go?" or "Who ultimately do we think the audience is going to be?" we said, "Well let's make it an international symposium". I'll have to say I doubt that first symposium had much in the way of international participation. If there were one or two people from another country, I'd say that would have been generous. But the name of it was something obviously that stuck. It was also something that we thought, "Ultimately this is going to be global and it is something that's going to be important to forensic and police departments and people involved with human identity worldwide", and so that's why we named it what we did.



How did you get the word out about ISHI #1 and who did you invite?

There was a very limited audience at the time and so we pretty much knew the major scientists who were involved in developing the methodologies and starting the early ideas of validation, and so the communications went out to those people that we knew. It was probably a combination of letters, phone calls and (to the extent that there was email then), probably some email.

What was the response?

We probably had between 40 and 50 people at that first meeting. I think the fact that we had 40 or 50 people coming here to Madison and being involved so early in that event, we were very pleased with it, particularly the caliber of the people we had coming here. Most of the people involved were in academic research. There was some FBI involvement as well.



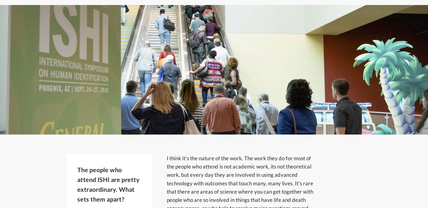
What were your memories of that first meeting?

I do recall, as we talked to people afterwards, that they wanted to continue it. They thought an annual meeting was good because of how rapidly the technology itself was emerging. Very early on, we had the recommendation to move it outside of Madison principally because of the weather. [The second one was in Madison as well], but the third symposium was moved to Arizona only because, at that time of year, the weather was unpredictable.

In those early years, did you expect the eventual growth of ISHI and its reach within the industry?

I think we all felt that this field had enormous potential and that we were an early mover in the market and in the adoption of the science and the technology going into manufactured kits, so there was that understanding. As the meetings occurred year after year, the feedback was always extraordinarily positive; the way it was managed by Promega, the way that we put together the programs, the people that were invited to speak. I always like to pay attention to the comments that people make during a meeting, and after a meeting, to reflect back on "Is this worthwhile to continue to do?" and as we heard from everybody, they said, "Yes, absolutely continue doing this."

I'm not surprised that it's gone on for 30 years, I'm not surprised that we have the people that we do today. I am surprised by the number of people who have been at virtually all the meetings year after year after year. I think that's extraordinary. I also expected at the beginning that there would probably be more meetings that would compete with this meeting; that there would be other companies or other organizations that would try to do something of a similar nature. But I also think that because of how well ISHI was done, how well it was managed, that people look to that and said "That's the meeting to go to." I think we kind of took that high-ground early on to establish something that really was seen as the most important meeting for many of these forensic people to go to year after year. I'm just happy that it's worked out as well as it has and that we still have that level of quality.



The people who attend ISHI are pretty extraordinary. What sets them apart?

I think it's the nature of the work. The work they do for most of the people who attend is not academic work, its not theoretical work, but every day they are involved in using advanced technology with outcomes that touch many, many lives. It's rare that there are areas of science where you can get together with people who are involved in things that have life and death consequences, or who help to resolve major questions around accidents or crime scenes. All of these things really touch so many people, and so these are people that are just involved in that day after day after day.

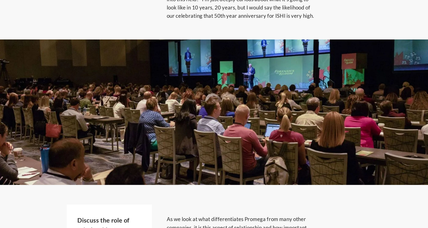
They like to get together with other people to share stories and to meet with other people they saw last year and the year before, and it's really become like this immense club, but not an exclusive club. We see new people coming in every year. Something similar would be a meeting where you have healthcare providers that are providing solutions to medical problems at very advanced levels and where you have people who are really engaged from a practice standpoint. So that's what it is, it's a meeting of practitioners, and of course people who are continuing to advance the science and the technologies, to support the work the practitioners are involved with.

Your thoughts on the development and innovation of human identity technology through the years...

Forensic science is both a mature field and a field that continues to evolve, so in the very early days it's like an infant and you're feeding an infant with a lot of caregiving. And then I'm sure we could point to the adolescence phase where it was really emerging. I'd say the development of STRs and that whole other generation of technology corresponded to the adolescence stage. Today, I really see it as being more in the adult phase but still an adult with growth potential.

What do you envision for the future of ISHI?

Obviously ISHI is 30 years old this year. I hope I'm around to celebrate the 50th year anniversary. I think it would be a great accomplishment! I think it's highly likely that Promega will still be very much involved in the field. And what's curious to me is to ask, "What will be the new applications? What will be the new technologies that will begin to come in and really make a difference in this field? And what is our part in helping bring that into this field?" I'm just deeply curious about what it's going to look like in 10 years, 20 years, but I would say the likelihood of our celebrating that 50th year anniversary for ISHI is very high.



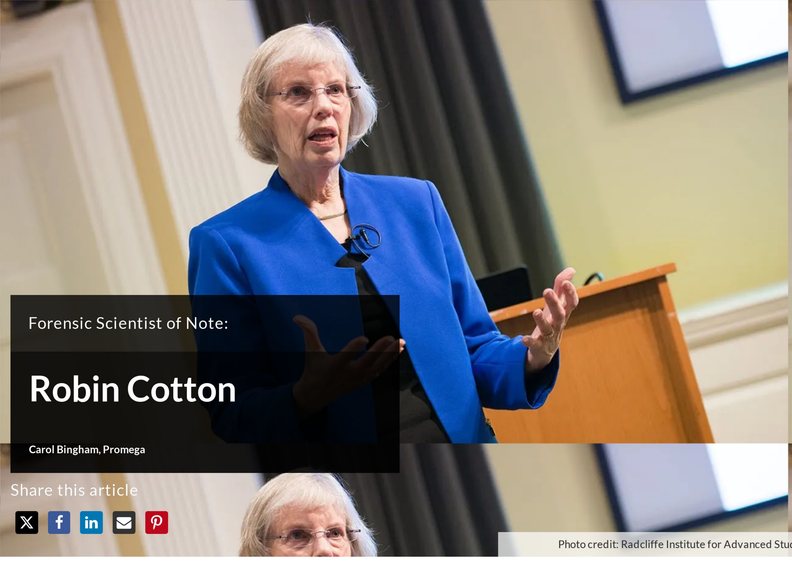
Discuss the role of relationship at Promega and how that translates to ISHI.

As we look at what differentiates Promega from many other companies, it is this aspect of relationship and how important that is; both within the company, relationship with self, relationship within the company itself and our colleagues, and then relationship with our customers and the community. In community, we often think about the geographical community. With the forensics field, I think there's a sense of community with the people who work in that area of science, the practitioners, and I think their reflection is that Promega has been there for them since the beginning, and for many of them, we've been there when they started their careers; and so, there's this sense of support, of commitment, of a long-term view that gives them the confidence that we really do have a partnership with them.

I think the nature of ISHI itself is a reinforcement of this sense of relationship. For example, the event we have every Wednesday night at ISHI, I think they call it the "Promega Party," is a reinforcement of how we look at their work, and how we look at them as going beyond simply a transaction. It goes beyond business and goes into this idea of relationship and the importance of what they do and who they are. It just continues to reinforce relationships and how that then cements and reinforces our connection with the people that use the products and the technologies.

Much has changed since the first meeting was organized in 1989, but some aspects have remained constant. These include ISHI's focus on offering the forensic community a forum to gather each year to learn from each other and share the latest advances in the field.

As always, Bill Linton will be on hand at ISHI 30 to deliver opening remarks at the general session. Unlike the first symposium, this meeting promises plenty of warmth, sunshine and updates on technologies undreamt of thirty years ago.



Forensic Scientist of Note:

Robin Cotton

Carol Bingham, Promega

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Photo credit: Radcliffe Institute for Advanced Study

It was Alec Jeffrey's paper on RFLP analysis that proved the catalyst for Robin Cotton to alter her career path from research on the Y chromosome to the applied science of DNA typing.

A biochemist by training, Cotton had a PhD in molecular biology and biochemistry from UC Irvine and two post-docs under her belt. She was working in a lab at NIH in 1988 when a chemist friend showed her a newspaper clipping from *The Washington Post*. It featured an interview with people who were starting a lab in Germantown, Maryland to do DNA identification. She contacted the lab, Cellmark Diagnostics, to see if they were hiring, and since accepting her first position there, Robin has been involved in some of the most consequential events in the field of forensics.

Prior to the late 1980s, DNA typing did not exist. The most informative information to be obtained from blood stains were blood groups, which were of limited use. RFLP analysis coupled with Alec Jeffrey's discovery of the massive amount of length polymorphism in some types of repeated DNA sequences was a game changer. Early DNA labs, like Cellmark, used single locus RFLP probes on Southern blots. Compared to today's tests, the technology was quite simple, but even so, results were orders of magnitude more informative than blood group typing.

In the early days of DNA fingerprinting very few police departments had their own in-house laboratories with DNA capability to process samples. Most agencies outsourced their samples to service labs including Cellmark which was one of the first laboratories in the United States to provide services for criminal investigations. As luck would have it, Dr. Cotton had been working on samples submitted by the Los Angeles Police Department for a number of years and was requested to analyze the first 24 samples submitted by the LAPD for the O.J. Simpson case. Testifying on the DNA results for this case proved to be one of the most interesting and intense experiences of her career.

