

Consumer genetic testing customers stretch their DNA data further with third-party interpretation websites

June 14 2019, by Sarah Catherine Nelson



Credit: Lan Yao from Pexels

Back in 2016, Helen (a pseudonym) took three different direct-to-consumer (DTC) genetic tests: AncestryDNA, 23andMe and

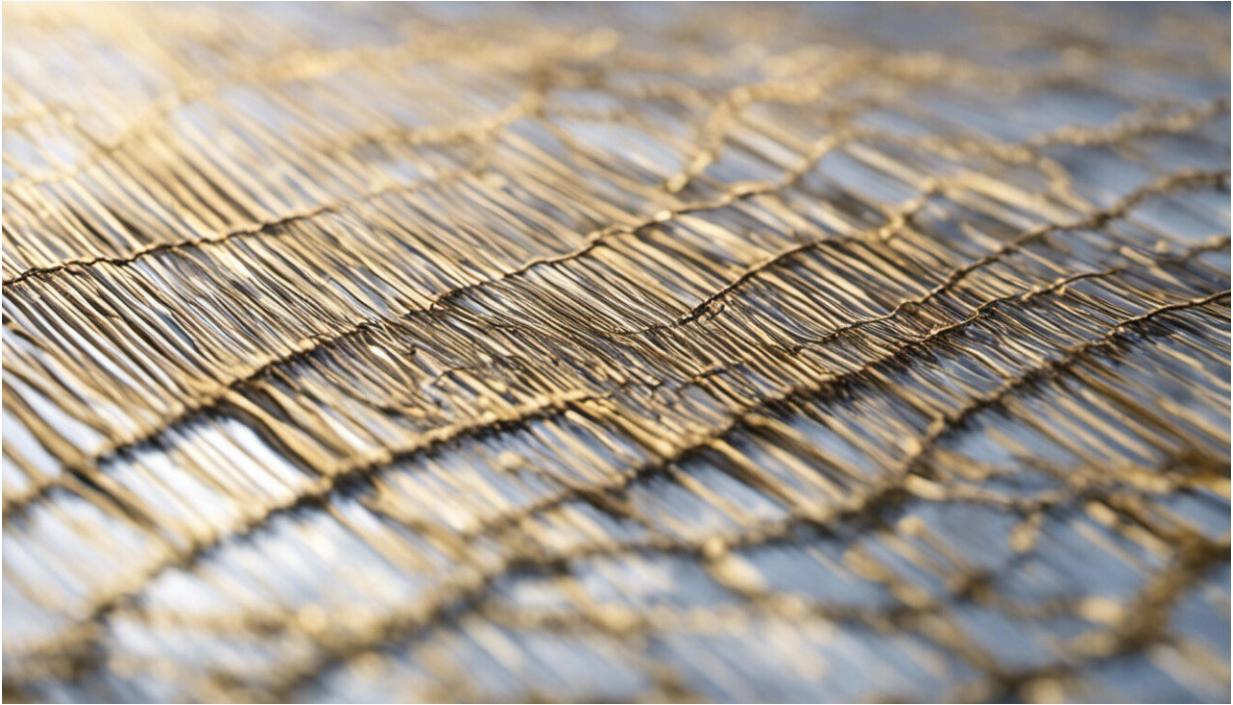
FamilyTreeDNA. She saw genetic testing as a way to enhance her paper trail genealogy research, and it panned out when she matched with several new relatives.

Helen is one of over 26 million individuals who [have reportedly taken](#) a DTC genetic test. That's a lot of spit in tubes being mailed to companies that promise customers information about their health, ancestry and family trees.

Notably, the search for genetic insights doesn't always stop with the interpretations provided by the DTC companies. One of Helen's matches on AncestryDNA told her how she could stretch her personal genomic information further: by downloading her raw genetic data, that long list of As, Cs, Gs and Ts at each of the DNA sites the DTC company measured, and then uploading it to third-party interpretation tools online such as GEDmatch and DNA.land to find more relatives.

Helen enthusiastically did so and joined Facebook groups dedicated to helping people use their genetic data to flesh out their family trees. While Helen wasn't initially seeking [health information](#), on these forums she learned about the third-party tool Promethease and decided to upload her data there as well. She thought, "Well, for five dollars—we'll see what it says."

Researchers don't have a very clear or comprehensive picture of how DTC customers use their raw data and these kinds of third-party tools. [As a genetics researcher](#) interested in the ethical and social implications of genomics in research, clinical care and everyday life, I think it's important to address this knowledge gap—particularly given questions about whether and to what extent these third-party tools are or should be regulated.



Credit: AI-generated image ([disclaimer](#))

Making the most of raw genetic data

[I interviewed Helen as part of a larger research study](#) to better understand the perspectives, experiences and motivations of those accessing their raw DTC data and using third-party interpretation tools.

