

15 DNA AND LAW

Wild, Woolly World of DNA

The first in a series of articles in the *New York Times*, titled "The DNA Age" presents case histories of people whose DNA tests are turning out to be mixed blessings, arousing more expectations than may be justified. From the adopted twins who are looking for financial aid after finding out they are part African and part Native American to the man raised a gentile attempting to invoke the law of return to Israel following the revelation his DNA matched Ashkenazi Jews, the series by Amy Harmon apparently intends to explore the two sides of DNA—the answers it brings, along with the new questions it raises.

On another front, Indian tribes routinely refuse to accept DNA evidence. According to the article, though, this has not deterred prospective new enrollees.

"It used to be 'someone said my grandmother was an Indian,' " says Joyce Walker, the enrollment clerk who regularly turns away DNA petitioners for the Mashantucket Pequot tribe, which operates the lucrative Foxwoods Resort Casino in Connecticut. "Now, it's 'my DNA says my grandmother was an Indian.'"

The title of the first of the series is "Seeking Ancestry in DNA Ties Uncovered by Tests." One of the featured DNA test takers was a customer of DNA Consultants.

April 13, 2006

DONALD N. YATES and TERESA A. YATES

Uncle Sam Wants Your DNA

U.S. Set to Begin a Vast Expansion of DNA Sampling

Julia Preston

New York Times

February 5, 2007

"The Justice Department is completing rules to allow the collection of DNA from most people arrested or detained by federal authorities, a vast expansion of DNA gathering that will include hundreds of thousands of illegal immigrants, by far the largest group affected.

"The new forensic DNA sampling was authorized by Congress in a little-noticed amendment to a January 2006 renewal of the Violence Against Women Act, which provides protections and assistance for victims of sexual crimes. The amendment permits DNA collecting from anyone under criminal arrest by federal authorities, and also from illegal immigrants detained by federal agents."

February 5, 2007

DNA and Prisons

Exoneration Using DNA Brings Change in Legal System

Solomon Moore

New York Times

October 1, 2007

"State lawmakers across the country are adopting broad changes to criminal-justice procedures as a response to the exoneration of more than 200 convicts through the use of DNA evidence.

"All but eight states now give inmates varying degrees of access to DNA evidence that might not have been available at the time of their convictions. Many states are also overhauling the way witnesses identify suspects, crime labs handle evidence and informants are used. ...

"Nationwide, misidentification by witnesses led to wrongful convictions in 75 percent of the 207 instances in which prisoners have been exonerated over the last decade, according to the Innocence Project,

a group in New York that investigates wrongful convictions. ...

"The legislative reform movement as a result of these DNA exonerations is probably the single greatest criminal-justice reform effort in the last 40 years,' said Peter J. Neufeld, co-director of the Innocence Project."

October 1, 2007

When Objects Become Subjects

Review of Paul Brodwin, " 'Bioethics in Action' and Human Population Genetics Research"

Population-genetics experts who lecture in the groves of academe or trudge through the jungles of the Amazon are not immune to racist bombshells and political dynamite. In 1991, Stanford geneticist Luigi Luca Cavalli-Sforza announced a project to study human genetic diversity. The ponderous monograph that issued forth in 1994 became as revered as it was unreadable. His *History and Geography of Human Genes* posited two main limbs in the human DNA tree, the African and non-African, with the latter branching off into Europeans (Caucasians) and Northeast Asians. Included in Northeast Asians were the so-called Amerindians. Amerinds were closest in genetic distance to Northern Turkic, Chukchi and other Arctic and Mongolian peoples.

Little did Cavalli-Sforza and his team expect to encounter any opposition to their benign project, much less withdrawal of funding by the U.S. government and United Nations, but this is exactly what happened. The genial professor was surprised one day by a letter from a Canadian human-rights group called the Rural Advancement Foundation International. The group demanded he stop his work immediately. It accused the Human Genome Diversity Project of biopiracy, stealing DNA from unsuspecting indigenous people and mining it for valuable information pharmaceutical companies could use to make drugs Third World people could not afford.

Paul Brodwin's article published in 2005 in the journal *Culture, Medicine and Psychiatry* (29:145-78) reviewed this controversy, which had some positive repercussions in forcing researchers to rethink colonialist attitudes toward their subjects. But in the second case of

"bioethics in action," Brodwin painted a much-more-ambiguous picture. It concerned the use of genetics by the ethnic group called Melungeons of Tennessee and Virginia to prove identity claims and press their ideas of special entitlements.

In the section of the article titled "The Reinvention of Melungeon Ethnicity," Brodwin chronicles the conflict between scientific genetics and the Melungeons' demand for collective recognition. Complicating this issue is that the academics were by no means certain among themselves about who or what Melungeons were from an anthropological perspective. A rancorous standoff between Virginia DeMarce and N. Brent Kennedy was matched by the tendentious nature of the Melungeons' own theories and assertions about themselves. Was there even such a thing as Melungeons or were they simply genealogical ghosts and lurid creations of popular journalism? Did they truly have some black and American Indian ancestry? Was the title only to apply to people in and around Newmans Ridge in Hancock County, Tennessee, or be extended to a wide range of persons of mixed ancestry like the Carolina Turks and Lumbee Indians? If the Melungeons went back before the arrival of Europeans, could they seek legal recognition as an indigenous American Indian tribe?

Questions abounded, and it seemed all of them were murky, emotionally charged and political. Unlike the Human Genome Diversity battle, neither party seemed to gain any advantages in the free-for-all. There were apparently no lessons to be learned on either side. At the end of the day, everyone just gave up and went home, exhausted.

Brodwin obviously sympathizes with the forces of the Academy in all this. He throws his lot in with the geneticist Kevin Jones, who found "he did not control the goals of research or the interpretation of findings." The Melungeon fracas illustrated "the political and conceptual vulnerabilities of human population genetics." In my opinion, however, Brodwin missed the point. Whom do university professors and academic researchers serve, if not the public? They should rejoice that so many of the great unwashed (even in the hills and hollers of Tennessee) are engaged by and even interested in their research.