One thing that surprised Robin about providing testimony for the OJ case was the recognition that she received from the televised testimony. She recalled being recognized as the "DNA lady" in an elevator on the first day of her testimony. The recognition from being on television lasted almost a year. Despite the high stakes of her testimony she was able to maintain a cool head and focus on presenting forensic science as the strong tool that it is.

The Simpson case was one Robin's most high-profile cases, but it represented only one of the hundreds of times she was called on to testify as an expert witness. As she explains, "in the early days, there were admissibility hearings, not in the occasional case, but in every case, in every state. Every place you went there was a Frye hearing. Some of them very contested; some of them would last for days".



In total, Robin testified in 35 states, a total of more than 250 times. In describing the legal landscape, she said, "if you looked at the legal precedents in each state for allowing DNA to come in as evidence, you would see that there were a handful of maybe 20 to 40 people across the country who did all these hearings over a period of about five years. After that time, most states had some sort of precedent". In other words, only a handful of people performed all the casework and gave all the courtroom testimony which resulted in the acceptance of DNA results in U.S. courtrooms.

It was against this backdrop that the first International Symposium on Human Identification, was organized by Promega. Robin attended one of the first meetings in Madison, Wisconsin and recalled "Attendees comprised a tiny group of people in a small room. Drinks and some little snacks were provided, and the weather was freezing. At the time there were few opportunities for forensic scientists to get together with others from around the country. Of course, there was no email at the time or cell phones – and so communications from lab to lab would occur within a state but having communications with DNA people from elsewhere was a big deal." Through the years, Robin has continued to participate in the International Symposium on Human Identification, presenting numerous talks during the general session and helping to organize workshops on subjects from Probabilistic Genotyping to Tips for Courtroom Testimony.

In recent years Dr. Cotton has embarked on a new chapter in her forensic science career. She is now Director for the newly established Biomedical Forensic Sciences Program at Boston University School of Medicine where she is focused on grooming a new generation of scientists. One highlight of her job is the opportunity to collaborate with students. Every participant in her program is required to complete a program of independent research culminating in a written thesis of publishable quality per FEPAC requirement. Students brainstorm with Robin and other faculty to identify a problem in the forensic work flow that can be improved and often collaborate with vendors to develop data that will be used in real world applications.

Not long ago, Sir Jeffreys, (having received a knighthood for his scientific work) retired from Leicester University. Robin was fortunate to receive an invitation to his retirement party. She took the opportunity to attend the event along with her husband. As she explains, "I just wanted to thank him, without him, I wouldn't have had a chance to have this career. He was the one that published the information, he was the one who did the research and came up with the idea of using DNA for identification. His work has impacted so many people, it's a shame he was never awarded a Nobel Prize". Robin relates that Dr. Jeffreys is both humble and without pretense, as she explains, "he's the real deal, brilliant and nice, that's Alec Jeffreys". The forensic community could certainly say the same about Robin Cotton. Her work has impacted the field since the early days of the science and continues today with the mentorship of the next generation of scientists.

Editor's Note: Participate in this year's ISHI in Palm Springs and you'll have the opportunity to see memorabilia from the OJ Simpson trial provided by Robin and others who were involved. Artifacts on display will include copies of the expert testimony, contemporary photos and RFLP autorads.

Forensic Genealogy:

What Your Second Cousin's DNA May Say about You

Ken Doyle, Promega

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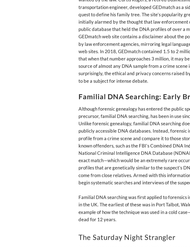
"The DNA people said, 'That was him. You got him.' And the whole room just exploded. Some detectives were crying. Some detectives were sitting there with their mouths open, staring into space."

Thanks to Colleen Fitzpatrick, a cold case that had baffled investigators for over two decades finally reached a breakthrough in 2014. She described the reaction in the Phoenix, AZ police department as a mixture of disbelief and elation. Fitzpatrick had just received the results of a DNA analysis obtained using a technique known as genetic genealogy that narrowed the DNA profile down to just one individual out of over 2,000 suspects. Fitzpatrick, who has a PhD in nuclear physics, is a founder of IdentifiGenes International, a company that provides genetic genealogy consulting services to solve forensic cases around the world.

Although genetic genealogy itself isn't new, its application to forensics is a relatively recent development. The technique relies on Y-DNA testing, which follows the male family line, making it popular among genealogists around the world. Forensic applications use the DNA profile derived from evidence at a crime scene to search multiple, public DNA profiles available in genealogy databases, with the goal of identifying possible family members related to a suspect. CeCe Moore, founder of The DNA Detectives Facebook group, explains how genetic genealogy works in this video:



Forensic genealogy relies heavily on access to public genealogy databases. Consumer DNA testing services such as Ancestry.com and 23andMe are well-known among genealogy hobbyists, and they maintain their own collections of DNA sequence data. However, the site that gained fame for forensic genealogy doesn't offer any DNA testing services at all. GEDmatch, unlike commercial DNA testing services, is a free, open-source database that relies on users to voluntarily upload their DNA profile information obtained from commercial DNA testing.

At the time the site was launched, however, the founders of GEDmatch had no idea that their creation would be used to track down serial killers, rapists and others wanted by the law. Curtis Rogers, a retired businessman, and John Olson, a transportation engineer, developed GEDmatch as a side project during Olson's spare time. Rogers' own quest to define his family tree. The site's popularity grew rapidly, but Rogers was initially alarmed by the thought that law enforcement officials were searching a public database that held the DNA profiles of over a million Americans. Today, the GEDmatch web site contains a disclaimer about the potential use of the information by law enforcement agencies, mirroring legal language on consumer DNA testing web sites. In 2018, GEDmatch contained 1.5 to 3 million DNA profiles, so it's estimated that when that number approaches 3 million, it may be possible to determine the source of almost any DNA sample from a crime scene in the US (source). Not surprisingly, the ethical and privacy concerns raised by forensic genealogy continue to be a subject for intense debate.

Familial DNA Searching: Early Breakthroughs

Although forensic genealogy has entered the public spotlight in recent years, its precursor, familial DNA searching, has been in use since the end of this century. Unlike forensic genealogy, familial DNA searching does not rely on open-source or public genealogical databases. Instead, forensic investigators obtain a DNA profile from a crime scene and compare it to those stored in DNA databases of known offenders, such as the FBI's Combined DNA Index System (CODIS) or the UK National Criminal Intelligence DNA Database (NDNAD). The goal is not to find an exact match—which would be an extremely rare occurrence—but to identify profiles that are genetically similar to the suspect's DNA, indicating that they could come from close relatives. Armed with this information, investigators could then begin systematic searches and interviews of the suspect's putative family members.

Familial DNA searching was first applied to forensics in 2003, to solve several cases in the UK. The earliest of these was in Port Talbot, Wales, and it was a dramatic example of how the technique was used in a cold case—after the suspect had been dead for 12 years.

The Saturday Night Strangler

In 1973, two teenage girls left a nightclub in Swansea and decided to hitch a ride. They never made it home; their bodies were found in the woods near Port Talbot, and they had been raped and strangled. The team of detectives working on the case —the first serial killer identified in Wales and dubbed the "Saturday Night Strangler" by the public—soon became overwhelmed by its magnitude. More than 10,000 drivers who could have possibly given the girls a ride had to be interviewed. Further complicating the investigation, several events near the town had led to a large cline of strangers, who could also be potential suspects. By the middle of 1974, the case—then known as the Llandarcy murders—was closed in the absence of any strong leads and the evidence was shelved.

In 1990, a forensic biologist named Colin Clark with Forensic Science Services (FSS), a government-owned company, reviewed the case. He found that much of the evidence had been damaged by mice and the damp conditions in Port Talbot. Clark was interested in attempting DNA analysis, but the techniques available at the time were inadequate for the challenging task.

Ten years later, another FSS scientist, Jonathan Whitaker, was working with a recently developed, innovative DNA profiling technique called low-copy-number (LCN) analysis. LCN uses the polymerase chain reaction (PCR) to amplify DNA from very small or damaged samples, making it possible to obtain a DNA profile from just a few cells. However, due to the method's extreme sensitivity, it is also susceptible to errors from contamination and PCR artefacts, and it has gained its share of controversy over the years.

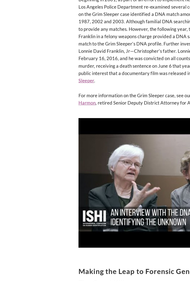
Whitaker realized that LCN might hold the key to finally unlocking the mystery hidden in what was left of the evidence from Port Talbot. Using LCN, his team was able to obtain a DNA profile of the perpetrator. However, after searching the UK national DNA database of known offenders, the team was unable to find a match. Despite that initial failure, the case was officially reopened.

A team headed by detective chief inspector Paul Bethell inherited the case archives. They had 25,000 persons of interest and knew their best hope of solving the case rested in obtaining DNA profiles, but it was clearly impractical to do so for the entire list. It took eight months to narrow down the list to around five hundred. Following further investigation, they obtained DNA samples from 353 men. None of those profiles matched the one obtained from the evidence.

Meanwhile, another cold case provided evidence that was to become vital in solving the Llandarcy murder mystery. In 1973, another teenage girl who had hitchhiked was brutally raped and strangled. Her body was found in Tonmawr, less than 10 miles from Port Talbot, three months before the murders of the girls in the Llandarcy case. The DNA sample showed that the same killer was responsible for all three murders.

