Personal Genomics and Social Sciences and Humanities

Well, from the universe of the very big (climate) to the universe of the very small (the human genome)! Once again, I wanted to start off the discussion with a reminder of the basic philosophy of the course. I stressed that personal genomics is not just a physical and life sciences issue: it is a political, economic, and social issue. It is also another example of the relationship (and sometimes the clash) between a concept grounded in science, and the social context in which policy decisions are made and implemented. My bottom line: if we are to understand personal genomics, we must have a basic understanding of both the physical and life sciences aspects and the social sciences and humanities aspects of the subject.

The review article by Peter Hatemi and Rose McDermott is a crucial starting point for us, because it summarizes the current state of research and dialogue on the nexus between genetics and human political behavior. This is at the very core of one of the most fundamental debates in the social sciences and humanities: the (in)famous nature/nurture debate. While there is widespread acknowledgement that behavior is influenced by both biological and environmental factors, the larger social consensus of our time is that human behavior is mostly the product of environmental influences: family, schooling, community, society, culture. The idea that this might not be entirely true, or as true as we have thought, comes as something of a shock or even an affront. The idea that genetics influence human political behavior is usually greeted with incredulity. Of course, the influence of genetics may be less than what some studies suggest (read the article), but the point is, to most of us, this is a scary, disturbing, or uneasy thought.

So here was an opportunity to return to the need for awareness about how we are all embedded in a larger social discourse on the subject of genetics. This has manifested itself before in our social history in a variety of forms, from *Frankenstein* to *A Brave New World* to *Jurassic Park*. We have been very uneasy about the biological sciences and what "monsters" may be lurking in that science. I returned to the "zeitgeist" theme to illustrate this point: we are not examining any of these issues in some kind of objective vacuum. One environmental impact

influencing how we see human genomics is the discourse in our own history and in contemporary society. Or is it the case that the very way you are approaching this subject, and receiving this idea, is influenced in some way by your genetics? Hmmm.

Now I am being deliberately provocative. Most studies of the relationship between genetics and human behavior are self aware of the limitations of the studies, the vast amount of work yet to be done, and the challenges of drawing this linkage (more on this below). Nevertheless, it is becoming increasingly clear that to some extent, exploring the question of the origins of human behaviour will require an understanding of genetic influences, and what was once a largely exclusive reserve of the social sciences and humanities will become a shared research project of both the life sciences and the social sciences and humanities.

The second article by Jaime S. King highlights another aspect of the relationship between human genomics and society: advances in genetics impact some of the most volatile and contentious debates in societies around the world. Abortion is just one of them, and this article reflects on advances in non-invasive prenatal genetic testing (NIPT), not only for gender and (for example) Down's Syndrome, but for a wide variety of genetic traits in the future. This raises a number of questions about rights, choice, prejudice, and law.

This illustrates how the personal genomics issue is embedded in a larger set of issues related to microbiology/biotechnology and society. A few examples of the relevant interfaces between the sciences and the arts on the genetics front include:

- Medicine (genetic testing, gene therapy, cloning, pharmaceuticals)
- Health care (practices, policies)
- Industry (chemicals, energy)
- Agriculture (GM crops/food)
- Legal and regulatory systems (court decisions, government legislation)
- Domestic politics (rights, minority groups)
- International/Global contexts

A word on how we got here

Personal genomics has a history embedded in the study of genetics, but our story begins with the sequencing of the human genome. This achievement was announced on June 26, 2000, after an effort that took 13 years at an estimated cost of 2.7 to 3 billion dollars. The scientists involved (J. Craig Venter and Francis Collins) won a prize and a visit to the White House to shake the hand of the then US President Bill Clinton. This event ushered in a new era in the biological sciences: the Economist magazine suggested that the history of biology could now be divided into the pre- and post-genomic periods. Accurate or not, what was interesting was how President Clinton felt it necessary to refer to the "language in which God created life" in his speech.

But as always, there was an interesting backstory behind the announcement. First of all, the announcement was premature: a truly complete sequencing of the genome was not accomplished until 2003.

Second, the attempt to sequence the genome had been a competitive one. One competitor was a public consortium, which included the International Human Genome Sequencing Consortium under Dr. Francis Collins, and the Whitehead Institute under the leadership of Dr. Eric Lander (other public partners included the National Institute of Health and the Sanger Institute). The other competitor was a private venture, established under Dr. J. Craig Venter and his company, Celera, with the intention of beating the public project to sequence the human genome first.

Third, there was a philosophical division among the scientists pursuing the human genome: some energetically pursued patents on various gene sequences (Venter), with a view to the commercial potential of the patents, while others saw gene sequences as public goods (Collins). A race developed between some scientists to discover genes first, in order to patent them or release them into the public domain.

Fourth, the effort to sequence the human genome was accompanied by extraordinary predictions about the promise for a new generation of treatments and drugs to address disease. More than ten years later, relatively few treatments have emerged, as it has proved much more

challenging than previously thought to link genes to diseases. That does not mean such benefits will not happen, it just means it is taking a lot longer than initially thought, and raises a caution flag with respect to any predictions made about the future impact of current or unforeseen scientific advances.

