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Genetic Profiling of Predispositions and Behaviors

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Introduction

John Palmer sits in the waiting room of yet another telecom company as he waits for the Human Resources representative to meet him for his interview. Looking around at the room, he is overwhelmed by a feeling of déjà vu, as if he has seen this waiting room a thousand times. He suddenly becomes aware of his clammy palms and shaking legs. He knows what is going to happen today - the same thing that happened in the past twenty interviews. John will be told that he is not a match for the job as the Human Resources Representative looks uneasily at the report she has just pulled up online. He doesn't have to ask what it is; he knows. His genetic code had been sampled as a baby and has been haunting him ever since. Based on his genetic code, John has a genetic predisposition for sexual offenses. Although he would never hurt a fly, he often thinks that he might as well become what the technology says he is: a sexual predator. The despair from another failed interview settles in as he thanks the representative and returns to his car.

Louise Thompson sits at home on hold for what seems like the thousandth time. One health insurance company after another has given her the same politically correct response - she and her daughter, Annie, are too high risk to insure. Although she is a healthy 34-year-old non-smoker and her daughter is a bright and happy four-year-old, premiums offered are more than she can afford, if she is lucky enough to get offered anything. According to the database of genetic code, Louise has a predisposition to develop breast cancer and Annie is predisposed to Type I Diabetes. These two predispositions have left Louise with no way to get affordable health insurance for her or her young daughter.

Imagine a society where genetic identification and information is taken at childbirth and stored in a database, following you around for the rest of your life. Despite what many may think, we are not that far from this type of society. The U.K. has begun a process they call "bar-coding babies" where a unique electronic bar code is linked to health information taken from them at birth. Currently, this does not contain genetic code, but there is a push from certain areas of society to include a genetic sample with each record.

Different segments of society use genetic profiling in different ways. Objectively, categorizing people into different groups is neither good nor bad. But, when taken together with a person's personal agenda, it could mean exclusion or bias towards certain groups or specific individuals. This has become such a concern in recent years that on February 8, 2000, former President Bill Clinton signed an executive order to prevent the use of a person's genetic profile to determine if that person is fit for employment. Clinton stated:

By signing this executive order, my goal is to set an example and pose a challenge for every employer in America, because I believe no employer should ever review your genetic records along with your resume (Clinton 2000).

Although this law would protect U.S. citizens from overt discrimination by employers, there is currently no federal law that protects an individual against discrimination by an insurance company who happens to gain access to an individual's genetic profile. This could result in higher premiums or prevent individuals from receiving health insurance at all. Also, discrimination is a difficult charge to prove in such cases. If an employer had access to an individual's genetic profile, they may find alternative reasons not to hire that person, thus escaping the bias charge but still discriminating nonetheless.

Other areas of society could use genetic profiling to discriminate. Law enforcement agencies such as the police, FBI or CIA could compile lists of people who are predisposed to violence or anti-social behavior. During the investigation of a crime, police might focus on this group of people rather than the population as a whole. Also, police with only circumstantial evidence could look more towards an individual's predispositions rather than their actual motives.

Still, there are very powerful arguments for using genetic profiling. Currently, scientists affiliated with the American Diabetes Association are trying to perfect a vaccine that could be given to children with a predisposition to Type I Diabetes in hopes of preventing them from actually developing the disease. In addition, parents are using genetic tests to determine the risk of their future children developing certain diseases. Based on the genetic codes of the parents, doctors can ascribe probabilities that their children will inherit certain traits or will develop certain diseases.

Ethical concerns arise not with the testing itself, but with the use of the results. This creates the need for society to develop strict guidelines that define appropriate usage and control of the databases that house the results. Agencies possessing databases containing genetic profiles strictly for scientific research should not be permitted to share

information with law enforcement or insurance agency databases. If this were to occur, a person's genetic profile could be used in a manner that would invade his or her privacy. For example, if insurance companies can use research information to delve into a person's past, they could discover information that can be used to deny coverage.

The regulation of data sharing could mean the difference between the benevolent use of genetic code and a society in which personal information is sold to the highest bidder. Society must work to prevent the unethical use of this information. If we do not, we could find ourselves in a society where people are judged not by the content of their character, but by the content of their genetic code.