Whitaker—working with Dark on the Llandarcy murders—had a stroke of inspiration. Although the DNA profile of the perpetrator hadn't matched any of the profiles in the national database, he suggested that a close family member of the killer might be represented in the database, thereby providing a partial match—50%, in the case of a son or father. One name rose to the top—Paul Kappen, a well-known car thief. The name was familiar to Whitaker. One of the five hundred people on the original Llandarcy list was named Joseph Kappen, but the investigators hadn't obtained his DNA, because he'd died 11 years earlier. Under Whitaker's direction, the team managed to obtain DNA samples from Joseph Kappen's ex-wife and daughter. Together with Paul Kappen's sample, these results gave them a match to 75% of the DNA profile from the Llandarcy murder case.

One final piece of evidence was required: a DNA sample from Joseph Kappen himself. The forensic team had sufficient evidence to exhume his body, despite his family's objections. On a night that seemed straight out of a horror movie—complete with pouring rain, lightning and thunder—Kappen's coffin was dug up and his remains sent to a local mortuary, where samples were taken from a femur and teeth. The DNA profile was a perfect match, providing the first triumph for Whitaker's technique of "familial DNA searching."



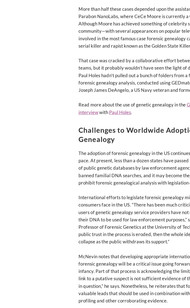
The Grim Sleeper

Across the Atlantic, initial adoption of familial DNA searching was slower than in the UK. Although the Denver, Colorado Police Department began studying the technique in 1999, California became the first state to make familial DNA searching legal in 2008. Cases in which the technique was employed had to meet two criteria: 1) the crime scene DNA profile had to be a single-source profile; and 2) all other investigative leads must have been exhausted.

The first—and arguably the most infamous—test of familial DNA searching in California was in pursuit of a serial killer known as the Grim Sleeper. The nickname derives from the 13-year break that the perpetrator apparently took after a killing spree that started in 1985.

Beginning in 2001, as part of an effort to implement new DNA analysis methods, the Los Angeles Police Department re-examined several cold cases. Continuing efforts on the Grim Sleeper case identified a DNA match among the victims killed in 1987, 2002 and 2003. Although familial DNA searching was used in 2008, it failed to provide any matches. However, the following year, the arrest of Christopher Franklin in a felony weapons charge provided a DNA sample that resulted a partial match to the Grim Sleeper's DNA profile. Further investigation led to the arrest of Lonnie David Franklin, Jr.—Christopher's father. Lonnie Franklin's trial began on February 16, 2016, and he was convicted on all counts of murder and attempted murder, receiving a death sentence on June 4 that year. The case captured so much public interest that a documentary film was released in 2014, Tales of the Grim Sleeper.

For more information on the Grim Sleeper case, see our interview with Rock Harmon, retired Senior Deputy District Attorney for Alameda County, California.



Making the Leap to Forensic Genealogy

The Canal Killer

Forensic genealogy, which enables investigators to bypass the constraints of only searching DNA databases of known offenders, also has its share of high-profile cases. Foremost among these was Fitzpatrick's Canal Killer case. The murders of two young women occurred in 1992 and 1993; in each case, the woman had gone for a bike ride along the Arizona canal in Phoenix and never returned. The first woman's decapitated body was discovered in a parking lot and, later, her head was found in the canal. The second victim's body was also found floating in the canal.

"I happened to be at ISHI in 2014," Fitzpatrick says, "and I had the opportunity to speak to the cold case investigators there…telling them about forensic genealogy." The Phoenix police department told Fitzpatrick they had a cold case that she might be able to help them with. They later sent her their Y-STR DNA testing results. Fitzpatrick searched several public genealogy databases to compare the profile from the case.

"I applied the software to find a match in those hundreds of thousands of profiles, and it came up with three exact matches to the name 'Miller'," Fitzpatrick recalls. "They had a list of two thousand suspects, and having the name narrowed it down to five." Further investigation narrowed the search down to one individual, based on information from a forensic professional society and the FBI. The Phoenix police obtained a DNA sample from the suspect, the profile matched the one Fitzpatrick analyzed, and the rest is history.

The Golden State Killer

In the years since Fitzpatrick solved the Canal Killer case, forensic genealogy has advanced to the point where even several of the most notorious cold cases have become solvable. In 2018 alone, over a dozen cold and active cases were solved using forensic genealogy in the US (Erlich et al., Science 362, 690–694 (2018), see Table 1).

More than half these cases depended upon the assistance of a single company: Parabon NanoLabs, where CeCe Moore is currently a Chief Genetic Genealogist. Although Moore has achieved something of celebrity status in the forensics community—with several appearances on popular television shows—Parabon wasn't involved in the most famous case forensic genealogists can yet the capture of the serial killer and rapist known as the Golden State Killer.

That case was cracked by a collaborative effort between forensic and genealogy teams, but it probably wouldn't have seen the light of day if an investigator named Paul Holes hadn't pulled out a bunch of folders from a forgotten file cabinet. The forensic genealogy analysis, conducted using GEDmatch, led to the arrest in 2018 of Joseph James DeAngelo, a US Navy veteran and former police officer.

Read more about the use of genetic genealogy in the Golden State Killer case in our interview with Paul Holes.

Challenges to Worldwide Adoption of Forensic Genealogy

The adoption of forensic genealogy in the US continues to proceed at a cautious pace. At present, less than a dozen states have passed legislation permitting the use of public genetic databases by law enforcement agencies. Maryland has already banned familial DNA searches, and it may become the first state to expressly prohibit forensic genealogical analysis with legislation currently pending.

International efforts to legislate forensic genealogy mirror the concerns that consumers face in the US. "There has been much criticism of the fact that existing users of genetic genealogy service providers have not explicitly given permission for their DNA to be used for law enforcement purposes," says Dimitri McNevin, PhD, Professor of Forensic Genetics at the University of Technology, Sydney, Australia. "If public trust in the process is eroded, then the whole idea of forensic genealogy could collapse as the public withdraws its support."

McNevin notes that developing appropriate international guidelines for the use of forensic genealogy will be a critical issue going forward, but the process is still in its infancy. Part of that process is acknowledging the limitations of the method. "Any link to a crime suspect is not sufficient evidence of direct association with the crime in question," he says. Nonetheless, he reiterates that forensic genealogy can provide valuable leads that should be used in combination with standard forensic DNA profiling and other corroborating evidence.

If wider adoption of forensic genealogy methods is inevitable, forensic laboratories around the world will need to decide how best to incorporate the technology into their workflows. McNevin recommends that agencies start by working with their own intelligence analysts to determine how the capability can be implemented within the broader intelligence cycle. They should take into account the businesses and qualifications of their investigators and prepare to tackle the complex questions that will arise from the use of forensic genealogical methods. "Finally, education of investigators and district attorneys' offices is also crucial," says McNevin.

Clearly, forensic genealogy has the potential to provide breakthroughs for law enforcement agencies, especially in cold cases where conventional avenues of investigation have been exhausted. However, wider adoption of the technology must be guided by policies that address privacy concerns and builds public trust. Technological advancements—such as the development of detailed DNA profiles from blood-sample-using next-generation DNA sequencing—may make forensic genealogy an even more powerful crime-solving tool. With that great power comes the great responsibility of addressing the public concern surrounding the ethical use of their data.

The subject of genetic genealogy will be highlighted at two workshops during the International Symposium on Human Identification, to be held in Palm Springs, CA, September 23–24, 2019.

Judy Hynds will chair a workshop titled Family Ties Using Genetic Genealogy to Solve Violent Crime. Hynds is a senior forensic scientist with the Orange County District Attorney's Office, Santa Ana, California. This workshop includes three components: 1) an overview of consumer DNA testing and whole genome testing for forensic genealogy and bioinformatics; 2) demonstration of genealogical databases, family tree building and use of common websites; and 3) discussion of best practices developed by Californian prosecutors.

Attendees will develop a holistic understanding of investigative genetic genealogy and receive material that enables them to educate their respective communities on this revolutionary tool. The workshop will also enable attendees to develop a more comprehensive understanding of legal issues surrounding genetic genealogy and best practices to balance public safety with privacy concerns.

Workshop Schedule:
Monday September 23, 2019
8:30 am–5:00 pm

Learn more

Dizhan Southard will chair a workshop titled Can You Solve Your Case Using Genetic Genealogy? Southard runs Your DNA Guide, a company providing education and assistance to anyone with DNA who wants to learn more about their family history. She also runs DNA Eyewitness, a company providing educational opportunities for law enforcement to learn how to apply DNA and family history in their investigations.

"The workshop will be a hands-on case study from start to finish," says Southard, "so attendees will learn about what kinds of cases qualify for the use of genetic genealogy, as well as the step-by-step process involved." Southard believes that the use of genetic genealogy in law enforcement is here to stay, and that it's a powerful technology that has the potential to revolutionize how investigators approach casework.

Southard is actively working to educate law enforcement agencies on how they can use genetic genealogy themselves, as well as the important protections that need to be in place regarding the use of DNA profiling data. She believes that the technology itself is not beyond the reach of most forensics laboratories. "The hard part," she says, "is learning to use that profile, and the results of a genetic genealogy database, to get what you want."

Workshop Schedule:
Thursday September 26, 2019
1:00–4:00 pm

Learn more

Using the YHRD Database for Casework Analysis

Lutz Roewer



EDITOR'S NOTE:

Funded by the National Institute of Justice, the U.S. Y-STR Database has been managed by the National Center for Forensic Science at the University of Central Florida since 2007 (Ballantyne et al. 2006), and its Release 4.2.1 (January 26, 2018) contains 35,658 Y-STR haplotypes generated by thirty U.S. forensic, academic and commercial laboratories. As of November 26, 2018, 217,758 search queries of the database had been performed.