And if they cannot achieve a satisfactory dialogue with their lay critics, whose fault is that? The debate should continue, not be swept under the rug of philosophical reflection. Whatever else they might be,

Melungeons are people. As such, they should not be dismissed when they become intractable.

September 1, 2009

Clearing out the Closets

Proposal Raises Bones of Contention

Anthropologists lobby to retain Native Indian skeletons for study

Rex Dalton

Nature 450/469 (November 22, 2007)

"Alarm is growing among anthropologists in the United States over a plan that could empty institutions of about 120,000 human skeletons currently stored for research purposes," according to this news item in *Nature*."

A new proposal under the Native American Graves Protection and Repatriation Act (NAGPRA) would repatriate the bones at museums, universities and federal facilities across the nation to local Native American tribes "even if the skeletons are not culturally identifiable to the tribes."

Affected would be ancient skeletons similar to the disputed Kennewick Man specimen from Washington state.

"The rules would be disastrous," says Phillip Walker, an anthropologist at the University of California, Santa Barbara. He is a former member of NAGPRA's seven-person review committee and helped prepare the American Association of Physical Anthropologists (AAPA) comments.

Donald Yates comments: Even if the Federal Government decides to extend ownership of ancient remains and relics to locally visible American Indian groups for disposal as they see fit, authorities will have to take American Indian spokespersons' word on who they are, how old they are, where they came from and how long they have been where they are—in other words, the very questions science seeks to answer by study of those remains and relics. It's like giving students the chance to set course policies and grade themselves. I say this as a university professor and American Indian.

American Indian cultural continuity and collective identification is quite recent. The oldest Indian writing, the *Walam Olum*, says the last-but-one phase of Lenape history comprised 36 reigns of their chiefs from their settlement along the Ohio to a figure known as Lekhihiten the Author down to about 800 C.E. Before about the year 1 C.E., all these nations were still in Asia around the area of Lake Baikal. The Cherokee, Iroquois and Lenape lived together in the former territory of the Moundbuilder Indians (likely a mixture of Mexican Indians and peoples from the Old World) for an estimated 500 years. Another 36 generations passed until the Lenape arrived in their last dwelling place along the Delaware River, where they were encountered by the Dutch and English. A special black wampum belt was made to commemorate this event. When the beads were counted in historical times, the belt showed that the Lenape arrived in their easternmost homeland in 1396.

So much for ancient roots on the land, and the Algonquian Indians are regarded as the grandfathers by other Indian nations. There are more Indian nations of Algonquian speech today in North America than any other. Yet they, like all others, appear to be relative newcomers, historically, genetically and culturally.

Scholars such as Isabel Stewart have suggested that the Athabaskan Navajo and Apache Indians, today's largest and most-favored tribes in the eyes of the Federal Government, did not arrive in the American Southwest until shortly before the Spaniards.

It's time to stop throwing political sops and go on with the science of objective study of peoples, whether that involves historical accounts, genetics or whatever means. The truth is far more empowering than anything else.

November 22, 2007

Is Science Only for Scientists?

That's the import of a trio of opinions in this week's *Nature*. One of them, "Genetics without Borders," criticizes a "UK government scheme to establish nationality through DNA testing (as) scientifically flawed, ethically dubious and potentially damaging to science." The "scheme" is a peer-reviewed program of the UK Border Agency to test whether some 100 asylum-seekers are Somali nationals. The testing uses a combination

of SNPs, mitochondrial DNA and Y chromosome, plus other forensic means, to determine whether they are actually Somali or not. (That is, within a high degree of probability, since all inductive conclusions are probabilistic.)

The editors of *Nature* fulminate against such methods. Yet, these are the tools of the trade used by law enforcement officials and academic geneticists, to say nothing of commercial DNA testing companies.

"The idea that genetic variability follows national boundaries is absurd," they scoff.

They are not impressed by the work of fellow scientists John Novembre et al., "Genes Mirror the Geography of Europe," in *Nature* 456, 98–101; 2008), saying that the idea that genetic variability follows man-made national boundaries is absurd. What is absurd is the idea that genetic variability is not molded and delineated by language, culture and historical events—the foundation of national boundaries. It seems to escape the opinion makers that Novembre et al. found that genetic patterns echoed linguistic divisions in Europe. This makes eminent sense in that courtship between most males and females is conducted in the same language. That means within the same nationalistic boundaries.

Random "mating" of an exogamous nature as envisaged by them is not in the nature of humans. It may be a generalization that can be formed of evolution, which is judged in sweeping retrospective, but it is not true of living people at any given time, in any given land or country. Until the 20th century (and perhaps even today) most people marry someone of the same rather narrowly defined ethnicity as themselves. In fact, until the modern period, an Englishman was most likely to marry a woman whose house was situated only an easy walk away. His horizons—and, thus, the eligible gene pool—was limited to a 24 mile square specifically labeled his "country."

Geneticists are wont to see human genetics in terms of geologic time, whereas the time depth and landscapes of history are more pertinent. The authors end by urging geneticists, "and indeed all scientists," to nip the government's "scheme" in the bud before the public finds out about it and an uprising ensues. This call to action seems to combine scientific cant with a patronizing view of the public.

Lay persons, and sometimes people outside one's narrow scientific specialty, just cannot be trusted to get anything quite right, can they?

Another day's blog will address the other two articles in this week's *Nature*, which exhibit similar mandarin attitudes.

October 6, 2009

US, EU Move to Regulate Direct-to-the-Consumer Genetic Testing

Discussion is accelerating in the United States and European Union to regulate private genomic testing that provides consumers medical information, according to *Science* magazine and the *European Journal of Human Genetics*. No mention is made in the reams of white papers about ancestry testing, but some of the pitfalls and bureaucratic morasses in the thinking about true genetic/medical testing are fairly ominous, if not silly.