Genetic Code

In April 2003, scientists were able to sequence the entire genetic code of a human being (Department of Science 2003). Using the map created by these scientists, it is now possible to find behaviors and predispositions that are directly linked to genetic code. Scientists are able to find and attribute anomalies in the DNA sequence, to a disease or behavioral disposition. Once genetic profiles become more widespread and collection of data is integrated across borders, issues of privacy will surface in several areas of society.

How Is It Done?

By using large families of 200 members or more, scientists are able to determine the causes of common genetic traits. Family members are the best sample set because they often share common genetic inheritances. Because of this, it is easier to isolate the genes that cause a disease with a great deal of certainty.

Another effective method of determining who shows predisposition to certain diseases is with genetic isolates, which come from areas with limited founder populations. This occurs when small groups of people migrate to a small geographic area and have remained there ever since with little migration after the initial wave. At present time, these populations are still all decedents from the founder population, and have inter-married to an extent where all members of the society are related. This creates a perfect environment and an invaluable resource for genetic testing. Because migration throughout the ages has been so prevalent and people rarely live in the same place as their ancestors did 1,000 years ago, there are very few genetic isolates left with large enough populations to study.

How Close Are We?

How close are we to a society where everyone has his or her genetic code on file? Newfoundland is a country with a strong genetic isolate (NewFound Genomics 2004). NewFound Genomics, a genetic profiling company based in Newfoundland, has begun studies based on the genetic profiles of volunteers (Taubes 2001). NewFound Genomics recruits native Newfoundlanders to volunteer to donate a genetic sample. These samples are kept based on the specific study for which the donor provided the sample and may not be used for any other purpose. NewFound Genomics must take Canadian policies including Personal Information Protection and Electronic Documents Act

(PIPEDA) and the Canadian Tri Council policy statement on Ethical Conduct for Research Involving Humans into account (NewFound Genomics 2004). These policies are designed to protect the privacy of those who donate genetic samples, including histories of donations, data kept in the study-specific databases, as well as lists of donors.

In Sweden's Västerbotten County, one of the last regions in Europe to be settled, genetic studies are taking place based on the genetic isolate present in the homogenous population. Västerbotten County experiences strong founder effects and is thus a prime location for genetic studies. The Västerbotten Intervention Program, or VIP, established in 1985 has been used to take blood samples from over 70,000 individuals. Based on voluntary participation, UmanGenomics invites all citizens as they turn 40, 50 and 60 years old to participate. Participants give UmanGenomics exclusive rights to the information including any commercialization planned by the company (UmanGenomics 2004).

Based on Swedish law, however, UmanGenomics must follow certain guidelines. All participants are informed about the uses of the samples and must give informed consent, meaning they sign a waiver saying they understand their rights and responsibilities. Participants who wish to withdraw consent can do so at any time and must only inform UmanGenomics of their desire. UmanGenomics must then either return or destroy the sample. The Swedish Medical Research Council and the National Board of Health and Welfare have established guidelines for uses of samples and all uses must be approved by the regional ethics committee.

The UK Biobank plans to start recruitment of volunteers for a genetic study in 2005. They estimate a turnout of 500,000 volunteers between 45 and 69 years-old (UK Biobank 2004). All volunteers will need to give written consent, fill out a questionnaire, and visit the clinic where their weight, blood pressure, and genetic sample will be taken. The questionnaire will contain information about exercise patterns, diet, and employment, medical and family history. The most invasive part of this study is that all data will be tracked against the individual's health record contained within the United Kingdom's socialized medicine system, the National Healthcare System, or NHS.

A primary concern among participants is access to this information. The Biobank has set up regulations concerning this, but there is no guarantee these regulations will last forever. The current policy does not allow insurance companies or employers access to individual or anonymous data. However, pharmaceutical companies are allowed access to anonymous information under "strict controls." The Biobank's policy contains a loophole. Police are allowed access to any and all information under court order. Once this information is brought into the public light due to a court order, it is now public information. Employers and insurance companies will no longer have to look to the Biobank for an individual's genetic code, but instead, they will look to the local police department.

Even the Kingdom of Tonga, a small country in Africa, has gotten in on the genetic profiling trend. The Tonganese government decided to pass legislation to collect the genetic profiles of each of Tonga's 108,000 citizens. Participation in the program is strongly encouraged,

but optional. The government sold this data to Australian owned AutoGen in hopes of finding cures for diseases plaguing the Tonganese people, including cardiovascular disease, hypertension, certain cancers, and stomach ulcers (Griggs 2000). One might wonder why the government would sell this data. Here's the catch: all drugs developed by AutoGen as a result of this study must be given to Tonga for free (RAPS 2004).