The U.S. Y-STR Database haplotypes have been permanently transferred to the Y- Chromosome Haplotype Reference Database and the U.S. Y-STR Database will be decommissioned. Decommission dates is scheduled for June 30, 2019. Further details can be found here, including a YHRD Users Guide to enable the user to access SWGDAM compliant data output.

To learn more about the YHRD database, please register for the webinar hosted by Promega on May 8, 2019 at 9am CST, in which Dr. Roewer will present on using the YHRD database in forensic laboratories… To register click on the button below.

Learn more

Today, Y-chromosome typing is used to assist forensic investigations in a broad range of casework, such as for the rapid sexing of traces, the identification and profiling of masked male DNA component(s) in mixtures with an excess of female DNA, tracing male lineages in relationship testing or for analyzing the ancestry of unidentified corpses or unknown trace donors.

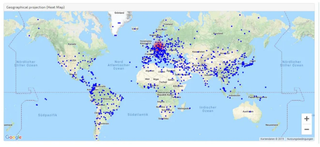
Like any forensic DNA analysis, judging the significance of a match between the trace and a reference person depends upon the population frequency of the DNA profile. Y-chromosomes are highly geographically structured, and there is the further complication that all members of a patrilineage are expected to share Y-haplotypes.

The forensic community has been proactive in establishing large quality-assured and publicly accessible databases of Y-haplotypes to address these issues. The Y-Chromosome Haplotype Reference Database (YHRD) now includes 135 national databases which have been populated over the past 20 years.

The YHRD project was started in 1999 with a collection of 3,825 haplotypes typed in 48 populations for up to 13 Y-STRs. Currently, the 60th edition of the YHRD is online with about 270,000 profiles from nearly 1,300 populations typed for up to 29 Y-STRs and a large number of phylogenetic Y-SNPs.

These data facilitate statistical analyses for a range of casework applications and interpretation purposes (e.g. frequency estimation, kinship and mixture analysis, ancestry prediction). All incoming data are validated first by the submitter using YHRD online tools, and then by the curators before being uploaded and accepted for publication.

To calculate the Y-STR profile, frequency constant and variable estimators can be used. Constant estimators (counts in a database accompanied by a confidence interval) are bound from below by the database size and are therefore often overly conservative.



More than 1,300 local population samples with more than 270,000 haplotypes in 136 countries

Expand Image

In contrast, variable estimators are independent of the database size but not from its allelic composition. The "Discrete Laplace method", used in the Y Chromosome Haplotype Reference Database (YHRD), is a model-based extrapolation method which makes use of an in-depth analysis of Y-STR profiles in more than thousand worldwide population samples. It can be used to estimate haplotype frequencies with a better approximation than the counting method.

Moreover, the YHRD is a useful resource to infer the ancestry of an unknown male person due to the availability of a consensus phylogenetic tree and geographic and ethno-linguistic information assigned to each profile. Y-based ancestry prediction and familial searching can therefore provide important investigative leads in crime cases without a suspect.

Lab Network YHRD



More than 300 laboratories in more than 70 countries

Expand Image



Lutz Roewer studied biochemistry at the University of Leipzig (Germany) and received his Ph.D. degree for molecular biology at the Humboldt-University Berlin (Germany) in 1991. His is currently an Associate Professor for Forensic Genetics at the Institute of Legal Medicine, Humboldt-University Berlin (Germany) and Director of the Department of Forensic Genetics at this institute.

Dr. Roewer has worked as a forensic expert since 1987. He is a specialist in the field of Y-chromosome genetics and its forensic, genealogical and anthropological applications. He is currently curator of the Y Chromosome Haplotype Reference Database (YHRD).

He is member of the editorial boards of the journals, International Journal of Legal Medicine and Forensic Science International: Genetics. He has been a member of the SWGDAM Y-STR committee for the past 5 years.

Dr. Roewer has authored more than 100 peer-reviewed articles in scientific journals, and publishes regular reviews and chapters in various textbooks.

Promega's hitting the road again with all the Power to Solve.

Presenting the Promega Tech Tour 2019 Workshop Series

Please join us for presentations and discussions on the latest advances in STR analysis and the forensics workflow. The workshops will feature presentations by leaders in the forensics community who will provide information and tips on maximizing success with challenging samples and improving laboratory efficiencies through workflow enhancements. The presentations will also include customer experiences and insights from using Promega products.

Entry-Level Coaching Inspires a Winning Culture

John Collins

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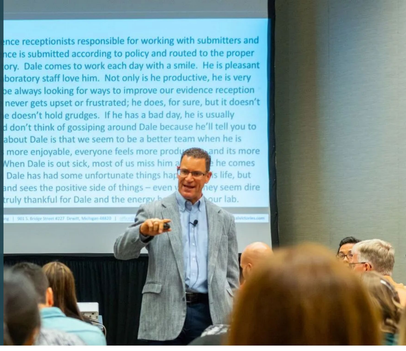

The first three years of a young forensic scientist's career matter greatly.

More personal and professional transformation occurs during this time than perhaps any other. It sets the tone for the scientist's life in the laboratory by establishing attitudes that will either expand or deflate her/his potential to be a high performer.

Whenever individuals are engaged in the process of transformation, the growth and improvement they experience depend heavily on the guidance and care of mentors who offer coaching and personal insight. Transformation in the absence of competent mentorship leaves a young scientist's future to chance.

Because there is so much to gain – and so much to lose – in the earliest years of a forensic scientist's career, it is worth a laboratory's time and energy to make it as positive an experience as possible. In fact, the entire culture of a laboratory will come to be strengthened or weakened by the experiences of its entry-level employees.

Coaching New Forensic Scientists

In forensic science, training-to-competency is a right of passage and an integral part of our occupational culture. But training is not enough. For forensic science laboratories to become high-impact teams, they must make coaching the predominant method for developing entry-level scientists who can be counted on to build the future cultures of their laboratories.

Coaching is a highly effective method for steering the growth and maturity of new forensic scientists who can become future champions of a laboratory's vision and mission. But it must be done right.

At this year's International Symposium on Human Identification (ISHI), to be held in Palm Springs, California this coming September, I will be leading a workshop in which I will outline a series of strategies for incorporating coaching into the training and development of new forensic scientists.

Learn More



Developing Your Program

In my opinion, a robust coaching program for new forensic scientists must focus on five specific priorities. All are very important, but laboratories often err in not placing an equal emphasis on each of them.

1. **Training and Assigning Coaches** - Prior to a new employee's first day of work, a laboratory should assign a coach to welcome the employee and provide encouragement during the first two or three years on the job. Ideally, the coach will be trained by the laboratory and instructed on how to interact with new employees and encourage them in a meaningful way. Not everyone is cut-out to be a coach, so select your coaches carefully. More important, if the coach is a practicing scientist or supervisor in the laboratory, it is best if he or she is not assigned to the same unit as the new employee.

2. **Technical Skills and Decision Making** - Most forensic science laboratories do a very good job at developing technical competencies. Training programs are geared toward building scientific excellence and ensuring that new employees become trusted case-workers within a reasonable period of time. Coaches can provide encouragement and guidance when new employees encounter challenges or frustrations.

3. **Self-Awareness** - Good coaches continually help their understudies pay attention to their own strengths and weaknesses. This is done by joining new employees in continuous self-reflection and discussion about how they are feeling, behaving, working, and developing as new forensic scientists. It is in this area that we see the advantage of coaches being independent of their understudies' work units. Many of the conversations can be deeply personal and even emotional as new employees work to overcome the inevitable snags and turbulence that come with transformation and personal growth.

4. **Professionalism** - As new forensic scientists develop their technical and scientific abilities, coaches can provide invaluable mentorship in helping employees build a strong sense of professionalism. Professionalism is what we see in people who are committed to their work while demonstrating self-control, interpersonal warmth, tactfulness, courtesy, and a willingness to place the interests of their teams and stakeholders before their own. Through effective coaching, the natural selfishness of youth can be replaced with humility and a genuine desire to serve others.

5. **Leadership Awareness** - We don't expect our newest employees to become supervisors in the near future. But we can expect them to appreciate the challenges of leadership and the importance of being gracious by offering support and understanding to those responsible for the administration of their laboratories. Coaches can build leadership awareness by championing the mission and vision of their laboratories, while also encouraging new employees to withhold judgement in matters about which they have limited information. Forensic science is uniquely challenging. Laboratory cultures require employees to focus on their core responsibilities without unfairly questioning the motives and methods of their senior colleagues and organizational leaders.

Today's young people are not prone to blind loyalty. They do not commit themselves to organizations or team priorities simply because they are told to do so. They commit themselves when they are inspired to do so.



Coach for Culture

If you are an administrator or organizational leader in a forensic science laboratory, know that you can develop an entry-level coaching program that can transform your organizational culture and its members in a short period of time. Your entry-level employees will benefit from a more personalized, encouraging style of training. At the same time, your coaches will set an example for other leaders by demonstrating how people are best influenced through support and positive reinforcement rather than direction and instruction.

Over many years, I have worked with a variety of clients both inside and outside forensic science. One thing remains clear. I can quickly evaluate the quality of an organization's culture by learning how the youngest, newest employees are treated and developed. It is the barometer, so to speak, that accurately reflects what is truly important to an organization.