My colleagues and I conducted a survey of about 1,100 DTC customers recruited via social media and followed up with interviews of 10 respondents—to our knowledge, the largest survey of this topic to date.

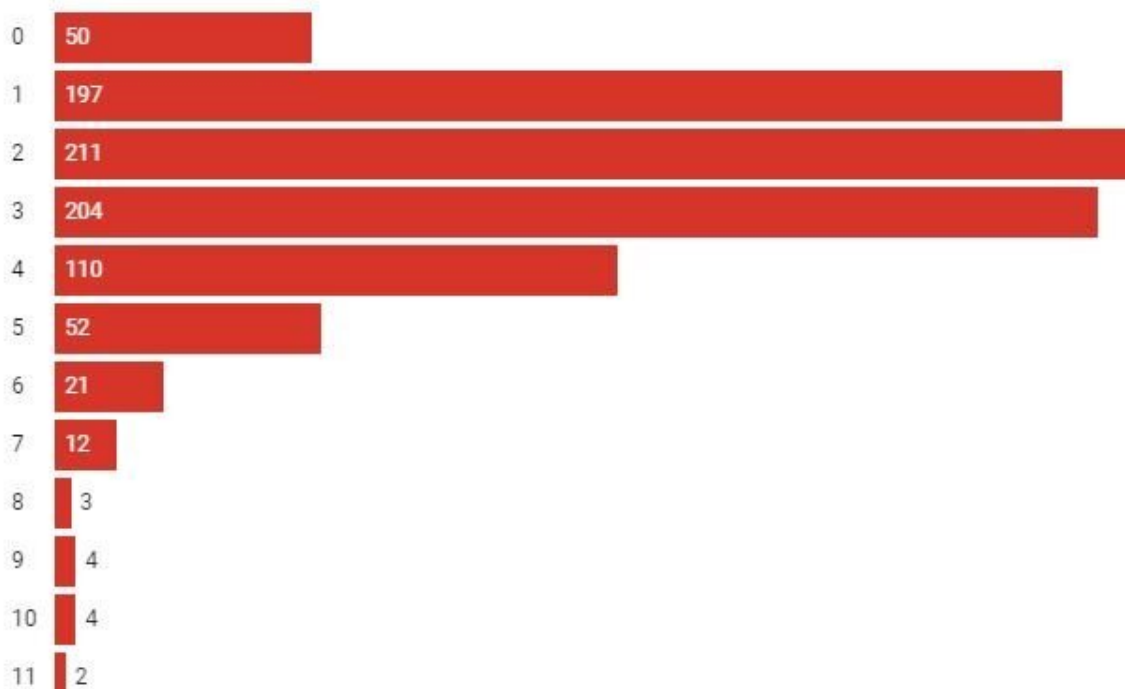
Eighty-nine percent of our survey participants had downloaded their genetic data from a DTC company, and most of those downloaders (94%) had also used one or more third-party interpretation tools—three tools on average. The most commonly used tools were GEDmatch (84%

of tool users), Promethease (63%) and DNA.land (55%).

One notable aspect of our results is that over half of tool users (56%) used both health-related and non-health-related, such as ancestry and genealogy, tools. We called this phenomenon "crossover" use. These crossover users were significantly different from people who used only one tool type in terms of demographics, which DTC tests they had taken and what initially motivated them to do DTC testing.

How many third-party interpretation tools did people use?

Researchers asked 870 people who had downloaded their genetic data from DTC companies which third-party tools they'd used to interpret it. People used an average of three different tools, ranging from zero tools used (50 respondents) to 11 different tools (two respondents).



Credit: The Conversation

For example, the percentage of users who had ordered 23andMe increased from the non-health-only to crossover to the health-only group, with a reverse trend for both AncestryDNA and FamilyTree DNA tests. While this trend is as you might expect, it was surprising how many respondents initially ordered DTC tests focused on ancestry and genealogy—like from AncestryDNA and FamilyTree DNA—who went on to use their genetic data from these companies in health-related third-party tools.

Imagine a DTC customer such as Helen who first focused on genetic genealogy. After matching with some new relatives on GEDmatch, she went on to plug her data into Promethease. There she saw thousands of reports of potentially increased genetic risk for diseases ranging from age-related macular degeneration to restless legs syndrome—quite a distance from where her genealogy quest started.