"Although there has been speculation about the potential psychosocial harms of testing (that is, genomic medical testing), such as an increase in anxiety or encouragement of fatalistic behavior, there are, to date, few studies addressing these concerns," writes the reporters for Policy Forum in the Oct. 8 issue of *Science*. "The limited evidence tends to be reassuring, even for risk information associated with relatively serious ailments ... however, the scope for potential harm from unnecessary or unproven treatment after genetic risk assessment is an important unstudied question" (pp. 181f.).

We commend scientists and physicians for finding a new field of study divorced from reality but have to wonder what they will do about ancestry testing once they have conquered and tamed Frankenstein's elder monster. We suggest the following guidelines (just kidding, folks):

- Labeling on Internet sites and Zen Shopping Carts that explicitly states, "The claims for this ancestry product have not been evaluated by the U.S. Government Accountability Office (GAO), U.S. Federal Trade Commission (FTC), House Energy and Commerce Committee, Food and Drug Administration, National Institutes for Health or Department of Bioethics and Humanities, University of Washington School of Medicine, Seattle, WA 98195 USA."

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- Predictive ancestry information may be hazardous to your progeny.
- No animal has been harmed in the production or clinical evaluation of this ancestry test.
- If you discover you have ancestry you did not expect, take a deep breath. Then take a healthy dose of skepticism, followed by two aspirins and a glass of water.

We're waiting for the next gambit from the genius bar in Washington!

November 28, 2010

Obstructionist Participants

In an article titled "Indian Tribe Wins Fight to Limit Research of Its DNA," Amy Harmon reports that Arizona State University has agreed to pay the Havasupai Indians of the Grand Canyon \$700,000 and return blood samples collected from them for diabetes studies in the 1990s.

The university's Board of Regents apologized to the tribe for ... well, that part of the story is not clear. Not informing them that the samples might be used for "wider-ranging genetics"? Not informing the subjects that they reached negative conclusions and found no "diabetes gene" as they believed they had in a Pima Indian study? Not getting permission (no, that was done with simple-to-understand, signed consent forms, as was proper)? Coming to different conclusions about the Havasupai's origins than their myths and legends? Allowing people to "get degrees and grants" using "our blood"? Implying that the Havasupai are inbred? One Havasupai woman found that offensive.

Many tribal members were disgruntled because they were still suffering from diabetes after the university "took their blood."

Sorry, Havasupai Indians, a project participation consent form is not a treaty. But if you signed it, you should honor your word. You cannot go back now and require the researchers who use your samples to come to research conclusions that suit you and be silent about those that do not. Science (and society) don't work like that.

The tribe's dictates to the university were mercenary, and the university's decision to pay the tribe off, wrong. The case sets a bad precedent and places another barrier between Indian peoples in remote areas and the real world.

April 22, 2010

Rigged Genetics

If the facts don't fit the evidence, change them ...

We always suspected the genetics community of clinging to stale dogmas and being slow to acknowledge emerging new evidence about American Indians. But we did not dream that their officiousness extended to changing the information given by test subjects to bring it into conformity with preconceived conclusions.

Not until we heard Marcy's story.

"Over the years, I've heard complaints that (a DNA testing company) is not really responsive when you have questions about unexpected results," Marcy said. "They usually suggest further testing, which, of course, means more revenue to them.

"I've had some major disagreements with (a DNA testing company) over how they list results for mitochondrial haplogroup ancestral origins. ... I found out they were taking dozens of T2's who had listed their earliest-known female ancestor as being from America or the United States, changing this and placing them in the 'unknown' category. They claimed that because our haplogroup was designated European, our ancestors couldn't be from the United States!

"Now, this was nonsense, because at the same time, they allowed people to claim other similarly-colonized western countries, like Cuba. It's my opinion that if participants list a country of origin for their earliest-known female relative, that should be what is on the web page, not something assigned by (a DNA testing company) because as they told me, it may 'confuse people,' or contradict current scientific data.

"As a consequence (the DNA testing company's) publicly reported ancestral origins has nothing to do with our haplogroup's ancient Cherokee clan mother. The chips should fall where they may."

Now, this is not professional behavior on the part of a DNA testing company and it prevents new findings from coming to light.

In a study of 52 individuals claiming direct maternal descent from an American Indian woman, mostly Cherokee, we found that they were unmatched anywhere else except among other participants. Haplogroup T emerged as the largest lineage, followed by U, X, J and H. Similar proportions of these haplogroups were noted in the populations of Egypt, Israel and other parts of the East Mediterranean.

DNA testing companies do a disservice to their customers and to science by failing to call results as they appear without doctoring them. It is time geneticists stopped bringing all American Indians over the Bering Straits and forcing test subjects into the Procrustean bed of outmoded theory.

August 23, 2011

Jim Bentley, DNA Frontiersman

We interviewed one of Chromosomal Labs Bode Technology's senior staff members, Director of Sales and Marketing Jim Bentley, to get his perspective on industry changes over the past 35-plus years.

When did you first get interested in DNA?

JB: I'll have to preface my answer with a few remarks on "the early days." When I graduated from Arizona State University in the 1970s, DNA testing as we know it, was not really a field that was in existence. There was not a lot going on. The little work I did with chromosomes was using electron microscopy. I worked in the biochemistry department, however, and performed hundreds of assays using poly-acrylamide gel electrophoresis, mainly for separation of proteins. This technique, although improved and streamlined remains in use today for DNA-STR separation. The field we're in today where we can determine a person's profile and compare it with others for forensics for relationships, ancestry, missing persons, adoptions and the like, that technology hadn't been developed yet. It wasn't quite as easy as it is today.

Tell us more about the evolution of DNA testing.

JB: It basically began with blood groups and types. The first paternity test was done in a court case with Charlie Chaplin in the 1940s. He was excluded as the father, but the court said he could go ahead and pay child support anyway—probably, because he could afford it. Since that time, scientists started moving past groups and types into some other techniques. Human Leukocyte Testing (HLA), DQ-Alpha, and Restriction Enzyme STR testing (RFLP) are examples of the evolution of DNA testing.

