


A genetic counselor and a research assistant looked online at the consumer-initiated genetic testing industry. We were left with more questions than answers. What does this mean for consumers?

APPROACH We performed a systematic evaluation of **consumer-initiated genetic tests** based on US companies' websites. We recorded test delivery model (e.g. DTC, physician-mediated), test type (e.g. pharmacogenetics), DNA analysis method (e.g. genotyping) and more. **We report our findings from 98 consumer-initiated health-related genetic tests offered by 47 US-based companies.**

Table 1. Features of US consumer-initiated health-related genetic tests

TOTAL		
	Total Individual Tests	98
	Total Companies	47
MODEL		
	Company Physician-Mediated	39
	Personal Physician-Mediated	29
	True DTC	27
	Unclear	3
TYPE OF TEST		
	Pharmacogenetics	21
	Genetic Health Risk	14
	Hereditary Cancer Testing	14
	Hereditary Non-Cancer Testing	12
	Carrier Testing	11
	Pediatric	3
	Polygenic Risk Score	2
	Unclear	1
	<i>Raw Data Analysis</i>	12
	<i>WES/WGS</i>	8
ANALYSIS		
	Genotyping	37
	Sequencing (NGS, WES/WGS)	34
	Unclear	14
	Raw Data Analysis	13
OTHER		
	CLIA/CAP Certifications	56
	GC Services Offered	33
	Risks Mentioned	31
	Raw Data File Generated	10
	FDA Authorized	5

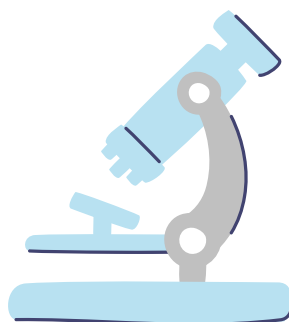
Want to know how we defined these terms? Check out our glossary! 



69% of these tests are physician-mediated, where a physician orders the test. The nature of physician involvement could not be ascertained from the websites. Lab developed tests (LDTs), such as these, do not require US Food & Drug Administration (FDA) oversight.



27% of these tests are DTC. Of these, 44% are raw data analysis products—often free or low-cost third party DNA interpretation tools that are unregulated by the FDA.



Eight companies offered WES/WGS tests. Since many do not clearly state what health-related results to expect, this raises questions about informed consent.



Most tests did not mention risks or pre-test considerations in their descriptions. Similarly, most tests did not offer (or even mention) genetic counseling services on their websites.

Consumer-initiated genetic testing: A look at the current landscape in the United States.

BACKGROUND & METHODS

There is no current resource addressing the scope of consumer-initiated genetic testing options in the US. To examine this and consider the public perspective, we conducted a systematic website review of genetic tests aimed directly at consumers. We narrowed in on 47 companies offering health-related genetic tests in 2021. Data (about the tests and how they are ordered) was collected using companies' public-facing websites; this is considered pre-test content. We did not access post-test content such as test reports.

DISCUSSION

We found substantial differences between website contents, readability and ease of use. Given this, some consumers may not have adequate information to make an informed choice based on the website alone, especially when compared to the standard-of-care in the clinical setting. What steps can be taken to protect public safety as this testing continues to grow in complexity and availability? This research calls for a broader discussion on how to better (and more consistently) communicate clinically relevant details (e.g. test accuracy, purpose, limitations, meaning of results, etc) on health-related genetic testing websites.

LIMITATIONS

We only included US-based companies; future efforts should explore the global market for a more complete look at tests that are available to Americans. There are numerous limitations to our website only approach, as our data was only as good as the websites themselves. That said, it did give us the scope of the industry from the public perspective. Using available search engines and public resources to generate our list of consumer-initiated genetics companies, there is no way to ensure our list is comprehensive, nor if the data we collected is current.

SOURCES

FDA. (2019, December 20). Direct-to-Consumer Tests. U.S. Food and Drug Administration. Retrieved September 22, 2021, from <https://www.fda.gov/medical-devices/in-vitro-diagnostics/direct-consumer-tests>; Genetic testing companies database. Crunchbase. Retrieved May 15, 2021, from <https://www.crunchbase.com> with Crunchbase Pro Membership; Phillips, A. M. (2018) Data on Direct-to-Consumer Genetic Testing and DNA testing companies. Open Source. doi: 10.5281/zenodo.1183565; International Society of Genetic Genealogy. (2013, June 2). Dna testing. International Society of Genetic Genealogy Wiki. Retrieved September 22, 2021, from https://isogg.org/wiki/Portal:DNA_testing.

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