Perhaps the most well-known and most controversial genetic study is that of Iceland by deCode Genetics. Founded by an American citizen and Harvard graduate, deCode Genetics is incorporated in the United States and backed by American investors. Its success is based on the genetic profiles of Iceland's citizens. Passed into law by the Icelandic parliament in December 1998, all of Iceland's 1.4 million citizens must give a genetic sample to deCode Genetics. As a genetic isolate descended from the Vikings, Iceland is an inbred population. Patriotism and heritage pride runs deep in Iceland and can be seen through the excellent medical and family tree records kept throughout the country. This creates the prime environment for a wide scale study containing more participants than any other study done thus far.

Although deCode Genetics has thus far been fairly successful, 25% of Icelanders object to this program. The law was passed into effect based on "presumed consent" rather than "informed consent" (Taubes 2001). Informed consent involves signing a waiver agreeing to give away your information and that you understand your rights. With presumed consent, one must submit a written "opt out" form to the proper authorities. Without the "opt out," deCode Genetics has the right to all records.

Within Iceland, there is much controversy and criticism over this study. Many advocates of the study claim it will be able to help the Icelandic people overcome diseases that have plagued them for centuries while some say this is misleading in that the chances of curing these diseases is so low, it is not greatly increased by this study. It has also been claimed that the study plays on the deeply rooted patriotism and national self-interest that Icelanders feel.

Why Should Society Be Concerned?

There are three main areas in which genetic profiling can be used in the future: law enforcement, the workplace and especially healthcare. It is important to understand the implications of the uses of data in these areas. Guilt of a crime, workplace hires, and insurance premiums may no longer be judged on the conventional methods we use today, but rather on the genetic code.

Law Enforcement. Throughout the police force, predispositions to violent behavior, sexual deviance, or anti-social behavior could be used in court to prove propensity where motive is lacking. For example, if two people are found at the scene of a violent crime, and neither have motive, the police are more likely to pursue the suspect with a propensity to violent behavior even if they have never committed a violent crime. Also, a jury is more likely to put this perceived violent individual behind bars because their genetic profile may eradicate the jurors' last reasonable doubt.

Workplace: Workplace discrimination is now, more than ever, a hot topic. Employers want to hire those who will be able to perform their job with minimal assistance and time off. Individuals needing extra time off or expensive medical care may be denied a job even before these needs arise. Historically, workplace discrimination charges are difficult to prove, especially when in an “at will” state where either the employer or the employee can end the employment agreement at any time. In these states, a prospective employee can be turned away for any or no reason. Protection is available through the Equal Employment Opportunity Commission (EEOC). However one must have enough evidence for the EEOC to pursue an individual’s case.

Healthcare. Healthcare issues are becoming more and more relevant in today’s society. Recently, geneticists have been conducting studies to predict the chances of a person developing a certain disease. In terms of scientific advances, this is an incredibly powerful tool for the preemptive treatment of diseases. However, privacy issues are already surfacing in the use of this information.

Finding predispositions for diseases, such as breast cancer, can be of great benefit to those with such propensities. Of those diagnosed with breast cancer, many women die because it was not detected early enough. With genetic profiling, doctors will be able to determine at birth whether or not a person is susceptible to developing breast cancer, and will most likely be able to detect the disease earlier with careful testing. Many women have selected voluntary mastectomies prior to any sign of cancer because their genetic code contains the BRAC I or BRAC II gene defect. While this may seem drastic, women are making this choice because it virtually guarantees they will not die from breast cancer.

Another area of healthcare that has advanced with the advent of genetic testing is the ability to tailor disease treatment depending on a person’s genetic make-up. This is useful because certain people react differently to certain drugs. For example, some cancer patients are more receptive to chemotherapy whereas others are more receptive to radiation. The use of genetic profiles will assist doctors in tailoring treatments to the individuals’ genetic make-up. Scientists believe that some day it may be practical to create a custom drug to treat an individual patient.

Although genetic profiling in healthcare has the aforementioned advantages, there are also several downsides. Information on a person’s genetic predisposition could be used by insurance companies to deny coverage. Even though a person may be more susceptible to develop a disease, they may not ever get it. In the mind of the insurance company, however, a higher risk means a higher premium, thus this person would pay for a lifetime of treatment for a disease they may never get.

