

I. Direct-to-consumer (DTC) Genetic Testing

- Mainstream since ~2007
- Biggest companies, **23andMe**, **AncestryDNA**, and **FamilyTree DNA**, each have over 1M customers
- How it works: companies...
 - Collect customer biological specimens through “spit kits,” distributed and returned via mail
 - Extract and analyze customer DNA using SNP genotyping arrays*
 - Measure ~700K-1M sites in the customer’s genome (~0.3% of total 3B sites)
 - *Note some DTC companies starting to offer whole genome or exome sequencing
 - Companies run algorithms to generate interpreted reports on
 - Physical traits (e.g., eye color)
 - Genetic ancestry (e.g., “XX% Northern European, XX% African ancestry,” etc.)
 - Relatives (i.e., which other customers are potential relatives)
 - Health and wellness (23andMe, though has been limited by FDA regulation)



II. “Raw” Genetic Data Files

- In addition to interpreted reports, customers can also download their “raw” genotype file
 - =flat text file with the customer’s genotype at each variant measured by the company

```
# This data file generated by 23andMe at: Tue Nov 26 21:21:54 2013
# Below is a text version of your data. Fields are TAB-separated
# Each line corresponds to a single SNP...
# rsid chromosome position genotype
rs4477212 1 82154 AA
rs3094315 1 752566 AA
rs3131972 1 752721 GG
rs12124819 1 776546 AC ... (and so on, for ~1M rows)
```

- Ok, but who would download their genetic data and why....?

III. Third-Party Interpretation Tools

- Several independent (“third-party”) tools/websites exist for DTC customers to engage in further self-analysis of their “raw” genotype file
 - I’ve catalogued ~20 such sites
- Third-party tools vary widely in terms of:
 - Business model
 - Can be for-profit companies, academic groups, or “citizen scientists”/non-specialists
 - Opportunities to participate in research
 - Some tools emphasize that uploading data to the site allows user to directly contribute to research
 - Types of analysis performed
 - Many tools focus on “genetic genealogy”
 - Others provide some type of health/wellness (perhaps medical?) interpretation
 - Several of the companies create “personalized” advice for nutrition and fitness regimens

Discussion Prompts

- 1) What conceptions of genomics are advanced by DTC genetic tests?
 - a. Are these helpful or harmful to genetic research and medicine?
- 2) Should people be able to access their “raw” or uninterpreted genotype data? What are the pros and cons?
- 3) What protections and regulations, if any, should govern consumer genomics companies? What about third-party, interpretation-only tools and websites?
- 4) You can view DTC genetics and subsequent self-directed analysis as part of the growing self-tracking/quantified self (QS) movement (think Fitbits and other wearables). Is genetic data (the potential risks and benefits of personal access and use) the same or different as other types of tracking/QS activities?

References & Further Reading***DTC genetics testing in general***

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Third-party interpretation tools

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