Don't just train your new forensic scientists; coach them over the first few years of their careers - and prepare to be amazed at the benefits it brings to your entire laboratory.

Free Coaching at ISHI - Special Offer

If you will be attending the 2019 ISHI in Palm Springs this September, and if you are a forensic science professional working in a public/governmental forensic science laboratory, I will be offering free 45-minute coaching sessions. Please consider signing up for a coaching session if you:

- want to discuss your career goals
- are seeking to improve your professionalism or leadership skills
- are trying to solve a challenging problem at work
- want to become more motivated and engaged in your work

Sessions will be held on Monday September 23rd and Tuesday September 24th from 8AM to 8PM. To sign up, please visit www.criticalvictories.com/ishi. For questions, please call (517) 803-4063 or email me at john@criticalvictories.com. Space is limited so please register your interest in this program as soon as possible.

John M. Collins is senior leadership consultant and executive coach at Critical Victories. After a twenty-year career in forensic science - having worked as a scientist and/or administrator in state, federal, and local laboratories - John has become one of the most active and respected speakers and writers on contemporary forensic science issues. He is the author of HR Management in the Forensic Science Laboratory - A 21st Century Approach to Effective Crime Lab Leadership (2018, Academic Press), and his executive coaching practice continues to motivate, inspire, and develop current and future forensic science leaders in the United States and overseas.

Advertorial

Forensic Genealogy:

New Technology to Solve More Cases

Andrew Singer, Bode Technology

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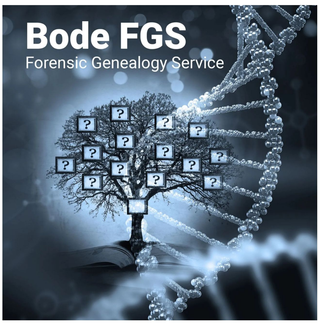
A new technological breakthrough is revolutionizing the way that DNA is used to drive investigations. Through advanced Single Nucleotide Polymorphism (SNP) testing and the use of publicly available DNA databases, scientists and genealogists are working together to link unknown DNA profiles to distant relatives to advance investigations in violent crimes and the identification of unidentified remains.

Forensic genealogy introduces a new way that DNA can be utilized to close cases. Bode Technology, a leader in forensic DNA testing, launched the Bode Forensic Genealogy Service (Bode FGS) in early 2019 as a supplemental application to forensic DNA testing. Bode's FGS program is a logical expansion of its service offerings and aligns with its mission to make a positive contribution to society, create a safer and more secure world and continue to support the law enforcement community.



Background

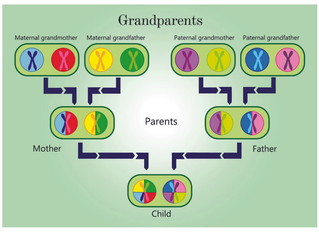
Since the DNA Identification Act of 1994, DNA has been a critical tool for law enforcement to drive investigations by identifying suspects and clearing the innocent. DNA has become the gold standard in forensic identification because of the quality standards by which crime laboratories abide and the power of identification that DNA provides. Advances in technology have allowed forensic analysts to develop DNA profiles from evidence containing just a few cells while new analytical tools can determine each contributor to a crime scene sample derived from multiple unknown contributors. These technological improvements continue to drive the importance of DNA in investigations.

Due to the visible impact of DNA testing, demand continues to increase. Legislation across the country mandating the tracking and testing of sexual assault kits is driving more cases to the laboratories. CODIS continues to grow rapidly. By the end of 2018, there were more than 915,000 unknown forensic profiles entered into CODIS, increasing at a rate of approximately 85,000 per year. With approximately a 50% hit rate in CODIS, testing cases more quickly will help solve cases and improve public safety. However, for the **~40,000** cases every year that do not result in a CODIS hit, forensic genealogy can help develop more investigative leads.

How FGS Works

Forensic genealogy relies on voluntary submissions of DNA profiles to publicly available databases. Currently, there are more than 26 million DNA profiles in ancestry related databases[1] which continue growing at an exponential rate. While not all of these profiles are available for searching by law enforcement, some of the databases, such as GEDMatch and Family Tree DNA, have allowed for law enforcement usage for violent crimes such as rape and homicide. These databases contain more than 1 million profiles and it is projected that a database of this size can identify about 60% of the individuals of European descent within a third-cousin or closer match[2]. With the combination of CODIS and forensic genealogy, the ability for DNA evidence to assist law enforcement in solving crimes vastly improves.

In order to search databases such as GEDMatch, genotyping of SNPs from the unknown evidence must be conducted. Using standard chips such as the Global Screening Array genotyping array from Illumina, the resulting SNP profiles are compared to genotypes from individuals in the database. The amount and quality of overlapping DNA segments help determine the predictive relationship between two profiles out to several generations. From this initial genetic clue, genealogists then use available resources to develop family trees to determine close and distant relatives, and in some cases the identity of the actual individual. By implementing forensic genealogy, investigators have successfully identified suspects in many high-profile cold cases, including the Golden State Killer[3].



FGS Best Practices

As forensic genealogy provides more investigative leads to law enforcement, and cases move towards adjudication, questions may be raised regarding the use of forensic genealogy. In order to preserve the integrity of the science and the justice process, it's imperative that best practices be implemented during the forensic genealogy process. These best practices should mirror those currently used in traditional forensic DNA testing. The quality system must start with proper handling of evidence and avoiding evidence consumption. DNA testing should be performed utilizing accredited laboratory practices with robust quality systems in place to ensure the accuracy of the resulting data generated. The development of a CODIS-eligible profile should be a non-negotiable step in the process, along with maintaining proper chain of custody of evidence. In the event that a profile is eligible, but no match is found, then forensic genealogy could be considered.

Best practices should also be followed while performing genealogical research. This starts by only utilizing experienced and board-certified genealogists to help develop these investigative leads. The genealogist should move forward by evaluating only credible sources, applying stringent quality and ethical standards, and employing diverse investigative methods to address complex questions of identity and kinship. Finally, law enforcement should ensure that any investigative leads are ultimately verified in an ISO 17025 accredited laboratory. These are all critical elements that have been thoughtfully and carefully incorporated within Bode's FGS program.

Solving More Cases

For more than 20 years, Bode has been a leader in forensic DNA analysis. Bode's highly refined protocols and rigorous quality systems are used every day to identify perpetrators of crimes, identify the missing and exonerate the innocent. Bode has incorporated the same principles to deliver a forensic genealogy solution for our clients. Through the use of accredited testing laboratories and board certified and experienced genealogists, Bode has developed a high-quality, turnkey solution for crime laboratories, law enforcement agencies and attorneys to take advantage of this new investigative tool.

Forensic genealogy should not, and will not, replace the need for CODIS. In order for DNA to remain the gold standard for DNA testing, it is important to maintain the integrity of a highly-regulated system. However, it is also important to recognize the impact that genealogy is having in solving cold cases by providing key investigative leads to law enforcement. As the general public continues to voluntarily populate the database, this will enable law enforcement to identify suspects when other avenues have been exhausted. Bode FGS is a tool that can prevent future crimes and provide the justice that victims deserve.

For more information visit https://bode-labs.com/pages/bode-forensic-genealogy-service **or email** Bode.Service@Bodetech.com.

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Forensic Genetic Investigation of Unidentified Human Skeletal Remains Discovered in the Himalayas:

Exploring the Value of Biogeographic Ancestry in the Absence of a DNA Database

Dr. Angie Ambers, University of New Haven

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There are many scenarios encountered in forensic casework in which bone may be the only viable sample type for DNA testing, including fires, terrorist attacks, natural disasters, war conflicts, airline crashes, homicides, human trafficking, and mass graves. In these cases, the condition of recovered human remains can range from relatively intact to highly degraded and/or fragmented. In some homicides, perpetrators dismember the bodies of their victims and hide or disperse the individual parts across a wide geographic region in order to impede discovery of the remains, as well as to hinder subsequent identification efforts. Additionally, in terms of forensic investigation, unidentified remains in an advanced state of decomposition often essentially become 'skeletal' cases because the soft tissue is too decomposed to obtain usable data for positive identification. Decomposition of soft tissue prevents visualization of distinguishing tattoos or scars on the skin, and can nullify the possibility of a fingerprint match or facial feature recognition. In these cases, bone is the most viable sample for DNA testing and therefore may be the only avenue for identifying a decedent.

In the U.S., thousands of children and adults vanish without a trace and/or under suspicious circumstances every year. The latest statistics available indicate that the National Crime Information Center (NCIC) contains 88,089 active missing persons records and a registry of 8,634 unidentified (skeletonized) remains. However, the actual number of unidentified skeletal remains that exists likely is substantially higher, given that many victims remain undiscovered to date.

Fortunately, the U.S. has a well-established database containing DNA profiles of missing persons and/or their relatives, for comparison when skeletal remains are recovered and to assist in identification efforts. However, although forensic DNA typing is widely conducted around the world, many countries still have not developed and/or populated reference DNA databases to assist in cases involving unidentified human remains (UHR).

India "missing persons" case: Skeletal remains discovered in the Dhauladhar Range of the western Himalayas (Himachal Pradesh)

In 2017, I traveled twice to the Gurgaon/Gurugram region of Delhi, India to train scientists from various Indian states and the Maldives Police Service on the processing of bone samples in forensic DNA casework. During the workshop training, I had the opportunity to work on an unidentified skeletal remains case with Dr. Vivek Sahajpal and Dr. Arun Sharma, two exceptional forensic DNA scientists from the State Forensic Science Laboratory in Himachal Pradesh.