This is not just a Hollywood portrayal of a future problem. This issue is surfacing across the globe with genetic studies happening in the most unexpected places. To help solve this problem of discrimination in healthcare, insurance companies in the United Kingdom, London issued a five-year moratorium in October 2001 on using genetic profiling. This means that all insurance companies in London have agreed not to use genetic profiling for any decision making with regards

to denial of insurance or increase of insurance premiums. This moratorium will end in October 2006 leaving people in the United Kingdom with the same dilemma with which they began.

Of course, the question needs to be asked; "Is it ethical to gamble on a person's health in the first place?" Perhaps, the problem lies in the fact that insurance companies need to make a profit to stay in business. While socialized medicine has its own problems, if implemented fairly, it is honest in its intent. From this perspective, genetic profiling for medical purposes in a socialized medical environment would have positive benefits greatly exceeding the negative.

Sandra's Story

A Coloradoan middle-aged mother of two, Sandra Pinkensen, had undergone a DNA test when her doctor detected breast cancer. Her insurance company was ordered to pay for the test because the cancer had already been detected, thus it could not be classified as preventative care and denied. Based on Colorado law, the insurance company was not allowed to see the results showing the BRAC II defect known to cause breast cancer. Sandra knew this defect was genetic and her blood relatives would most likely have it, including her mother and ten-year-old daughter. She felt scared and alone, thinking no one would understand, so she didn't share this information with anyone other than her doctor and genetic counselor.

Her genetic counselor suggested she voluntarily have her ovaries removed because the BRAC II defect greatly increases the chance of ovarian cancer. After pondering her options, Sandra decided to go ahead with the procedure. The insurance company has no way of knowing that Sandra has the BRAC II defect and the only way to get coverage for the procedure is to tell them that, allowing them to use Sandra's genetic profile in a risk assessment. Her doctor contrived an excuse for the procedure to avoid telling the insurance company that she had the defect. Is it ethical for the doctor to lie to the insurance company solely due to fear of future profiling? Is it ethical for Sandra to withhold the information from the rest of her blood relatives who have a high likelihood of the defect? If the insurance company were able to gain Sandra's profile, would it be ethical to use it in a risk assessment? These are all issues that could become prevalent in our society based on genetic profiling.

How Can We Protect Ourselves?

While most or all of the chapters in this book discuss how technology can invade our privacy, this chapter will discuss how technology could protect information, specifically information concerning genetic predispositions.

When asking a technical consultant how this could be done, his response was that to truly protect the data, one could "unplug the computer from the network/internet, make sure there are no removable disk drives, CD burners or the like in it, put it in a closet, lock the door, and post guards 24/7" (Simmons 2004). While this would work, it is certainly not practical in the business world. Data needs to be accessed by more than one or two people. It may even need to be sent from one physical location to another.

The first consideration that must be made is where the data is housed within a company. Although the safest place for the data to be is on a computer that is not connected to anything else, this would mean that only a handful of people could access it. Precautions should be taken though in regards to where on a company's network the data is stored. Data should not be stored on a computer that has direct access to the Internet. By doing this, it makes it more difficult for someone who has broken into a network to locate the file (Monarchi 2004).

The next consideration is the type of encryption used on the data. Thousands of years ago, people were already using encryption to prevent people from reading messages en route to their destinations. One method was to shave a messenger's head, write the message there and then wait until his hair grew back to send him along to the recipient (Monarchi 2004). This was a time consuming method, which could only be used for non-urgent messages. Another encryption method was used by the Roman Emperor, Julius Caesar. He used a simple letter replacement method where he would substitute a letter for one three places further along in the alphabet. Thus the letter 'A' would be replaced by the letter 'D', the letter 'B' by the letter 'E' and so on. This method of letter replacement is known today as the "Caesar Cipher". Today, this method would be far too easy to decrypt, especially with computer tools. There are many infinitely more sophisticated methods of encryption available for protecting computer data.

Data can be seen in two different ways, stored data or "data at rest" and data that is being transferred or "data in motion". In looking at the first type, stored data, we can encrypt the data at two different levels. The first is at the file level where the entire database file is encrypted. While this is simple and easy to do, there are a few problems with this method. One of them is that by encrypting the entire file there is a large amount of overhead both when the file is encrypted and each time it is decrypted for use. Another problem is that only one key can be used to encrypt the data thus making it easier to break into (Application Security, Inc. 2003).

