“Well for five dollars, we'll see what it says”: A Mixed-Methods Study on Consumer Use of Third-Party Genetic Interpretation Tools

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Motivation

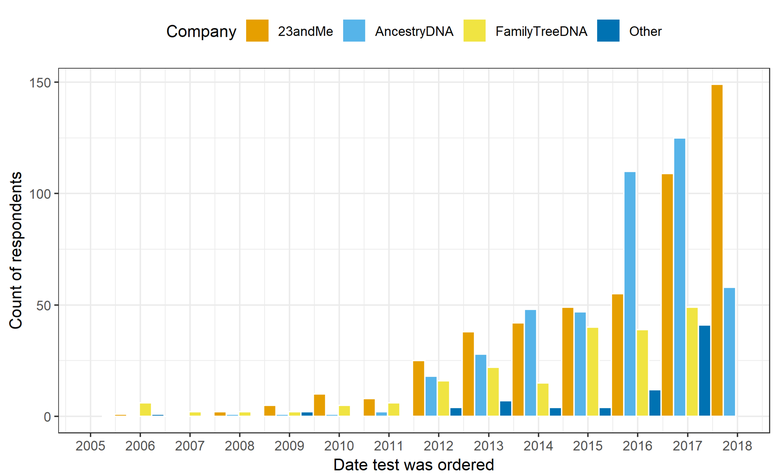
Individuals have increasing access to **“raw” or uninterpreted genetic data** via direct-to-consumer (DTC) genetic testing,[1] research participation,[2,3] or clinical sequencing.[4] Raw genetic data can be used in a heterogeneous set of online, **third-party interpretation** (TPI) tools,[5-7] which has raised concerns about false positives, data privacy, and overall accuracy of information provided. But **little empirical data exists** about how widespread raw data download is or what users do with the information from TPI tools.

Research goal: To better understand motivations and behaviors of DTC customers from testing to data download to TPI tool usage, with the broader goal of understanding how raw data return in non-DTC contexts may unfold.

Participants & Methods

- **Online survey** with questions about DTC testing, data download, and TPI tool usage
- **Recruited** convenience sample via social media, primarily Facebook groups and Reddit
  - 1,137 respondents recruited in Oct-Nov 2017
- **Grouped** tool users based on type(s) of tools used: non-health only, health only, or both (“crossover”)
- Conducted 10 **follow-up interviews** with volunteers from crossover group
- **Analyzed** quantitative survey data in R; thematic analysis of qualitative survey done in Atlas.ti
- **Posted** survey data and analysis code on openICSPR[8]

Results 1 : DTC testing



- 37% (413/1,116) ordered 2 or more different DTC tests
- 89% (870/974) downloaded raw data

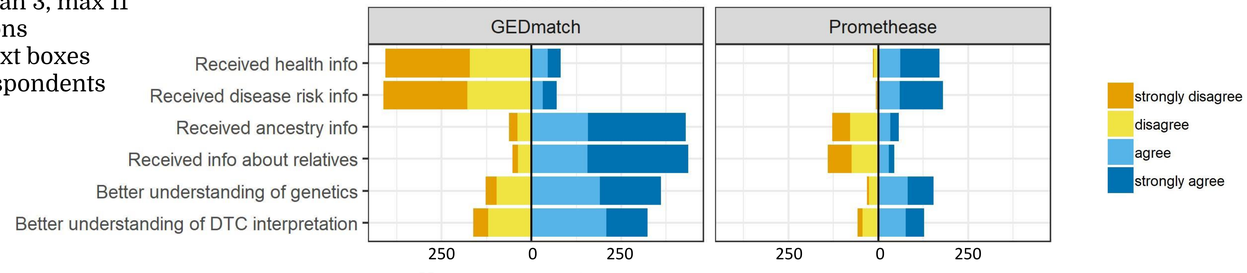
Results 2 : Third-party tool usage

- Respondents used multiple tools: median 3, max 11
- 13 tools listed in fixed response questions
  - additional tools listed in open text boxes
- Table lists tools used by 20 or more respondents