The big breakthrough came when Dr. Alec Jeffreys at the University

of Leicester discovered STR testing in England the late 1980s. He used STR profiling on the Colin Pitchfork case. Colin Pitchfork became the first criminal convicted on the basis of DNA evidence and as a result of a mass DNA screening operation. He was charged with raping and murdering two teenage girls. Since that time, the forensic community has really refined the techniques to perform STR testing. They've made it simpler and more accurate. It's really moved exponentially in the last 20 years. Today, competent biologists and chemists can produce excellent results, every time. Dr. Jeffreys has been knighted for his contributions.

So, what got you involved?

JB: I came out of college as a chemist, one interested in the medical field. I started out working in clinical chemistry and toxicology. The work we did with DNA was extremely limited and very costly. But I did stick with a career in clinical chemistry. Within four years after graduating from school, I was managing a clinical laboratory in Houston, Texas, called National Health Laboratories. It was a laboratory of about 100 scientists and support staff. After mergers, acquisitions and such, that company remains as Lab Corp. (It performs more than 1 million tests on more than 370,000 specimens each day.)

What opportunities for professional growth did you have over the years?

JB: Through taking a lot of continuing-education coursework, I became proficient and qualified as a general supervisor in clinical chemistry, toxicology, hematology, parasitology, microbiology, serology—everything except for tissue work like histology and cytology, which was done by certified medical experts in those specialties. My interests kept me in touch with the staff pathologists, however, as well as all the rest of the laboratory. Though my present-day field did not exist at the time I graduated, by staying current I was able to benefit from the changes and be part of an emerging valuable service provided not only to the medical community but also to the forensic one, and the general population at large.

What are some famous cases you've been involved with ... that you can talk about?

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JB: Actually, that's my problem. We've been involved in a number of high-profile cases, but we're not allowed to talk about any of them. Most have been on the forensic side, serial-killer trials in Arizona, also in California, some that made the news in Florida ... Texas ... Georgia.

Were you involved in catching the Grim Sleeper?

JB: Actually, that's an ongoing case in Los Angeles we are familiar with, but we didn't do the work on it, so we can talk about that one. The importance of the Grim Sleeper case has to do with familial testing and autosomal DNA. It was termed the Grim Sleeper case because there were a number of homicides that took place beginning in the mid-1980s, all with the same basic MO (modus operandi), and then the murderer went underground for 14 years. The victims were typically prostitutes shot with a firearm. In 2010, a suspect, Lonnie David Franklin Jr., 57, was arrested and charged with multiple counts of murder. He has not yet been convicted, nor the evidence against him tested in court.

How was DNA used to catch him?

JB: So, here were a number of cold cases, but they were being tracked, and the law enforcement authorities in Los Angeles continued to monitor progress. The sole survivor of one of the Grim Sleeper's attacks furnished a description of him as a black man in his 30s, along with other details. According to her story in the press, he lured her into an orange Ford Pinto, shot her in the chest with a pistol, took Polaroid's and raped her, leaving her for dead. In 2008, the body count was 13, and a \$500,000 reward was put out for "America's Most Wanted."

It became the first use in California, and one of the first three cases in the United States to use familial DNA searching, that is, drawing on the FBI's CoDIS database to match one family member's profile with a suspect's profile. The L.A. police were able to provide a close partial match to Franklin's crime-scene profile with that of his son, whose CoDIS markers were on file for a minor crime. They then set up a kind of mini-sting operation at a pizza parlor in Buena Park, where they knew the family liked to eat.

Undercover detectives masqueraded as waiters and busboys. When the family left, they whisked away an unfinished pizza slice. The crust yielded DNA, which police linked on a more-solid basis to Lonnie

Franklin. It was the first high-profile case in which a family member's DNA had been used to catch a criminal. The ACLU and others had been critical of familial searching on grounds of privacy, and there is still a lot of debate over familial searching because it might open up the search and include those who hadn't committed any crime.

Did this help produce new commercial products like the "cousin finders"?

JB: Only a few states are doing familial searching, and they are pretty guarded about it. It's hard for me to make a connection. Certainly, these developments have been concentrated in the past three or four years, but the use of this technique is spreading.

Are people legitimately suspicious about DNA databases?

JB: Fears surface from time to time. There have been claims that keep popping up that someone's going to take everything that's in the database and use it to determine genetic deficiencies that could lead to medical issues down the road. Once it was speculated that if such information was released, insurance companies would begin denying people coverage based on their profiles.

This is the mother of conspiracy theories, isn't it?

JB: It really is. For the most part—not for everyone—the vast majority of the markers we are using are in the "junk DNA" area. That is, they don't by themselves "do" anything or give you genetic information on the face of things. There may be one or two markers that possibly could be construed as yielding some medical information—such as a trisomy at vWA or TPOX (a CoDIS locus). But by and large, you are not going to be able to do any medical diagnostics with the markers we run. Usually trisomies such as Down's syndrome would be physically expressed and not hidden. It's a little different with SNP panels (single nucleotide polymorphisms) such as those run by 23andMe. With a high number of those, it's entirely possible to predict medical predisposition. That's what they base their business on.

Let's talk some more about the CoDIS database.

JB: It's important to realize that even law enforcement doesn't provide

much access to the CoDIS (Combined DNA Identification System) databank. That's something I have to give the FBI credit for. They have developed a system that is secure. It's the DNA administrator at each facility who has undergone FBI training and uploads the data under very strict rules, and they are notified of any "hits" that involve them, but, otherwise, there is very little access, and the use of the database is very even across the country. There are not a large number of portals that can be used to access the CoDIS database. There are several hundred law enforcement laboratories that are running profiles across the country, and the database is best thought about on three different levels: LDIS, SDIS and NDIS, local, state and national versions. Between our labs in Phoenix and Virginia, we've tested over a million profiles for entry into CoDIS. That's about one-tenth of the entire number. I can tell you there is tight security. Hundreds of thousands of investigations have been aided by a DNA hit (we don't like to say "match" so much, because statistically nothing is 100 percent) generating a lead.