Himachal Pradesh is a northern state of India, situated in the western Himalayas. Most of the state lies in the foothills of the Dhauladhar Range, with a scenic landscape of snow-clad mountains, deep gorges, densely forested valleys, large lakes, and cascading streams (**Figure 1**). Due to the majority of the region, tourism is a major contributor to the state's economy and growth. Popularly referred to as "The Land of Wanderlust," it is a well-known trekking and climbing destination for adventure- and spiritual seekers. Additionally, Himachal Pradesh is the home of the Dalai Lama and the Tibetan government-in-exile. The Tibetan presence is reflected in the state's Buddhist temples and monasteries, as well as during its vibrant festivals and celebrations.

In 2016, human skeletal remains were discovered along Indrahar Pass, a mountain pass in the Dhauladhar range of the Himalayas, located at an altitude of 4,342 meters (14,245 ft) above sea level. Indrahar Pass is near the town of Dharamshala in Himachal Pradesh and attracts substantial tourist traffic during the trekking season (April – October). In the last two decades, approximately 30 trekkers on tours have gone missing in Himachal Pradesh, notably near the tourist hotspot of Dharamshala. The majority of these tourists were of European descent, including trekkers from the United States, Australia, France, England, Russia, Italy, Sweden, Ireland, Yugoslavia, and the Netherlands. When unknown skeletal remains were discovered in 2016, Indian authorities understandably assumed that the remains belonged to one of these missing tourists, and implored forensic scientists to attempt to recover DNA in an effort to identify the decedent.





Figure 1. Map showing the location of Himachal Pradesh, a northern state of India situated in the middle of the western Himalayas. This scenic "land of wanderlust" is a popular tourist destination for adventure seekers and trekkers. In 2016, human skeletal remains were discovered along Indrahar Pass in the Dhauladhar range of the Himalayas, near the town of Dharamshala.

The skeleton discovered was incomplete, consisting only of disarticulated cranial bones, a lower mandible (sans teeth), bones from the pelvis, the diaphyses of five unassigned long bones, and severely degraded fragments of a pelvis. Additionally, four disarticulated (loose) molar teeth were recovered from the scene.

The gender of the remains was unknown prior to DNA testing. Anthropological gender assessment can be challenging without access to the cranium and the pelvis, two of the most sexually dimorphic skeletal elements in the human body. In this case, the skull and pelvis were not intact, and only portions of each were recovered. Cranial plates were disarticulated and fragmented, and only portions of the highly degraded pelvis were found. Therefore, it was necessary to determine the gender of the decedent using genetic analyses.

In addition to the poor condition of the skull and pelvis, many of the long bone epiphyses were missing or severely degraded, likely from advanced bone diagenesis due to environmental exposure. *Bryophyta* (moss) was growing on the epiphyses of a few of the long bones recovered. Additionally, fragmented long bones were selected for DNA testing after extensive visual examination to determine the most viable candidate samples for typing success. These two bones were the most structurally intact, with no visible fractures to the diaphyses, and appeared to contain the most dense cortical bone material, as estimated by comparative manual handling of all available bones in the sample set. Another factor guiding the long bone in the sample set was not selected for testing due to a significant longitudinal fracture that extended along the length of the majority of the diaphysis, which likely would have compromised the quality of DNA within the bone microstructure over time.

Multiple DNA extractions using 0.5g- 1.0g bone or tooth powder were performed in a designated low copy number (LCN) area of the Thermo Fisher Scientific laboratory in the Gurgaon/Gurugram region of Delhi, India. DNA quantification and autosomal STR genotyping were conducted using the Quantifiler® Trio and Globalfiler™ kits (Thermo Fisher Scientific, Waltham, MA), respectively. In total, extractions were performed on nine different bone cuttings and two molar teeth. A complete 24-locus consensus profile was obtained, with complete concordance in allele calls between the various bone sections and tooth tested. Moreover, DNA was not detected in any of the extraction reagent blanks or amplification negative controls. A female analyst (myself) conducted all testing for each set of reactions, including surface cleaning, bone grinding, DNA extractions, quantification, PCR amplifications, and genotyping. Results confirmed that the unknown skeletal remains were male.

Although the decedent's endogenous STR profile had been determined, a DNA database was not available for kinship analysis and/or comparison to exemplars. At this stage, the only feasible option for identifying this individual involved reaching out to government officials in the "home" countries of each of the 30+ missing tourists and requesting that family reference samples be collected for comparison to the profile obtained from the skeletal remains. Aside from logistical challenges in undertaking this extensive effort, there are sensitivity issues that must be considered with such an approach. The families of these 30+ missing persons had already experienced a tremendous amount of trauma and grief due to unanswered questions associated with the disappearance of their loved one. Although discovery of a loved one's remains and the ability to provide a proper burial often can provide some measure of "closure," the reality in this case is that we had no reasonable level of confidence in assuming that the remains belonged to one of these families.

Assessment of biogeographic ancestry: Y-STRs and ancestry-informative SNPs (aiSNPs)

Since STR genotyping revealed that the remains were male – and Y haplogroups can be used to infer biogeographic ancestry – a decision was made to perform additional testing with markers that are specific to the Y chromosome using the 27-locus Yfiler™ Plus multiplex by Thermo Fisher Scientific). With the alleles in the decedent's Y-STR haplotype, Y haplogroup prediction was performed using the ancestry feature and metapopulation tool of the Y-STR Haplotype Reference Database (YHRD). The YHRD-based prediction was cross-checked with HaploGrep software. Analysis of the Y-STR alleles in the profile obtained from the skeletal remains revealed that the decedent was of Asian descent (not European), as shown in the heat map and diagram generated by YHRD (**Figure 2**).

Figure 2. Biogeographic ancestry prediction for unidentified skeletal remains discovered in the western Himalayas of India, using the ancestry feature and metapopulation tool of the Y-STR Haplotype Reference Database (Y-HRD) (accessed 10-18-2017). The remains were originally presumed to be of one of the 30+ European and American tourists who have gone missing in the last two decades while trekking in Himachal Pradesh.

In addition to traditional capillary electrophoresis (CE)-based methods for Y-chromosome typing and the utility of Y-STR markers for inferring biogeographic ancestry, the advent of massively parallel sequencing (MPS) technology has greatly expanded the tools available to scientists for predicting the ancestral origin of unidentified skeletal remains. In this case, samples from the decedent were further tested using the ForenSeq™ DNA Signature Prep Kit (Verogen, San Diego, California USA) and the MiSeq™ MPS platform. The ForenSeq™ panel co-amplifies 24 Y-STRs, 95 human identity SNPs, 56 ancestry-informative SNPs, 22 phenotype-informative SNPs, 29 global autosomal STRs (plus amelogenin), and 7 X-STRs.

The ForenSeq™ Y-STR data were concordant with the Y-STR haplotype obtained using the traditional CE-based method. However, the additional ancestry-informative SNP (aiSNP) data provided an opportunity to support or refute the ancestry prediction previously inferred using YHRD. Of the 56 aiSNPs in the ForenSeq™ panel, results were obtained for 43 loci and were analyzed using the Forensic Research/Reference on Genetics knowledge base (FROG-kb) and FROG-kb (introduced in 2012) as an open access web tool that utilizes SNP data to infer biogeographic ancestry of an unknown sample via comparison to known populations data sets contained within the software. The aiSNPs obtained from the skeletal remains discovered in Himachal Pradesh were compared to 139 reference populations in FROG-kb. Statistical calculations support that the decedent is of Asian descent, which is consistent with the prediction made by YHRD. **Figure 3** shows the top 10 most likely populations of origin for the unknown decedent, along with statistical likelihoods.

Population (region, sample size [N])	Probability of genotype in each population	Likelihood ratio (LR)
Sri Lankan Tamil (SLT) (Asia, 206)	1.96E-13	1
Kurukh (Asia, 60)	1.01E-13	1.94
Tripuk (TU) (Asia, 206)	1.47E-14	13.4
Punjabi (PJL) (Asia, 192)	4.423E-15	40.8
Gujarati (GIH) (Asia, 206)	1.466E-15	96.5
Tharu (Asia, 26)	1.046E-15	96.5
Bengali (BEB) (Asia, 172)	2.68E-15	73.4
Gujarati (GIH) (Asia, 206)	0.774E-16	341.0
Kachari (Asia, 36)	3.634E-16	542.0
Telugu (Asia, 68)	1.71E-17	8760.0

Figure 3. Biogeographic ancestry prediction for unidentified human skeletal remains discovered in the western Himalayas of India, via comparison of endogenous autosomal ancestry-informative SNP (aiSNP) data to 139 reference populations in the Forensic Research/Reference on Genetics-knowledge base (FROG-kb) (http://frog.med.yale.edu/). The top 10 FROG-kb results are shown, indicating the most likely populations of origin for the remains.

Ultimately, although the identity of this individual remains unknown to date, the possibility of causing further emotional trauma to families of 30+ missing European and American tourists was circumvented due in large part to recent advances in forensic DNA technology. Although massively parallel sequencing (MPS) is also known as next generation sequencing (NGS), the reality is that it is no longer "next generation." Research laboratories have been using the technology for years, and many of the accompanying assays/kits have been through the validation process in preparation for implementation into forensic DNA casework. The changing face of forensic genetics now offers the capability to obtain biogeographic ancestry and phenotype information from unknown samples, which could provide important investigative leads in cases that otherwise may have progressed in the wrong direction. The capability to obtain far more information from an evidentiary sample than was previously possible in the traditional casework landscape likely will alter the way forensic genetic investigations are approached in the future, particularly with unidentified human remains (UHR).