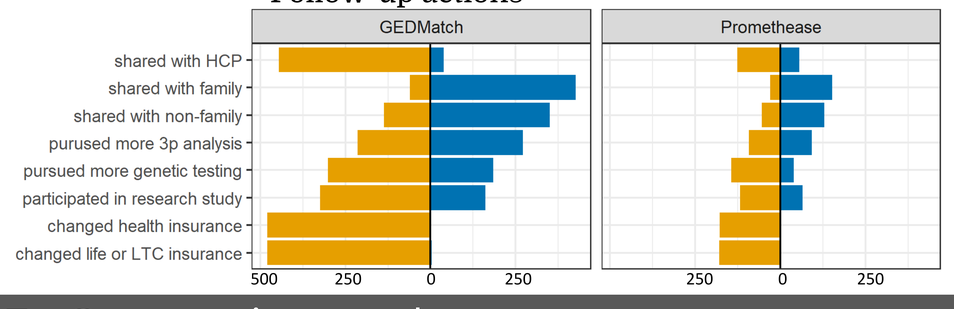
Tool	Total users (N, % of 820)	Selected for responses (N, % of 820)
GEDMatch	688 (83.9%)	505 (61.6%)
Promethease†	515 (62.8%)	188 (22.9%)
DNA.land	450 (54.9%)	58 (7.1%)
openSNP†	113 (13.8%)	12 (1.5%)
GeneticGenie†	60 (7.3%)	4 (0.5%)
Interpretome†	54 (6.6%)	1 (0.1%)
Livewello†	47 (5.7%)	7 (0.9%)
WeGene	39 (4.8%)	4 (0.5%)
Athletigen†	35 (4.3%)	1 (0.1%)
FTDNA	35 (4.3%)	1 (0.1%)
MyHeritage	34 (4.1%)	2 (0.2%)
NutraHacker†	27 (3.3%)	1 (0.1%)
Codegen†	27 (3.3%)	3 (0.4%)

†Includes health-related information

Information received



Follow-up actions



Results 3 : Comparing tool user groups

Demographics

Variable	Non-health only tools	Crossover	Health only tools
Number of respondents	263	458	98
Mean age* (SD; range)**	51.8 (14; 18-83)	45.5 (15; 18-84)	39.4 (12; 20-73)
Women (%)*	69.8	68.7	53.3
Works in genetic research/medicine* (%)**	3.0	2.3	13.3
Participant in genetic research* (%)	11.5	19.0	15.6

Additional demographics collected that were not significantly different (p>0.05): occupation, max education, race, lives in US

DTC tests and testing motivations

Variable category	Variable	Non-health only tools	Crossover	Health only tools
DTC tests ordered (%)	23andMe***	40.7	68.1	87.8
	AncestryDNA***	73.0	62.0	14.3
	FamilyTreeDNA***	37.3	32.1	3.1
Rating of DTC testing motivations[b] (% very important)	General curiosity***	55.5	71.4	82.7
	Ancestry***	69.6	69.8	41.2
	Find relatives***	62.4	50.4	14.3
	Risk for specific diseases***	16.1	34.2	54.1
	Limited family health history	21.4	21.0	17.7
	Other family members using	10.8	11.0	8.3
	Participate in research	26.3	33.8	34.7
	Raw genetic data file**	44.4	56.4	60.4

*** p<0.001, **p<0.01, *p<0.05, comparing three tool user groups

Results 4 : Interviews

Initial use of health-tools

> I was interested in finding out exactly what my DNA meant, not just what 23andMe wanted to tell me...I think I used Promethease first...from what I could tell, it was the most information...[On additionally using GEDmatch] I may have just Googled ‘things to do with raw DNA.’

Initial use of non health-tools

> I wasn't [using third-party tools] for health at all and just when I saw the Promethease and I thought, ‘Well for five dollars, you know, we'll see what it says.’

Initial use of both

> I'm adopted...I was doing all kinds of testing...partly I was trying to take control of whatever health information I could get, but then also fishing around for family connections. I was doing some of each.

Conclusions

- We observed **high volumes** of raw data download and TPI tool usage in this sample of DTC customers.
- Users often leveraged their raw data **across the domains** of health, ancestry, and genealogy
- Interviews illustrated how **social networking, initial lack of interesting findings, and general curiosity** contributed to use of multiple tool types
- DTC testing is currently the most common route to raw data; however, further research is needed to understand how broadening **access from research participant and clinical sequencing** may unfold

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References

1. Regalado, A. *MIT Technology Review* (2019)
2. NASEM (2018). doi:10.17226/25094
3. Thorogood, A. *et al. Hum. Genomics* **12,** 1–7 (2018)
4. Evans, B. J., Dorschner, M. O., Burke, W. & Jarvik, G. P. *Genet. Med.* **16,** 799–803 (2014)
5. Badalato, L., Kalokairinou, L. & Borry, P. *Eur. J. Hum. Genet.* **25,** 1189–1194 (2017)
6. Wang, C. *et al. Mol. Genet. Genomic Med.* **6,** 35–43 (2018)
7. Nelson, S. C. & Fullerton, S. M. *J. Genet. Couns.* **27,** 770–781 (2018)
8. http://doi.org/10.3886/E105721V2

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