How did you get bitten by the genealogy bug?

JB: I've always been fascinated with ancestry. I think it came about because my father took an interest in discovering our family's roots and had to do so at the time by traveling to Salt Lake City, Utah, and poring over whatever records he could find there about our fathers, and great-grandfathers, and great-great-grandfathers, and so forth. He had tintypes of some of the relatives. We had various pieces of the puzzle. My father pretty much consolidated everything back to William Bentley, who settled in Rhode Island in the early 1700s and had come from Bedfordshire, England. He put together a book for family use. He glorified a few of them and left a few out that weren't ready for glorification. For the sensitivity of some of the relatives, he left a few details out, but it was a pretty solid piece of work. For me, it kind of fostered this interest in ancestry and its importance. Certainly, when I started at Chromosomal Labs • Bode Technology, we started looking at the various tools that could be used. Our history, to be sure, is passed down from generation to generation. Initially, we were using mitochondrial DNA, Y-SNP's and Y-STRs and then autosomal STRs to determine how we're connected to general and specific individuals back to the Revolutionary War days and how you are linked with the world

population, what your roots were. I have a particular Y haplogroup of G2a, which is not one of the more common ones.

Hmm ... you and Joseph Stalin.

JB: [Laughs]. Is that what his haplogroup was? Uh-oh! He was one of the worst. Well, I got interested in G2a and hooked up with about 50 other Bentleys, and we identified our founder patriarch haplotype. I get emails from them on a regular basis. The other thing we tried to find out was what in the world were all these G2a's doing in England. I don't know. But one of the things I find in the literature most often was that the Sarmatians were horsemen that gave the Romans a pretty rough time. Eventually, they were decimated. The Romans took their remaining cavalry and pressed them into service for 12 to 13 years or longer. Some were dispatched to Hadrian's Wall. Now, do I know for a hundred percent certainty that's where I came from? No, but it's fun to regard that as a hypothetical personal history.

You have a Scythian gene, don't you?

JB: Yes, I do, according to the analysis DNA Consultants did for my autosomal ancestry. The work Dr. Yates has done on the rare alleles supports a lot of the stuff the family has been putting together for years and years. I was very pleased to get my Rare Genes from History report back showing I had the Scythian gene. That seems to go along with the Sarmatian theory about the Bentleys.

How do you see the industry changing over the next few years?

JB: I can speak best about changes I am seeing in the field. They're getting closer to having rapid DNA testing on a chip. This gives flexibility to those who want to use DNA as "point of use" testing. The FBI, this past year, came out at the Promega conference and said that within the next two years they would like to see wide adoption of "point of use" testing. The IntegenX prototype allows you to put your swab into a cartridge, insert it into the instrument on the fly and get your STR results in a few hours. Previously, Rapid DNA testing was not only time-consuming and lab-bound, but it was very expensive. It cost several hundred dollars in reagents alone. As the technology improves to allow two-hour testing, in our lab or on a chip, reagent and personnel time

continue to drop. Now, the FBI would like to see point-of-use testing in every booking station in the country. At the last show, I also saw an instrument from Illumina that would run Y-STRs, mtDNA and autosomal DNA profiles simultaneously on one sample. Another change that is coming is we will see an expanded profile becoming the standard, perhaps something similar to the GlobalFiler kit from Life Technologies with its 24 loci. With the new technology you can increase the speed for amplifying the specimen by five times and achieve nine times the discriminating power or resolution.

Any final remarks?

JB: The DNA testing field is on the threshold of even greater accolades of appreciation both from the scientific community and the public. If DNA wasn't even in anyone's mind 20 years ago, soon it will be part of everyone's daily lives.

January 26, 2013

Junk DNA

We are our DNA. It was not a surprise to find that our entire DNA is functional ("Junk DNA Isn't Junk, and That Isn't Really News"). The surprise is in the discovery of what we can do with what we once thought was junk.

According to that recent NPR article, "It is a massive control panel that regulates the activity of our genes." Our genes "would not work" without it. So, instead of being junk, they are critical and "control how cells, organs, and other tissues behave." But we can also now read the markers and mutations on this "panel" and discover much more information than knowing it is just working efficiently for our body. This knowledge is considered a "major medical and scientific breakthrough" (Ibid.). We just have to read it well.

But first, what is DNA exactly? John Wilwol, in his recent NPR article, "A 'Thumb' on the Pulse of What Makes Us Human," quotes Sam Kean, author of the book *The Violinist's Thumb And Other Lost Tales of Love, War, and Genius, As Written by Our Genetic Code*, as saying that DNA is what makes us who we are. Wilwol further quotes

Kean to help us understand what DNA is and how it differentiates from genes: "While DNA is a thing—a chemical that sticks to your fingers," he writes, genes are more conceptual in nature, "like a story with DNA as the language the story is written in."

So, if DNA is a language how are we able to read it? All parts of our genetic code are now readable and meaningful. Marker locations (loci) are spread across one's entire genome, not confined to one's male (Y chromosome) or female (mitochondrial) DNA. (This is how sex-linked, haplotype tests that follow one line at a time are analyzed). Different mutations are handed down genetically—different according to the region where one's ancestors lived.

Because of this new ability to read markers, consumers are now able to buy autosomal DNA tests that provide a complete analysis of where all one's ancestors' ethno-geographic origins may lie—reflecting the entire spectrum of all their ancestral lines. Not just one line at a time as in haplotype testing. This is next-generation ancestry DNA testing and the wave of the future. Anyone can take an autosomal DNA test because it does not rely on X or Y chromosomes. A female is unable to take the male Y-linked test and must entice a male in her line, if one is available, to take this test. The future is now in many ways.

