

By Kim Ribbink



## Trend:

# Genetic Testing and Data Privacy and Ownership

**G**enetic testing is a booming market. According to reports, as of April 2018 more than 15 million people had taken direct-to-consumer genetic tests, and with prices for autosomal DNA tests now costing less than \$100, they have become very accessible.

Companies such as 23andMe, Ancestry.com, and MyHeritage are making it easy for consumers to test their DNA using a simple cheek swab.

With a growing number of applications for genetic tests for early detection and prevention of oncology and genetic diseases, the value of this market is projected to exceed \$22 billion by 2024.

Indeed, there are suggestions that the human data marketplace represents an estimated \$150 billion to \$200 billion annually.

With the growth of the consumer testing market, the opportunity to draw insights from this data for research into potential new treatments for numerous diseases is compelling.

In July 2018, GlaxoSmithKline and 23andMe unveiled an exclusive agreement to focus on R&D into innovative new medicines and potential cures, using human genetics as

the basis for discovery. GSK is investing \$300 million in 23andMe with the objective of mining genetic information in the company's database. According to 23andMe, the information shared is aggregated data and protects privacy.

However, doubts remain. David Forster, chief compliance officer, WIRB-Copernicus Group, says it is very important that patients are informed about all of the intended uses of their data, including if the data might be shared with other parties.

"For instance, if the provider of the DTC genetic testing plans to share that data with pharmaceutical companies, patients should be informed and have a right to decline such sharing," he says.

Jennifer Miller, Ph.D., founder of Bioethics International, agrees, saying there is a lot of money being made around data and at a minimum, patients need to be informed where their data are going.

### Taking a Cautious Approach

Despite the potential to use data to advance drug development — and even to trace killers, another way the data are being used —

With data from genetic tests being tapped by companies for R&D purposes, questions about privacy and data rights are being raised, and steps are being taken to protect people's data.

there are a number of concerns when it comes to genetic testing and how the data are used.

One concern raised by Dr. Miller is the limitations of data.

"Currently, 80% of genomic data are from Caucasian so if genome data are going to be used to develop new drugs, biologics, or any kind of medical intervention, it's important to know using these data may not yield a generalizable result," Dr. Miller notes. "There have been cases where we've developed therapies that don't work for everybody and it's taken us time to realize that."

She notes the example of Type 2 diabetes blood tests, which turned out didn't work for African-Americans. As a result, an estimated 650,000 African-Americans may have had undiagnosed diabetes because common blood tests didn't work for them.

"We don't want something similar to happen by relying on genomic data that are 80% Caucasian," Dr. Miller warns. "All scientists need to be aware of the limitations of their data and be very transparent about them."

She cites the Food and Drug Administration's Drug Trial Snapshots, which highlight who participated in trials and whether there



It is very important that patients are informed about all of the intended uses of their data, including if the data might be shared with other parties.

**DAVID FORSTER**  
WIRB-Copernicus Group

were differences among sex, race, and age groups. Dr. Miller says the same approach should be used with genomic data where it's made clear what the demographic profiles look like.

Patient privacy and the rights of the individual to control their data are also key. Obtaining informed consent is essential with genetic testing of any type, and especially DTC testing, Mr. Forster says. "Patients should clearly understand the risks and benefits of the testing, the confidentiality provisions, the expected accuracy of the genetic results, any planned uses for the data, and what results will be shared with them," Mr. Forster says. "The stated risks should include the possible effects of the data on future insurance and healthcare, and the reliability and meaning of the data."

Dr. Miller says patients and consumers very often don't understand the value of what they are providing and how their data will be used.

"I'm fairly well-educated, I read the websites of companies, and I find it hard to understand what the process of opting out is," she says. "Can I opt out after I provided my sample? Do I have the right to be forgotten? What if I decide I don't want my information out there, can I pull it back? It's not clear to me whether someone can opt out if data have been de-identified and aggregated with other peoples' information. There tends to be more control around data that hasn't been de-identified. Once it's stripped of certain identifiers that are defined under HIPAA, it's no longer considered your data and it doesn't have a lot of protections around it."

Confidentiality and privacy present significant ethical issues as well, experts say.

"Genetic information can be important to healthcare decision making, life planning, and reveal family relations such as misattributed paternity and adoptions," Mr. Forster says. "It is therefore essential that providers of DTC genetic testing have strong security protections in place and inform patients of potential privacy issues that might arise."

