Questions or comments? We'd love to hear from you.

Jill Furnival, MSc, CCGC
Email: jfurnival@soundrocket.com
Phone: +1 (734) 527-2170
Twitter: @jillwordsoncall

Sophia Bradley
Email: sbradley@soundrocket.com
Phone: +1 (734) 203-7552
Twitter: @bradley_sophia

Check out what we're doing at SoundRocket by visiting www.soundrocket.com or follow us on Twitter at @soundrocket !

Glossary

This glossary describes how each of these terms were used in our study. This is not a list of official definitions for these terms.

Analysis:
These definitions were straightforward; anything described using the proper language, such as SNP analysis or sequencing, was categorized as such. Some websites did not report on the type of analysis; they were labelled unclear.

Carrier Testing:
Defined based on the FDA regulation for autosomal recessive carrier screening gene mutation detection system, ref. 21 CFR 866.5940.

CLIA/CAP Certifications:
Any website that mentioned their lab certification was recorded here.

Companies:
Any US-based company that offers consumer-initiated genetic testing.

Company physician-mediated:
A test that requires a physician's sign off where the test company provides a physician to order the test.

Consumer-initiated:
A genetic test that is marketed to consumers. This was defined by 1) the use of consumer-directed language and tone on the website, and 2) the option for a consumer to take some action in ordering the test, this included instructions to speak to your healthcare provider about the test, or the ability to purchase the test themselves, with or without physician oversight.

FDA Authorized: Any test mentioning they have received FDA 501k authorization to market their DTC test to US consumers.

GC (Genetic Counseling) Services:
Any website that offers GC services as part of the test, either included in the price tag or as a referral were included here.

Genetic Health Risk (GHR):
Defined based on the FDA regulation for genetic health risk assessment systems, ref. 21 CFR 866.5950. Typically, these provide a risk of developing a disease to inform on lifestyle choices and conversations with a health care provider.

Health-related test:
This study focused on consumer-initiated genetic tests for medical purposes, which may have an impact on a person's medical care. Of note, this study did not include non-medical (e.g. ancestry, paternity) or general wellness genetic tests.

Hereditary Cancer Testing:
Defined based on the FDA regulation for cancer predisposition risk assessment systems , ref. CFR 21 866.6090.

Hereditary Non-Cancer Testing:
Added this category for hereditary single-gene conditions (e.g. hereditary cardiac conditions) that are not cancer. Typically these are medically-actionable.

Individual Tests:
We categorized *each* genetic test based on the type of test performed (e.g. pharmacogenetics, carrier test, hereditary cancer). For example, if a company offers Factor V Leiden and BRCA1/2 testing, even in one combined test panel, we would categorize it as two tests: Genetic Health Risk and Hereditary Cancer, respectively.

Pediatric:
Defined as any test where the targeted test subject is under the age of 18. We added this as a separate category as there are the additional complications regarding consent and autonomy when testing minors.

Personal physician-mediated: A test that requires a consumer to order the test through their own health care provider (e.g. physician, physician assistant, nurse practitioner, etc)

Pharmacogenetics:
Defined based on the FDA regulation for pharmacogenetic assessment systems , ref. 21 CFR 862.3364.

Polygenic Risk Score:
Defined as any test marketed with the words "polygenic risk score."

Physician-mediated:
A test that requires a physician's sign off to be ordered.

Raw Data Analysis:
Any program, paid or unpaid, that takes raw data files as input and creates a health-related genetic interpretation/report(s) from that data.

Raw Data File Generated:
Any website that offers to include a raw data file after testing.

Risks Mentioned:
Any website that offers any discussion of risks before testing at all was included here. This did not mean that the risks were fully described.

True DTC (Direct-to-consumer):
A test that can be ordered by the consumer without any physician involvement or sign off. We defined a test as "true DTC" if there was no language anywhere on the website -- in FAQS, test description or in the checkout -- to indicate that a physician was involved in the test ordering process. (This approach misses any physician involvement introduced after test checkout.)

Unclear [Model, Type of Test, or Analysis]:
The company clearly offers a consumer-initiated health-related genetic test based on their website, but details regarding how the test is ordered, type of genetic test, or specifics regarding DNA analysis are unclear. For example, this would include companies which ask consumers to email the company for more information on ordering the test.

Whole Exome Sequencing/Whole Genome Sequencing (WES/WGS):
Any test offering sequencing of a consumer's whole exome or genome, as stated in the test description. *Note: We recorded WES/WGS tests as both 1) a type of analysis and 2) a type of test, as it was often advertised as the test product.*

Links To Our 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

International Society of Genetic Geneology. (2013, June 2). Dna testing. International Society of Genetic Geneology Wiki. Retrieved September 22, 2021, from https://isogg.org/wiki/Portal:DNA_testing.

Phillips, A. M. (2018) Data on Direct-to-Consumer Genetic Testing and DNA testing companies. Open Source. doi: 10.5281/zenodo.1183565