What else can you learn from autosomal DNA testing? Anne Tergesen, in a recent article in the *Wall Street Journal*, quotes Megan Molenyak, author of, *Hey America, Your Roots Are Showing*, as saying that this relatively new test deciphers the amount of DNA shared between those whose common ancestors lived within the last half-dozen or so generations. Tergesen explains it like this, "Y-DNA and mitochondrial DNA can connect people whose common ancestors lived recently or hundreds of years ago. But to find out how closely you are related—and to locate relatives besides those on your direct maternal or paternal lines—you will need an autosomal DNA test." (Of course, you would both need one to compare) and "in general, the more DNA two people share, the closer their connection".

But there are even more things on the horizon with autosomal DNA for the future. According to a recent *Smithsonian* article, "Fetal Genome Sequenced Without Help From Daddy," a fetus' entire genome can now be sequenced from the mother alone with "99.8% accuracy." How is that possible? It was just "last month clinicians announced that they could

sequence a fetus's entire genome by taking samples from the pregnant mother's blood and that of the father to be" ("Fetal Genome"). Now, they have a "more difficult, but more complete method (that) uses DNA from the pregnant woman and the fetus to map out every last letter of the fetal genome ...with the advantage that it can pick up mutations that a fetus has but its parents do not" (ibid.). Rob Stein quotes Dr. Alan Guttmacher, director of the National Institute for the Child Health and Human Development in a recent NPR article, "Genome Sequencing For Babies Brings Knowledge and Conflicts," as saying, "Instead of screening for currently something like 30 conditions, it would allow you to screen for hundreds if not thousands, of conditions) at birth." He goes on to say that "one could imagine a day where knowing someone's entire genome sequence at birth, you could really begin to think about structuring health care, their dietary choices, their exercise choices ... early in life, in a way that would have an impact on truly lifelong health." Stein says that this gene sequencing could "spot babies that are prone to conditions such as obesity, diabetes, heart attacks or cancer" and that we may soon be "sequencing all babies when they're born." It could be a wonderful tool. But we are not there yet.

According to Rob Stein in another NPR article, "Perfection is Skin Deep: Everyone has Flawed Genes," scientists have determined we are all more flawed than they thought. "Researchers discovered that normal, healthy people are walking around with a surprisingly large number of mutations in their genes." Chris Tyler-Smith of the Wellcome Trust Sanger Institute in Cambridge, England, and his colleagues analyzed the DNA of 179 people from several countries who volunteered their genetic information to the 1,000 Genomes Project.

In a paper published in the *American Journal of Human Genetics*, researchers reported that though none of the people whose DNA was studied were sick, the average person has about 400 minor flaws and one or two that could contribute to disease. Tyler-Smith says, "It's a bit surprising that people should be walking around apparently healthy yet we're seeing known disease-causing mutations in their genomes," he says. "But the answer was that these tended to be for mild and very often late-onset conditions. Things like heart disease, an increased risk of disease or developing cancer."

On its website, the American Diabetes Association highlights the

interaction of genetic and environmental factors: "You inherit a predisposition to the disease then something in your environment triggers it. Genes alone are not enough."

So, the problem is not so much with the analytical tool but rather the possibility of over-interpretation. Again, we just have to read it well, with the same critical eye for what is written *in* us as that written *by* us. And who knows what else we will soon be able to discover from reading our DNA?

January 14, 2013

Genes Are the Teachers

Sam Kean, author of the book *The Violinist's*, says, "While DNA is a thing—a chemical that sticks to your fingers," genes are more conceptual in nature. He says genes are the story while DNA is the words that make up the story.

Chromosomes are long, cylindrical-like blocks that contain the words allowing them to create the story, similar to children's brightly colored alphabet blocks. Alleles are any alternative forms of the story (read gene) that may occur at any given locus (point) on the chromosome, attaching themselves like people do on a train or bus or subway—grabbing on for safety and, perhaps, deciding to go a different route. Or tell a different story. Reminds me of the crazy, alternate endings that one sometimes finds on a DVD. For instance, there are Internet rumors of alternate endings for *The Titanic*—Jack surviving and swimming to shore and Rose dropping the diamonds in the ocean as her granddaughter watches. Personally, I don't like alternate endings. But alleles? Alleles we need. They make us unique.

What do alleles do? Alleles hang out together like cute little couples or a group of friends on the chromosomes. Just like people, some of them are dominant, and some of them are shy (recessive). If you get two dominant people in the party, you know who is having their way. It is the same with alleles though it usually takes more than two people to really get anything done just like it usually takes more than two alleles to determine a trait. It might take quite a crowd of them. They might call another party (interact with another site of alleles). All of our genetic traits are caused by alleles interacting. The genes we inherit are all the

same as humans, but it is how the alleles express them that makes us unique (*Encyclopædia Britannica*).

What about genes and DNA? According to Ananya Mandal, M.D., in her article, "Genes—What Are Genes?", DNA is the "chemical information database." The DNA carries the "complete set of instructions for the cell" and genes are the "working sub-units" of DNA. You might also see DNA as a library of information and the genes as the teachers and librarians who use the library and tell the students (each cell) how to work (how to make proteins), as each gene has a set of instructions, roughly speaking, for making a certain protein, be it melanin or insulin. Mary Kugler, R.N., in her article, "What are Genes, DNA, and Chromosomes?" says a gene is a "distinct portion of a cell's DNA" and has "coded instructions for making everything the body needs." These genes are "packed in bundles" of chromosomes. Something like all the librarians packed into offices that are really two offices in one.

How many chromosomes do we have? Everyone has 23 pairs of chromosomes (46) but only one is sex-linked. The other 22 pairs are autosomal chromosomes determining the rest of the body's makeup (Kugler). What do chromosomes do? Keep DNA in its place.

Or if they were people, alleles would be the artists, genes the teachers, chromosomes the disciplinarians, and the DNA would be the great Wizard of OZ—except with real instructions.

October 13, 2013

Too Big to Feel

We don't often write editorials in this space. Normally, you will see nothing but news in the DNA Consultants Blog. Some sparse marketing messages may appear whenever we have a new product or study. But the FDA's "stop and desist" letter last Friday to personal genomics giant 23andMe has sent shockwaves through the industry. Although we are not in the business of providing medical information to customers, only ancestral background analyses, we feel compelled to weigh in on the FDA's warning, which we think is overdue.