And finally, all through this process, there was the beginning of a wide-ranging discussion and debate about the implications of human genome sequencing. The extraordinary medical, pharmaceutical, and human health potential was very exciting, because wide availability of sequencing capacity promises a wider range of research efforts and potential products of significant benefit. Human genome sequences can be compared with each other and with those of other species to reveal gene function and improve understanding of species evolution, genome functions, and the development of disease. Pharmacogenomics promises the ability to match drugs to a patient's genome, and avoid adverse drug reactions. Our understanding of human life and our relationship to the natural world, of human origins and evolution, and how to treat disease, will increase dramatically.

This potential was deliberated alongside concerns about abuse of the technology, public education and awareness, the role government regulation, individual rights and privacy, reproductive ethics, patient and physician relations, among many others. Darker concerns, including the potential for social engineering, genetic discrimination, racial and social underclass marginalization, and an unequal distribution of benefits globally, hovered around the periphery of the dialogue.

An important caveat must be made at this point. The debate about human and personal genomics is very new, and much of the discussion is in its infancy. Currently, dialogue and debate is largely conducted within the scientific, legal, and academic community while news stories tend to narrate the rapid progression of the technology. Wider discussion of personalized genomics at the governmental level has barely begun. Domestic law and regulation (or public policy, if you prefer) on the subject is only just taking shape. Global regulation in the form of international treaties is developing at a slow pace, far slower than scientific discoveries. So we are heading into largely charted waters on this subject.

Personal Genomics and Human Society

1. Analytical and Policy Complexity

We know from Dave that the science behind genomics in general and human genomics in particular is complex. The social aspects of human and personal genomics are at least as complex. One again, the "levels of analysis" tool really helps us grasp this: human genomics is another classic example of the importance of being aware of how a large number of variables across individuals, groups and states, and the international system all play a role in how we understand the implications of the sequencing of the human genome and how we might respond to these challenges. To review, there are a lot of variables:

> Individual: education; life experiences; spiritual beliefs; ethical views; standards of living; personal health; medical histories; personal aspirations
>
> The state: governance (political systems); leadership; law and regulation; economic priorities; public opinion; role of interest groups; marginalized groups
>
> International: interests of states; governance and institutions; political will; negotiations; norms; bargaining; trust

The human genome project will continue to engage a large number of variables and so we need to understand them as best we can. These are the subjects of the social sciences and humanities, and we cannot understand the impact of the sequencing of the human genome without them.

2. The genie is out of the bottle

Human genome sequencing has never been in a publicly funded "bottle" of course, given that many sequencing technologies and techniques were developed in privately funded laboratories. But the extent to which human genome sequencing is already available to wider society is remarkable. Accessibility is increasing as costs fall, and this has raised

some concerns about how private firms have moved ahead of the ethical, legal, and social debates about how the technology should be used, managed or regulated.

For example, privacy of information is a major consideration. In an article in Science, a researcher recounted how he could take the anonymously donated genome from an open-source site like 1000 Genomes Project, identify an inherited pattern (markers on the Y chromosome called Y-STR markers) in the genetic sequence of that genome, take the age and region of the donor which provided on the website, match the pattern and age and location to the patterns and name information available on genealogy websites (which use the same genetic pattern along with surname to identify heredity for their customers), match individuals based on age and names, and then Googled their names and their relatives names to discover information about them, including where they lived, where they worked, etc.

Another example is direct-to-consumer testing. As a result of falling costs, the availability of full human genome sequencing has increased, but so have services like ancestry tracing and “predictive medicine” (a list of genetic variations that may put you at risk for certain diseases). This genetic data and ancestry tracing is accomplished through spit or blood samples, and a search for a certain number of genetic traits in each sample. Costs vary by service. These companies do not require high levels of consumer knowledge and awareness. When such tests are used for medical screenings, misinterpretations of test results can result in unnecessary stress and lead to misinformed decision-making.

Debate has also arisen over fetal testing and the impact this might have on familial relations and public policy. Will spousal genomic testing impact decisions on procreation? Would prenatal testing impact decisions on procreation? Illumina's CEO, Jay Flatley, stated in February 2009 that "A complete DNA read-out for every newborn will be technically feasible and affordable in less than five years, promising a revolution in healthcare" and that "by 2019 it will have become routine to map infants' genes when they are born."

So the genie is out of the bottle. And the question is no longer whether it can be put back, but rather what kind of regulations should be put in

place to manage or govern the activities of the private sector, which could have profound implications for individuals and society.

Currently, regulations in the US are mostly at the state level: 25 states permit direct-to-consumer genetic tests without restriction. The most common restrictions are the need for a permit (a state or federal clinical lab certification) and authorization from a physician. Many companies have been issued "cease and desist" orders based on a failure to comply. However, many companies argue their services are educational, not diagnostic, thus avoiding the more stringent regulations associated with diagnostic services.

There is no regulation of direct-to-consumer genetic testing in Canada under the Health Act (and very little anywhere else, for that matter). Studies of the habits of direct-to-consumer service providers suggest they fall well short of the recommendations of the Canadian College for Medical Geneticists, established in 2011.

Ethical Issue:

- Fairness in the use of genetic information by insurers, employers, courts, schools, universities, adoption agencies, and the military, among others.
    - *Who should have access to personal genetic information, and how will it be used?*

Ethical Issue:

- Reproductive issues include adequate informed consent for complex and potentially controversial procedures, use of genetic information in reproductive decision-making, and reproductive rights.
    - *Do healthcare personnel properly counsel parents about the risks and limitations of genetic technology?*
    - *What are the larger societal issues raised by new reproductive technologies?*

And that was it for this week!