A second option for encryption of stored data is to encrypt different sections of the data. In a file on genetic predispositions, it may be practical to encrypt only the sensitive fields of data (e.g. a company could encrypt rows of data that contain the types of tests a person has taken and the results, but not encrypt the name field). This gives a company the possibility of encrypting data using several different encryption keys and means there is less of a chance that an entire database can be compromised (Application Security, Inc. 2003).

Data that is being transmitted is much more vulnerable than data stored behind a company firewall on a secured computer. Because of this, it is important to encrypt data using a strong encryption method. One of the most common protocols is SSL (Secure Sockets Layer). This method, developed by Netscape, uses a private key to encrypt data that is sent over the Internet. While this method is the most popular by far, it is the least secure of the protocols mentioned in this paper (Application Security, Inc. 2003).

A second protocol is TLS (Transport Layer Security). This is made up of two layers, which ensures that the connection is private and

reliable and also authenticates between the client and server (Webopedia 2004).

A third method for sending and accessing data is by the use of a VPN (Virtual Private Network). One way to implement this is by using IPSec (IP Security) which uses a method called 'tunneling' which creates a virtual secure connection much as if the sending and receiving computer were connected by a single private wire. This would be one of the best methods for companies sharing data either within company offices at different locations or between different companies.

What Does This Mean for the Future?

The preceding shows that there are many methods to encrypt and protect data and databases containing genetic profiling information. However, it should be noted that the reason these protections need to be put in place has nothing to do with the advancement of genetic testing. Technology is neither good nor evil, rather is it what people do with it that defines how it is viewed by society. What this points to is a need for legislation defining how genetic testing can be used and by whom. It is unfair to give a person a choice between protecting their health and ensuring that they can afford insurance coverage as was seen in the case of Louise Thompson and her daughter.

The National Institute of Health (NIH) has a long-term open grant with the goal of getting technology to the point that it would be possible to have a person's entire genome sequenced for only \$1,000. The NIH will give money to anyone who has a convincing proposal for this undertaking. Most people think that this is at least ten years off, but many companies have taken up the challenge to work on selected parts of this problem. Mapping the human genome was achieved in the same manner. Because many companies were competing to be the first to do this, the estimated date for completion was shortened by years. The NIH has given out many grants for research efforts that promise to help achieve this goal. Since genetic testing is practically in its infancy, there will be much trial and error with legal issues and challenges before laws are put in place to protect the privacy of people who want to know more about themselves in an effort to live healthier lives. The combination of new technology, the right to privacy and laws to protect individuals can mean that someday soon, it may be practical for every individual to own their entire genetic code.

Works Cited

- Application Security, Inc. 2003. *Encryption of Data at Rest*. Forbes IT Research Library.
- Clinton, Bill. Speech: "Remarks by the President on Genetic Discrimination." The White House. Washington D.C. 8 Feb 2000.
- Department of Science. Speech: "International Consortium Completes Human Genome Project." Bethesda, MD. 14 Apr 2003.
- Griggs, Kim. 2000. *Wired News: Tonga Sells its Old, New Genes*.
- Monarchi, David. Professor in Information Systems Division, University of Colorado at Boulder. Personal Interview. 20 Sept 2004 and 26 Sept 2004.
- Newfound Genomics. 2004. *Newfound Genomics Website* 16 Sept 2004 <<http://www.newfound-genomics.com>>

Regulatory Affairs Professionals Society (RAPS). Nov 2004. Australian Company to Build Genetic Database of Tonga. 16 Sept 2004 <http://www.raps.org/s_raps/sec_RANews_Detail.asp?TRACKID=&CID=116&DID=20527>

Simmons, Jeremy. Technical Consultant. Personal Interview. 20 Sept 2004.

Taubes, Gary. April 2001. Technology Review. *Your Genetic Destiny for Sale*.

UK Biobank. 20 Oct 2004 UK BioBank Frequently Asked Questions 16 Sept 2004 <<http://www.ukbiobank.ac.uk/FAQs.htm>>

UmanGenomics 2004 UmanGenomics.Website. 16 Sept 2004 <http://www.umangenomics.com/en_index2.htm>

"What is TLS?" 2004. Webopedia Website. 18 Sept 2004 <<http://www.webopedia.com>>

Application Security, Inc. 2003. *Encryption of Data at Rest*. Forbes IT Research Library.