First, it is important to note that the FDA took action against 23andMe because the company has been selling an unapproved diagnostic device and medical service. The FDA demanded 23andMe "immediately

discontinue marketing the Personal Genome Service" after years of protracted and unsuccessful requests for proof of safety and efficacy to back up the company's marketing claims.

A check of 23andMe's website on November 26 showed little change in the promises it makes to consumers. Splashed across the welcome page in large letters was "Get to know you." The site boasted having "Reports on 240+ health conditions." In language that sure sounds medical to us, it speaks of carrier status, health risks and drug response.

An appeal to parents suggests, "Find out if your children are at risk for inherited conditions, so you can plan for the health of your family." Under "drug response," the company advises that you can take "information on how you might respond to certain medications" from your \$99 DNA test to your next doctor's visit.

The FDA found "some of the uses for which Personal Genomic Service is intended are particularly concerning." A false positive result for the BRCA gene, for instance, could cause a patient to remove breasts or ovaries to avoid getting cancer. A false negative could lead patients into an unfounded sense of security and make them ignore that an actual risk exists. An inaccurate result for warfarin drug response could lead a patient to self-manage their dosage or skip it altogether, leading to "illness, injury, or death." Some pretty dire concerns.

DNA Consultants simply does not do DNA testing for medical information. If a customer calls and asks for such a service, we explain that is something they should discuss with their healthcare provider. Neither our marketing nor fulfillment of tests contains any medical language.

We specialize in ancestry analysis exclusively and believe we do that better than anybody else.

Of course, the field of genetic screening has made enormous progress over the past 10 years. We monitor many of those advances in our blog. But we do not believe the field is by any means "there" yet, certainly not ready to be packaged and hawked to consumers. We doubt it ever will be. Or at least we hope not.

Despite popular anthems of genetic determinism, your health is not all in your genes.

But your ancestry certainly is. Find out what your ancestry is and you will be able to form an idea of what your ancestral medical history might

look like, going beyond the two generations of family medical history covered by standard questionnaires at the doctor's office. But that part is entirely up to you and your healthcare providers.

No company is too big to fail. Even the largest are subject to oversight by the government as well as consumer pressure and the natural forces of the market. Nor is any company too big to feel. We hope 23andMe will respond both to the FDA and the public with understanding and responsibility, not indifference and arrogance.

November 26, 2013

Top Stories of 2013

What were the top genomics and DNA testing stories of 2013? It has definitely been the year that DNA from genetic testing has been rocking in the news. I have chosen what I think are the top three.

First, last June, there came the Supreme Court decision that police can now legally take DNA from anyone they arrest. They then enter this into a database where they can match it with existing samples (Dan Noswoltiz, "It's Now Legal for the Police to Collect DNA," *Popular Science*). Since we live in a rather scary world since 9/11, I thought that might be constructive, at first glance, until I realized that the key word is "arrest" not "charged." What is the difference? People are falsely framed and arrested every day as well as arrested for minor offenses. Consequently, there any number of ways this could be abused. Even if someone is declared innocent, guess what? They still have your DNA.

(Do you want others to have your personal genomic data? That is a question you might want to ask any DNA testing company you use as well. What do you do with my DNA? Do you keep it and put it in a database or share it with others?)

Also, in the same month, the Supreme Court ruled that human genes cannot be patented, except for synthetic genes. This was in response to a lawsuit between "Myriad Genetics, a medical diagnostics company, and the Association for Molecular Pathology" (Young). Many saw this as a win for women with an elevated genetic risk of breast and ovarian cancers as well as researchers and scientists (Richard Wolf, "Justices Rule Human Genes Cannot Be Patented," *USA Today*) Why? Myriad had

a monopoly on the gene; as a result, no one else could produce or manufacture it, and the genetic test was inordinately expensive. Not every woman has a bank account matching Angelina Jolie's. Her decision to first take the genetic test and then have a preventive double mastectomy because of her high risk of breast cancer brought this case to the forefront.

But it also played into the biggest and most-controversial story of the year concerning genetics testing for disease and the battle between the FDA and 23andMe. The FDA told them to stop selling health- and medical-related information with their genetic tests. Some see this as the FDA stealing their right to their own personal genetic information. Since there is no genetic destiny for disease because of other lifestyle and epigenetic factors (Carl Zimmer, "Hope, Hype, and Genetic Breakthroughs" *Wall Street Journal*), others see this as part of a process to ensure that these tests are accurate and not misinterpreted. However, knowing you have a high genetic risk for a disease might mean you make better lifestyle choices. I think in the end it will not spell the end for direct-to-consumer genetic health services, but, hopefully, the industry will not only be resurrected, but there will also be better guidelines for the field as well as the consumer spurring a wider market so consumers have more choices. I am looking for that silver lining in the New Year.

January 2, 2014

Mired in Miracles

This past summer, the Supreme Court decided biotech companies could not patent human genes extracted through genetic testing. What are the implications of such a decision for businesses and consumers? This decision is advantageous for both. Why? It keeps prices down for the consumer assuring most anyone can afford needed genetic testing and prevents a monopoly on any genetic testing done for both research and medical purposes. That means more than one company can offer a particular type of genetic testing, and it is more affordable and accessible to the public.

The brouhaha all started with the company Myriad Genetics. First, this company discovered with genetic testing the "precise location and sequence" of two genetic mutations that are a significant risk factor for breast cancer, BRAC1 and BRAC2 genes. After making this discovery,

they created their own form of genetic testing to find these mutations and decided to patent it. Angelina Jolie made the decision to have a double mastectomy after discovering she had these genes based on a very expensive test. It cost some \$3,000 because of Myriad Genetics' monopoly. ("Biotech Companies No Longer Have the Right to Patent Human Genes").

