

What Can Your Genes Tell You?

A Community Conversation, March 18, 2014

Our genes provide instructions for our physical attributes, or phenotype. So literally our genes can - in some ways - tell us what we will “look like” on cellular, system and whole body levels. This sometimes translates to understanding or predicting health status when the genetic basis of disease is well understood.

In order for our genes to be informative for understanding and predicting health, we need **access** to our genetic information and the ability to **interpret** it. But *who gets access and how? Who is trusted with the interpretation?* These questions are at the heart of the recent actions of the Food and Drug Administration (FDA) against direct-to-consumer genetic testing company 23andMe.



What information does 23andMe provide?

23andMe is a direct-to-consumer genetic testing company founded in 2006. In exchange for a sample of spit and \$99, customers of 23andMe's Personal Genome Service receive a variety of genetic information, including:

- **Health Risk:** for 122 diseases, the customer is categorized as higher or lower risk compared to the general population. There is a range of evidence types underlying each of these risk assessments, from well understood to more provisional. Each result comes with a confidence rating and with links to the underlying research studies. (31/122 are high confidence)
- **Drug Response:** for 25 drugs, the customer is classified either (1) by metabolic status—that is, how quickly the body processes a drug: rapid, fast, increased, reduced, or (2) as a positive vs negative responder. (12/25 are high confidence)
- **Inherited Conditions:** for 53 inherited conditions, the customer is classified as being a carrier (or not), meaning if their partner is also a carrier they have a 25% chance of having an affected child. (53/53 are high confidence)
- **Traits:** for 60 different traits, the customer is provided likely outcomes based on genetic data. Examples are wet vs dry earwax, lactose intolerance, and norovirus resistance. (13/60 are high confidence)
- **Ancestry:** Information about your maternal and paternal lineage, your global origins, your % Neanderthal, and a relative finder.

Customers can also opt in to participate in the research arm of the company, “23andWe.” The company has conducted numerous research studies, including ones on Parkinson’s disease and sarcoma.

Why has the FDA gotten involved with 23andMe?

The FDA is a federal agency that oversees (“regulates”) many things: food, cosmetics, medical devices, drugs, and biologics (e.g., vaccines). The FDA’s role is to ensure both the **safety** and **effectiveness** of these products. The FDA has come to consider 23andMe’s Personal Genome Service a “medical device” and therefore subject to their regulatory oversight. On November 22, 2013, the FDA sent 23andMe a letter demanding that they stop selling their product until they can provide the FDA with evidence that their Personal Genome Service is **safe** and **effective**. 23andMe complied with this directive; *new customers can currently only receive their ancestry results*.

Big picture considerations in FDA regulation of direct to consumer genetic testing...

- Should individuals have direct access to their genetic information or should health care providers be the gatekeepers?
 - o *What are the harms and benefits of each model?*
- Should genetic information about health be regulated as a medical device by the FDA? What about other types of health information?
 - o *What are the harms and benefits of regulation? What alternatives exist?*
- What is the desired balance between individualism and paternalism when it comes to oversight of direct to consumer genetic testing?

Key Terms

Genotype vs. **Phenotype**: genetic data (i.e., sequence of DNA) vs expression of a trait or disease

Penetrance – The degree to which a genotype exhibits a phenotype. BRCA1 = 80% penetrance

Analytic validity – does the test accurately measure the genetic variation

Clinical validity – does the test accurately predict the presence, absence, or risk of a specific disease

Premarket approval – FDA process of scientific and regulatory review for **medical devices**, products designed to help diagnose or treat any disease or health condition

SNP (Single Nucleotide Polymorphism, “snip”) – Within a short section of DNA that is variable between people; single molecule changes are associated with different phenotypes and can serve as an identifying feature in the genomic landscape

Resources for further reading and listening

KQED, “FDA Orders 23andMe to Halt Sales,” 11/26/13 [Radio Program], www.kqed.org

Genomics Law Report Blog, posts from 12/3/13, www.genomicslawreport.com

Klein, E. “Should the FDA stop you from scaring yourself with 23andMe’s DNA test?” Washington Post, 12/6/13

Janssens, S. “How FDA and 23andMe Dance Around Evidence That Is Not There.” Huffington Post, 1/27/14

The 23andMe blog, “The Spittoon,” posts from Nov-Dec ‘13, blog.23andme.com

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