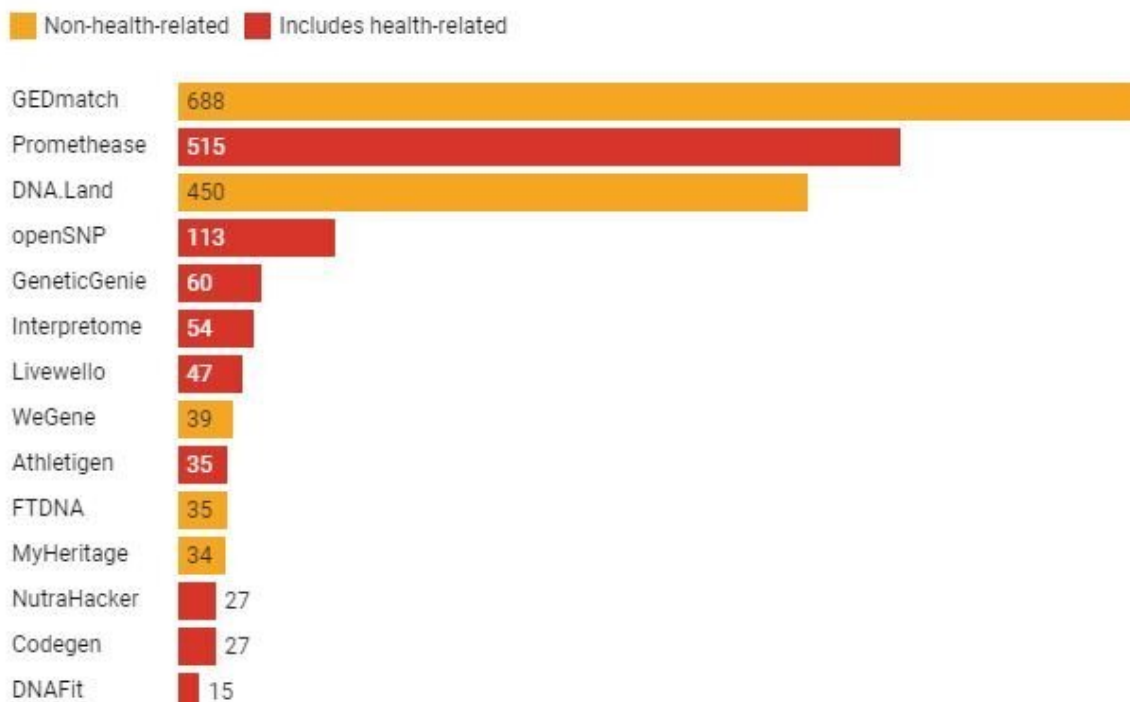
Uploading genetic data to a variety of third-party tools makes sense when you consider that DNA carries multiple kinds of information: health risks, family relations and more distant genetic ancestry. The genome is like a Swiss Army knife in that you can draw on different characteristics depending on what you want to do or know.

Through our follow-up interviews, my research group learned how crossover users arrive at such different third-party tools. As with Helen, a common reason was hearing about multiple tools on social media venues such as Facebook groups or subreddits.

And once you've started plugging your raw data into one tool, there is relatively low activation energy required to use additional tools. Other reasons people tried additional third-party tools included initial lack of interesting findings in one domain and general curiosity to extract as much information as possible.

Which third-party tools did users turn to?

These are the third-party interpretation tools respondents reported using. Some of the tools focus on family relationships and genetic ancestry, while others include some type of health- or wellness-related information.



Credit: The Conversation

More interpreters, more implications

Once relatively obscure, third-party tools have come into the spotlight over the past year. The third-party genealogy website GEDmatch helped crack the Golden State Killer murders and [subsequently dozens of other cold cases](#). The health-related tool Promethease [garnered headlines when](#) individuals were incorrectly told they were at increased risk for serious diseases—though these false positives ultimately came down to errors in the DTC [genetic data](#), rather than Promethease's interpretation.

Genetic professionals and especially [genetic counselors who are on the frontlines](#), seeing patients with their third-party reports, have well-founded [concerns about the quality and reliability of results from DTC testing companies and third-party tools](#) alike. This unease is likely exacerbated by the variability in [scientific approaches and privacy and security practices](#) across third-party tools. While some rightly question the adequacy of current oversight for both [privacy controls](#) and reliability of [health-related information](#), tighter regulation could restrict individual freedom to pursue personal genetic information.

Though my colleagues and I were able to reach over a thousand DTC customers in our study, we cannot say that our findings generalize to all DTC customers. Because of how we recruited participants, it's likely that we sampled a very motivated and engaged group of people. The DTC testing and third-party tool landscape is also changing rapidly. Just in the past year, for instance, GEDmatch changed its terms of service [regarding use by law enforcement](#) more than once, My Heritage [expanded into health-related offerings](#), and [Gencove retired its consumer-oriented product](#). Any research in this area is on shifting sands.

But this kind of study can also help inform genetics professionals who are considering delivering raw DNA data in other contexts. This includes researchers, such as those [planning to return raw data to participants](#) in the National Institutes of Health's [All of Us](#) precision medicine project. Clinicians who order genome sequencing tests may enable raw data access; patients have the [legal right to their full laboratory reports](#). Our study can shed some light on what individuals who receive raw data via these other routes might do with it.

Overall, our study illustrates the complexity and variety of third-party [tool](#) users' motivations, experiences and actions. These findings should inform, but probably won't simplify, the job of any regulators grappling with whether and how to respond to this growing field of genetic

interpretation.

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