Of course, the fight ended with the Supreme Court's decision, but our vision of it was of Angelina Jolie cast in the limelight once again. However, instead of being a hell-on-wheels wife doing her best to use every means to kill her husband in *Mr. & Mrs. Smith*, she sounded more like any woman afraid of getting breast cancer. The only difference was she could foot any bill they handed her.

Perhaps Angelina Jolie did not realize it, but the feeling many probably had when reading articles concerning her genetically inherited condition was not one of empathy alone but fear—fear upon the realization that most could ill afford genetic testing of this kind. Where did that leave the majority of women? Many, no doubt, were left wondering if they had the same genetic mutations especially if they had a high risk factor like Angelina Jolie's. But Angelina Jolie was not left to wonder. She got tested and made a decision.

The Supreme Court made the right call. No one should have a monopoly on genetic testing. Genetic testing should not just be for the Angelina Jolies of the world but affordable for anyone who needs it.

June 18, 2014

Forensic Fantasies

If DNA testing is to be done correctly, ~~one~~ has to follow certain procedures and protocols. Otherwise, a criminal investigation will misinterpret the results from contaminated samples. If the DNA testing results are not analyzed properly, the consequences could be devastating. It could even mean life or death. Amanda Knox's trial didn't follow correct forensic procedures.

Amanda Knox was a tourist in Italy in 2007 where she and her boyfriend were convicted of the murder of her British roommate, Meredith Kercher. Many Italian papers at the time suggested that they had wild orgies. It made for tabloid heaven. Where are the DNA testing clues? There is not any evidence. According to Claudia Dempsey, author

of *Murder in Italy: Amanda Knox, Meredith Kercher, and the Murder Trial That Shook the World*, in her *Seattle Post-Intelligencer* article, "The Carnival Ride Continues," the judge who acquitted them and released the pair in 2011, Judge Claudio Patrillo Hellman, said there was *nessuna prova* (no evidence) against them. The prosecutor, Giuliano Mignini, believes, according to Dempsey, in a "four-way orgy" led by Knox that ended with Kercher's murder.

It is one thing to have fantasies and use them to create fiction for the screen or novels. It is quite another to use one's fantasies and botched DNA testing results to ruin lives. The acquittal of the murder trial, however, has since been overturned as Italy does not have a double indemnity clause as the United States does (one cannot be tried twice for the same crime). Consequently, Amanda Knox, is not attending the second trial and is not expected to be extradited whatever the outcome.

For one thing, recent DNA testing evidence shows that there was no blood of the victim on the knife. (Dempsey, "Good News for Amanda Knox: Victim's DNA Not Found on Knife"). Has anyone seen a horror movie like *Psycho* or any zombie movie where someone was killed with a knife, and they managed to *not* get blood on it? It would not have taken much DNA to be able to get a DNA sample of Meredith Kercher if it was used as the murder weapon. It was not in my opinion.

What was wrong with the DNA testing results? According to the Huffington Post article, "Amanda Knox Case: DNA Experts Cast Doubt on DNA Evidence," it was not up to international standards, according to DNA and forensic specialists. They used "a dirty glove and did not wear caps" and the bra clasp of Kercher that the prosecution insisted had Knox's DNA on it was not collected for 46 days. Even if Knox's DNA was on the bra clasp what does that prove? That she asked her girlfriend to help her fix her bra. Maybe bra-less men do not realize that women sometimes do that kind of thing? Also, the crime scene was not secured and handled properly. People moved around, and objects were moved ("Amanda Knox Case").

Also, most people do not realize that they already have someone in jail for this crime. Rudy Guede, by his own admission, decided to burglarize her place and then afterward "fled to Germany with his bloody shoes and pants." He is now in jail though soon to be released.

DNA testing is a wonderful tool. However, like any other tool, it must

be used correctly following strict guidelines and procedures, or it is useless if not damaging.

October 11, 2013

Preserving Your DNA

We might have gotten a laugh from the movie *Identity Thief*, but it isn't that funny if it happens to you. Living in a fast-paced world full of online spammers, credit-card skimmers and petty thieves, most of us know to be careful with our finances, Social Security number and credit cards. But what about a more personal identity? What about your DNA? Your DNA is unique to you alone. Even identical twins show slight genetic differences with very expensive genetic testing costing thousands of dollars (Barry Starr, "Genetic Sleuthing"). Your DNA is you.

Why should I control my genetic information? Because of epigenetics and other factors, it is doubtful that prejudice will ever be used from your DNA testing results as it is portrayed in the film *Gattaca*. Nevertheless, in the near future, if a thief has your genetic profile, that would be the ultimate in identity theft. Why? It could well be how we soon identify replacing the now state-of-the art idea of using your voice or fingerprint. Your genetic information is like owning a mirror-image of you.

How is genetic information used? Forensics already uses genetics to catch criminals, exonerate suspects or find missing persons. And DNA testing companies that analyze DNA with autosomal DNA testing (non-cousin related) can give you a comprehensive analysis of ancestry based on and reflecting your unique genetic information.

How can I control my genetic information? First, if you have done autosomal DNA testing and received your genetic profile with your unique alleles, you should put it in a secure place. Many have done DNA testing but not as many have thought too much about how their DNA is being used. In order to control our genetic information, we need to become as careful with it as we are with our credit cards. You might wish to think twice about using a DNA testing company that shares your genetic information with third parties or stores it in a large genomics database for medical research without the individual's express permission to do so. Even if you have read all the fine print or given your permission for them to do this, you might want to rethink that option.

DONALD N. YATES and TERESA A. YATES

Why? Not all DNA testing companies share your DNA testing results, or gather it in a large database, so why not choose one that does not and keeps it secure?

How can I save my genetic information and keep it the most secure? Your DNA dies with you. Not only should you save your unique genetic profile, you should save a healthy copy of your and your loved ones' genomes now to take advantages of advances in medicine and gene therapies in the future. And there are other reasons. If someone goes missing, this will greatly help an investigation. Just a handful of DNA testing companies offer a way for you to securely and affordably save your genetic information. Take advantage of one that does and be ahead of the crowd.

March 26, 2013

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