Acknowledgements:

DNA testing of these unidentified remains discovered in the Himalayas was supported by Thermo Fisher Scientific, the State Forensic Science Laboratory (Himachal Pradesh, India), and the UNT Center for Human Identification. I am grateful to Dr. Sahajpal and Dr. Sharma for entrusting me with this case. It was an honor to work with such talented scientists, and to acquire new friends in the process. Lastly, many thanks to Les Gerren, Yogendra Dawadkar, Alima Agarwal, and Jeet Singh for their assistance, and support.

Dr. Ambers holds a Ph.D. in molecular biology (with emphasis in forensic genetics and human identification), as well as master's degrees both in forensic genetics and in criminology. She is currently an Associate Professor in the Forensic Science Department of the Henry C. Lee College of Criminal Justice and Forensic Sciences, at the University of New Haven. She began her career in forensic genetics studying under the late Arthur Eisenberg, one of the pioneers of forensic DNA testing in the United States.

Dr. Ambers worked as a full-time forensic geneticist for the University of North Texas (UNT) Center for Human Identification for 8 years, specializing in DNA analysis and genetic characterization of unidentified human remains (UHR). In 2016, she received the "Outstanding Forensic Genetics Alumni" award for her successes and contributions to the field. Her research has involved an investigation of alternative methods (e.g., whole genome amplification, DNA capture) for improving autosomal and Y-STR typing of degraded and low copy number (LCN) DNA from human skeletal remains and environmentally-damaged biological materials, as well as a development/optimization of a DNA-based multiplex screening tool for the sequestration of fragmented and commingled skeletal remains in mass graves.

Prior to joining the Henry Lee College, Dr. Ambers was an adjunct professor at the University of North Texas for 12 years (teaching molecular biology, genetics, heredity, human anatomy/physiology). In 2008, she developed the curriculum for a course in forensic molecular biology, and for a graduate-level DNA analysis/methodology to undergraduate students enrolled in UNT's FERAC-accredited forensic science certificate program.



Before pursuing her doctorate, Dr. Ambers was lead DNA analyst and lab manager of the UNT DNA Sequencing Core Facility. Her casework has involved DNA testing of an American Civil War guerrilla scout, several Finnish World War II soldiers, unidentified late-19th century skeletal remains discovered by a construction crew in Deadwood, South Dakota, unidentified skeletal remains of Special Operations soldiers killed during the 1974 Turkish invasion of Cyprus, skeletal remains of two soldiers from Napoleon's army, skeletal remains exhumed from Prague Castle in the Czech Republic, skeletal remains of soldiers from the Seven Years' War (1756-1763), and bone samples purported to belong to a member of Jesse James gang killed during a bank robbery in 1876).

Dr. Ambers co-presented a workshop on "Advanced Methods for DNA-based Analysis of Skeletal Remains" at the 26th International Symposium on Human Identification, and in 2015 was an invited speaker at an international bone workshop/conference in Prague, Czech Republic. In 2017, she traveled twice to India to train scientists from various Indian states and the Maldives Police Service on the processing of bone samples in forensic DNA casework. She specializes in characterization and identification of contemporary, historical, and archaeological human skeletal remains. Her casework and research has been published in various peer-reviewed journals (Forensic Science International: Genetics (FSI: Genetics), International Journal of Legal Medicine, Legal Medicine, BMC Genomics, Croatian Medical Journal, The Journal of Heraldry) and has received prizes in numerous local and national newspapers, including The Washington Times. Dr. Ambers' most recent casework includes DNA analyses of five sets of human skeletal remains associated with the French explorer La Salle's last expedition, and twenty-five sets of skeletal remains associated with Scottish royalty and the Kings of Aragon.

In addition to skeletal remains cases and research, Dr. Ambers collaborated with the National Institute of Justice (NIJ) and the Forensic Technology Center of Excellence (FTCoE) in 2014 to develop and disseminate a formal report on the use of "Familial DNA Searching" in cold case investigations, an approach which recently garnered media attention for its use in catching the Golden State Killer. She also recently (2017-2018) served as principal lead on a U.S. State Department grant to conduct human trafficking in Central America through the application of forensics. As part of the program objectives, she traveled to three Northern Triangle countries (Guatemala, El Salvador, Honduras) to perform gap assessments of government laboratories and train personnel in forensic DNA analysis, with the goal of providing quality casework methods based on ISO 17025 standards. In addition to providing both lecture and tactile training in DNA analysis methods to Northern Triangle laboratories, Dr. Ambers was part of a consortium built to help these countries develop and implement DNA databases to assist in the identification of missing persons related to human trafficking.

Experience Old Hollywood's Hidden Oasis:

Palm Springs



Tara Luther

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Hollywood began from simple roots and was once a sleepy farm town until the movie makers moved in. Yet, in the span of a decade, humble apple orchards became movie production companies, and actors and starlets had taken over. The early 1920's was a time of prosperity, and Americans loved to go to the movie theater. By 1929, there were 25,000 cinemas in the country and an average of 100 million Americans went to the movies on a weekly basis.

Stars like Rudolph Valentino, Clara Bow, John Barrymore, Mary Astor, and Charlie Chaplin became household names. Many early actors had "morals" clauses in their contracts, restricting their actions within 100 miles of the studio, and causing them to flee to Palm Springs.

As with many things, the truth is not so scandalous. Often, actors were under contract to stay within driving distance of the studio in the event of reshoots, and due to the proximity of Los Angeles and the relaxing environment, many chose to take up residence in the oasis of Palm Springs. In fact, Palm Springs became known as Hollywood's Playground.



As more and more actors flocked to the Coachella Valley, a neighborhood of stars was developed in the 1930's and 1940's. Termed "The Movie Colony", the area is named for those who called it home, such as Cary Grant, Dinah Shore, Van Johnson, Tony Curtis, Gloria Swanson, and Jack Benny. Located in central Palm Springs, the neighborhood now consists of four hotels, two parks, and 170 homes (some of which were designed by renowned Modernist architects like E. Stewart Williams, William Cody, and Donald Wexler). Frank Sinatra made the neighborhood "his" in the 1950's when his Twin Palms Estate became known as party central. He even had a flag adorned with a martini that would be raised to indicate he was in town and the drinks were flowing.

While the times have changed, you can still get a sense of what it was like to be a star in Hollywood's Playground. Start your day by joining a tour of the former homes of celebrities led by Desert Adventures Jeep Tours. Begin at Frank Sinatra's party pad, and work your way through the homes of Elizabeth Taylor, Elvis Presley, Debbie Reynolds, Kirk Douglas, Ann Miller, Katherine Hepburn, and Liberace. If a tour isn't enough to sate your appetite, some homes are for rent.



Next, take a stroll downtown where Palm Springs hosts its own "Walk of Stars". Three hundred gold stars embed the sidewalks and immortalize notable people who have lived in the Palm Springs area. Or, drive through town and take note of the street signs paying homage to the stars, such as Frank Sinatra Drive, Gene Autry Trail, or Jack Benny Road.

If you're feeling adventurous, saddle up like a cowboy in a Western movie at Smoke Tree Stables and explore the trails among the palms, just like Cary Grant used to do. Or venture to the outskirts of Palm Springs and hop on the aerial tramway which takes you to the top of Mt. San Jacinto and provides stunning views of the Coachella Valley below.

Less than an hour north of Palm Springs lies Joshua Tree National Park. Reminiscent of Dr. Seuss' Truffula trees, the park's namesake yucca tree inhabits a unique landscape that has been a favorite backdrop of filmmakers filled with nature trails among large desert rocks.



Finish your day with a dip in the pool and head out to dinner at any number of famous establishments. Try the risotto with mushrooms at the Purple Room, a supper club with a swanky, 1960's vibe where the Rat Pack once dined. If you prefer seafood, head to top-rated Copley's on Palm Canyon, which was once a guesthouse on Cary Grant's estate, and sample the ahi tacos. Or, check out Melvyn's, located in the Inglenook Inn, a hotel built in the 1920's. An homage to old-school continental dining, it was favorited by Frank Sinatra, and you can still order his favorite dish, Steak Diane, prepared tableside.

This September, help us celebrate the 30th anniversary of The International Symposium on Human Identification, which will be held in the Palm Springs Convention Center. Kirk Douglas once said, "If you want to see a star, don't go to Hollywood. Come to Palm Springs." It was true in the 1920's, and it's still true today. One look at the tranquil sunset over the purple mountains surrounding the valley, and you'll know why.



Calendar of Events

Deadlines

June 9, 2019

Oral Abstracts due

June 9, 2019

Interesting Case Abstracts due

July 9, 2019

Poster Abstracts due

July 15, 2019

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Familial DNA Searching: Early Breakthroughs

The Grim Sleeper

Across the Atlantic, initial adoption of familial DNA searching was slower than in the UK. Although the Denver, Colorado Police Department began studying the technique in 1999, California became the first state to make familial DNA searching legal in 2008. Cases in which the technique was employed had to meet two criteria: 1) the crime scene DNA profile had to be a single-source profile; and 2) all other investigative leads must have been exhausted.

The first—and arguably the most infamous—test of familial DNA searching in California was in pursuit of a serial killer known as the Grim Sleeper. The nickname derives from the 13-year break that the perpetrator apparently took after a killing spree that started in 1985.