Efforts are in place to give individuals more control over their data. Sir Tim Berners-Lee, the inventor of the world wide web, is building a start-up focused on helping people control their own data. The company, Inrupt, has developed a platform called Solid, which gives users control over where their data are stored. All data are stored on a Solid personal online data (POD) and each individual gives people and apps permission to read or write to their Solid POD. This allows both for ease of use, since data saved on one app are available on another, and protects the user's privacy.

Hu-manity.co, a data ownership start-up established by Richie Etwaru, is also focused on enabling citizens to restrict how their data are used. The company is using blockchain technology — called Human Data Consent and Authorization Blockchain — to enable people to negotiate consent and authorization terms with organizations so that their inherent human data can be respected as legal property.

There are also concerns about how sharing genetic data might affect the privacy of other citizens, and Dr. Miller notes that de-identification is not 100% foolproof.

"You can often re-identify somebody by triangulating information, particularly with social media information," she says.

According to a study published in the journal *Science*, if as few as 3 million people in the United States upload their genomes to public genealogy websites, around 60% of Americans of European descent — largely because most clients of consumer DNA companies are Caucasian — would be identifiable by their DNA and just a few additional clues. So far, more than 1 million people in the United States have published their genetic information.

### The Danger of Poor Data Understanding

Dr. Miller says at this point patients or users should treat DNA testing kits as entertainment or interesting information, including their interest in knowing more about themselves, their families, and where they come from, and not as medical diagnostic tools.

According to Karmen Trzupsek, director, clinical trial services, and director, ocular and rare disease genetics services at InformedDNA, while the rise of DTC genetic testing has, in some ways, helped the average American become more familiar and comfortable with the use of genetics, in some ways, it has also made Americans warier and more confused.

### Protecting Your Data

Research from Hu-manity.co estimates the size of the global human data marketplace to be between \$150 billion and \$200 billion annually.

In terms of privacy, some progress is being made. In the European Union, the General Data Protection Regulation 2016/679 addresses data protection and privacy for all individuals within the European Union and the European Economic Area. However, while the regulation speaks to how and for what purpose your data may be collected and traded, GDPR provides no means for you to negotiate how the data is used or any right to that data.

Without classifying this data as human property, companies can still mine it for free and sell it to anyone they wish.

Hu-manity.co allows you to claim your right to declare your inherent human data as property on an app and record your consent to a blockchain. The blockchain provides transparency to all participants in a marketplace for how you will allow your property to be used and under what conditions, including the ability to be compensated for its use.

They invite you to sign up for the #My31 App. It's an effective way to repeal the law of the jungle and protect the privacy of your data as your property.

Source: Derek Hasen, Chief Research Officer, and Michael DePalma, Co-Founder and President, Hu-Manity.co

"Despite the leaps and bounds that have been made in our understanding of the human genome, patients often have a limited understanding of basic genetic/genomic concepts," Ms. Trzupsek says. "This can complicate how genetic results are delivered, as patients may interpret DTC test results incorrectly."

Mr. Forster adds that a key factor in DTC genetic testing is the validity, accuracy, and clinical actionability of the results that will be returned to the patients.

"If the results are validated, accurate, and clinically actionable, there is more justification in returning them to the patients," he says. "If they are not, the justification weakens, particularly if the results are returned directly



While the rise of DTC genetic testing has, in some ways, helped the average American become more familiar and comfortable with the use of genetics, in some ways, it has also made Americans warier and more confused.

**KARMEN TRZUPEK**  
InformedDNA

to patients without clinical interpretation. Patients may misinterpret the meaning and importance of the results and take action that is not warranted.”

Ms. Trzupsek warns this could lead to disastrous results, such as unnecessary surgeries or and/or inaccurate risk identification for family

All scientists need to be aware of the limitations of the data and be very transparent about its use.

**DR. JENNIFER MILLER**  
Bioethics International



members. Patients typically turn to their physicians for help, but because most physicians are not well-versed in interpreting the ambiguities and limitations of genetic testing, those physicians may refuse to review the test report or may misinterpret the results as well.

“For example, some patients have incorrectly concluded that they were at heightened risk for cardiac arrhythmia based on DTC genetic testing results and have unnecessarily had a cardiac defibrillator placed as a result,”

she says. “Others have discontinued breast cancer screening after erroneously believing that a limited, negative BRCA result eliminates their risk of hereditary cancer.”

The popularity of DTC testing highlights a growing interest and willingness to participate in genetic testing, she says, but the pharmaceutical industry must reflect on these cautionary tales and approach the use of genetic testing with an ethically minded patient focus. **PV**

# Be inspired.

Be here.

**Pharma****VOICE**

THE FORUM FOR THE INDUSTRY EXECUTIVE