Beginning in 2001, as part of an effort to implement new DNA analysis methods, the Los Angeles Police Department re-examined several cold cases. Continuing efforts on the Grim Sleeper case identified a DNA match among three victims killed in 1987, 2002 and 2003. Although familial DNA searching was used in 2008, it failed to provide any matches. However, the following year, the arrest of Christopher Franklin in a felony weapons charge provided a DNA sample that resulted a partial match to the Grim Sleeper's DNA profile. Further investigation led to the arrest of Lonnie David Franklin, Jr—Christopher's father. Lonnie Franklin's trial began on February 16, 2016, and he was convicted on all counts of murder and attempted murder, receiving a death sentence on June 6 that year. The case captured so much public interest that a documentary film was released in 2014, Tales of the Grim Sleeper.

[https://www.rottentomatoes.com/m/tales_of_the_grim_sleeper/]
For more information on the Grim Sleeper case, see our interview with Rock Harmon, retired Senior Deputy District Attorney for Alameda County, California.
[https://www.ishinews.com/catching-the-grim-sleeper-an-interview-with-rock-harmon/]

Familial DNA Searching: Early Breakthroughs

The Canal Killer

Forensic genealogy, which enables investigators to bypass the constraints of only searching DNA databases of known offenders, also has its share of high-profile cases. Foremost among these was Fitzpatrick's Canal Killer case. The murders of two young women occurred in 1992 and 1993; in each case, the woman had gone for a bike ride along the Arizona canal in Phoenix and never returned. The first woman's decapitated body was discovered in a parking lot and, later, her head was found in the canal. The second victim's body was also found floating in the canal.

"I happened to be at ISHI in 2014," Fitzpatrick says, "and I had the opportunity to speak to the cold case investigators there…telling them about forensic genealogy." The Phoenix police department told Fitzpatrick they had a cold case that she might be able to help them with. They later sent her their Y-STR DNA testing results. Fitzpatrick searched several public genealogy databases to compare the profile from the case.

"I applied the software to find a match in those hundreds of thousands of profiles, and it came up with three exact matches to the name 'Miller'," Fitzpatrick recalls. "They had a list of two thousand suspects, and having the name narrowed it down to five." Further investigation narrowed the search down to one individual, based on information from a forensic professional society and the FBI. The Phoenix police obtained a DNA sample from the suspect, the profile matched the one Fitzpatrick analyzed, and the rest is history.

Familial DNA Searching: Early Breakthroughs

The Saturday Night Strangler

In 1973, two teenage girls left a nightclub in Swansea and decided to hitch a ride. They never made it home; their bodies were found in the woods near Port Talbot, and they had been raped and strangled. The team of detectives working on the case—the first serial killer identified in Wales and dubbed the “Saturday Night Strangler” by the public—soon became overwhelmed by its magnitude. More than 10,000 drivers who could have possibly given the girls a ride had to be interviewed. Further complicating the investigation, several events near the town had led to a large influx of strangers, who could also be potential suspects. By the middle of 1974, the case—then known as the Llandarcy murders--was closed in the absence of any strong leads and the evidence was shelved.

In 1990, a forensic biologist named Colin Dark with Forensic Science Services (FSS), a government-owned company, reviewed the case. He found that much of the evidence had been damaged by mice and the damp conditions in Port Talbot. Dark was interested in attempting DNA analysis, but the techniques available at the time were inadequate for the challenging task.

Ten years later, another FSS scientist, Jonathan Whitaker, was working with a recently developed, innovative DNA profiling technique called low copy-number (LCN) analysis. LCN uses the polymerase chain reaction (PCR) to amplify DNA from very small or damaged samples, making it possible to obtain a DNA profile from just a few cells. However, due to the method’s extreme sensitivity, it is also susceptible to errors from contamination and PCR artefacts, and it has gained its share of controversy over the years.

Whitaker realized that LCN might hold the key to finally unlocking the mystery hidden in what was left of the evidence from Port Talbot. Using LCN, his team was able to obtain a DNA profile of the perpetrator. However, after searching the UK national DNA database of known offenders, the team was unable to find a match. Despite that initial failure, the case was officially reopened.

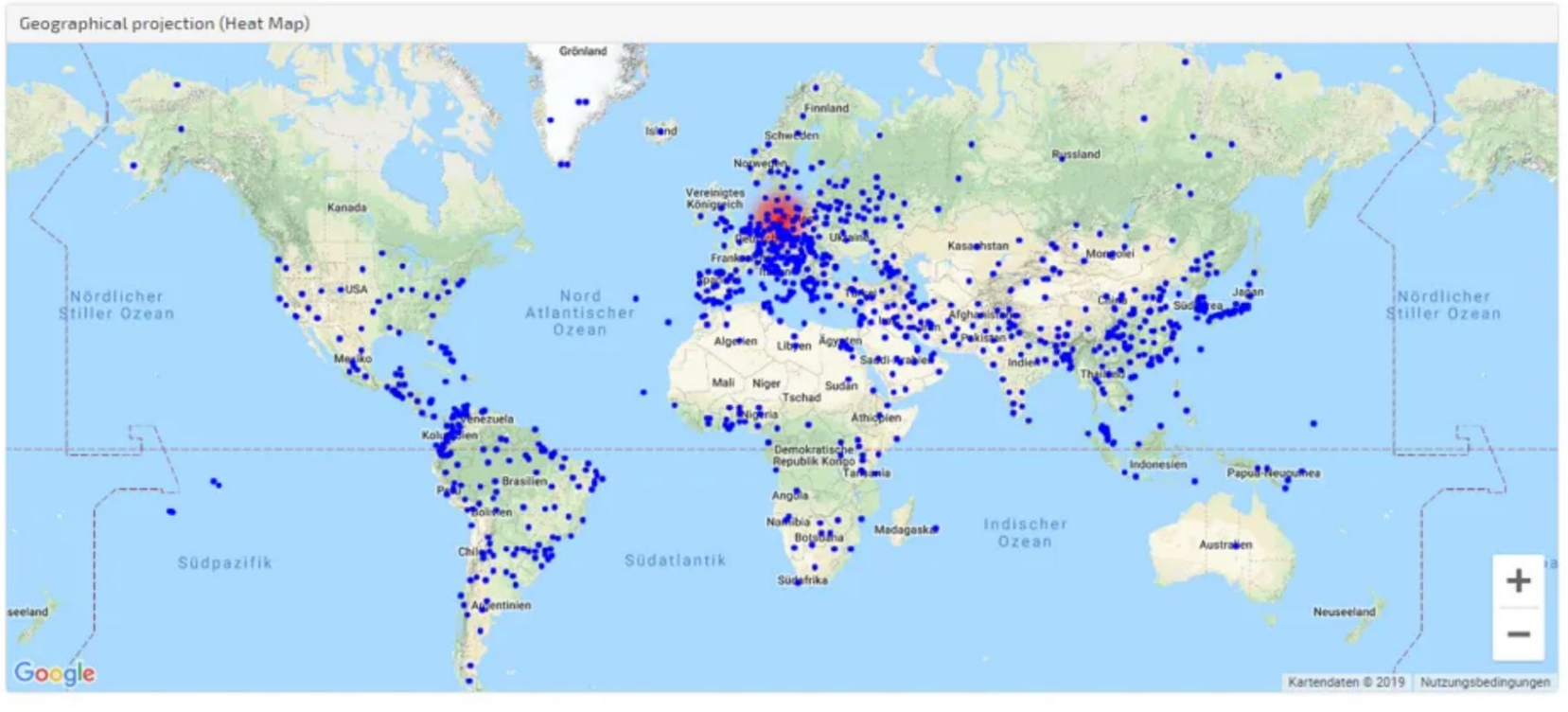
A team headed by detective chief inspector Paul Bethell inherited the case archives. They had 35,000 persons of interest and knew their best hope of solving the case rested in obtaining DNA profiles, but it was clearly impractical to do so for the entire list. It took eight months to narrow down the list to around five hundred. Following further investigation, they obtained DNA samples from 353 men. None of those profiles matched the one obtained from the evidence.

Meanwhile, another cold case provided evidence that was to become vital in solving the Llandarcy murder mystery. In 1973, another teenage girl who had hitchhiked was brutally raped and strangled. Her body was found in Tonmawr, less than 10 miles from Port Talbot, three months before the murders of the girls in the Llandarcy case. The DNA sample showed that the same killer was responsible for all three murders.

Whitaker—working with Dark on the Llandarcy murders—had a stroke of inspiration. Although the DNA profile of the perpetrator hadn’t matched any of the profiles in the national database, he suggested that a close family member of the killer might be represented in the database, thereby providing a partial match—50%, in the case of a son or father. One name rose to the surface: Paul Kappen, a well-known car thief. The name was familiar to Whitaker. One of the five hundred people on the original Llandarcy list was named Joseph Kappen, but the investigators hadn’t obtained his DNA, because he’d died 11 years earlier. Under Whitaker’s direction, the team managed to obtain DNA samples from Joseph Kappen’s ex-wife and daughter. Together with Paul Kappen’s sample, these results gave them a match to 75% of the DNA profile from the Llandarcy murder case.

One final piece of evidence was required: a DNA sample from Joseph Kappen himself. The forensic team had sufficient evidence to exhume his body, despite his family’s objections. On a night that seemed straight out of a horror movie—complete with pouring rain, lightning and thunder—Kappen’s coffin was dug up and his remains sent to a local mortuary, where samples were taken from a femur and teeth. The DNA profile was a perfect match, providing the first triumph for Whitaker’s technique of “familial DNA searching”.

More than 1,300 local population samples with more than 270,000 haplotypes in 136 countries



More than 300 laboratories in more than 70 countries

