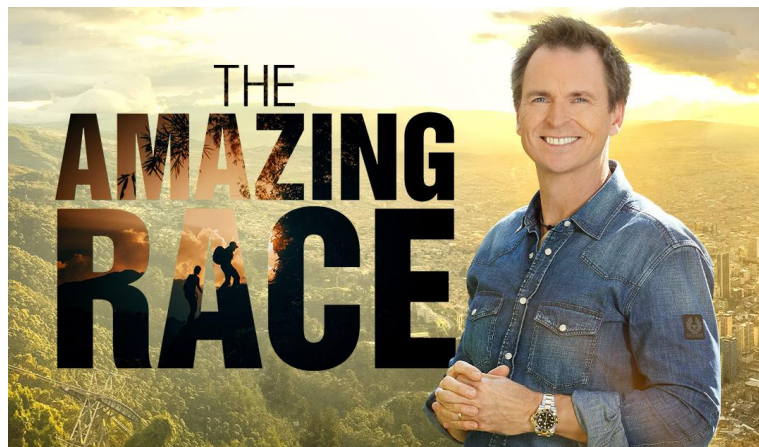
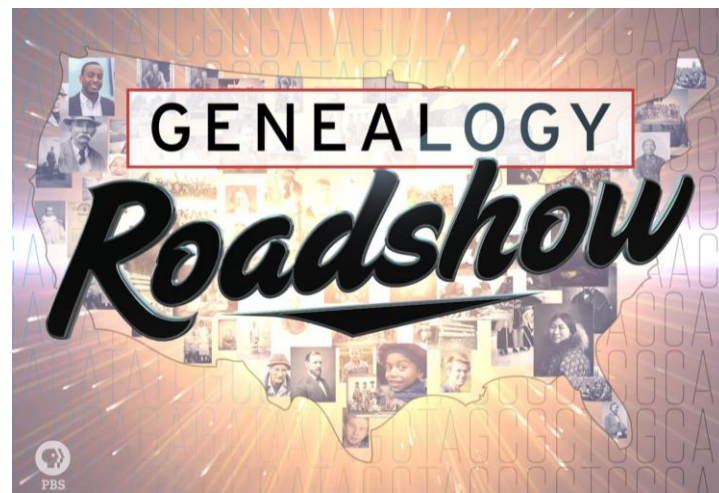


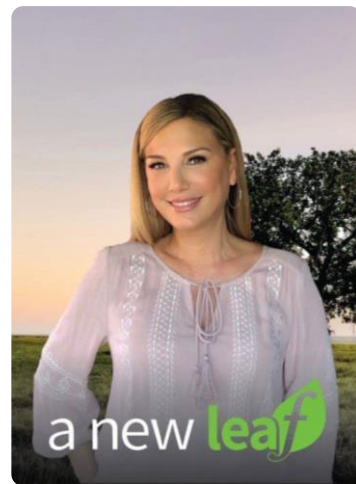
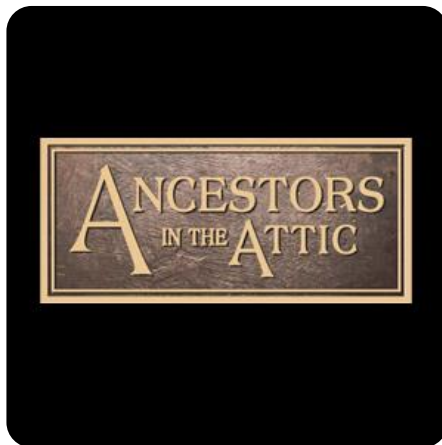
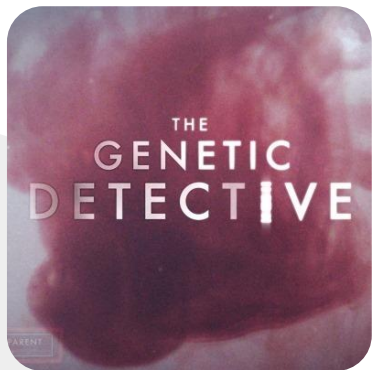
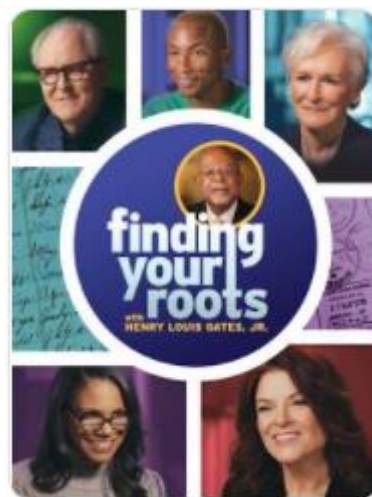


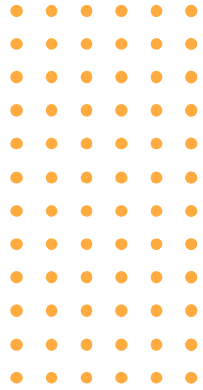
The Importance of Genetic Counseling

Genetainment





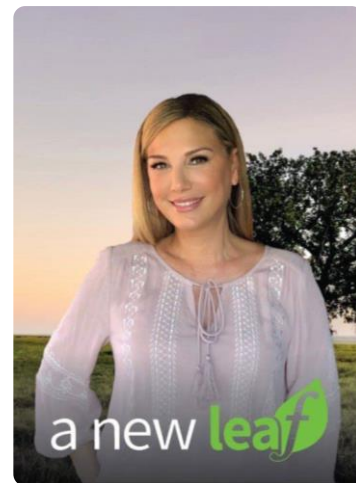
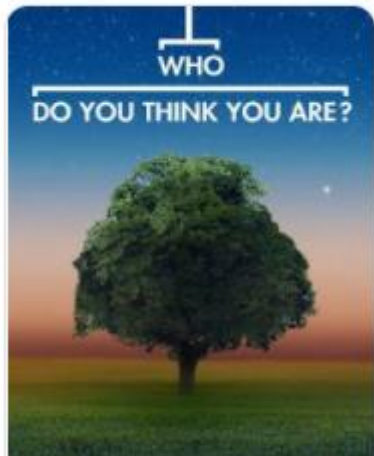




What does this have
to do with direct to
consumer testing?







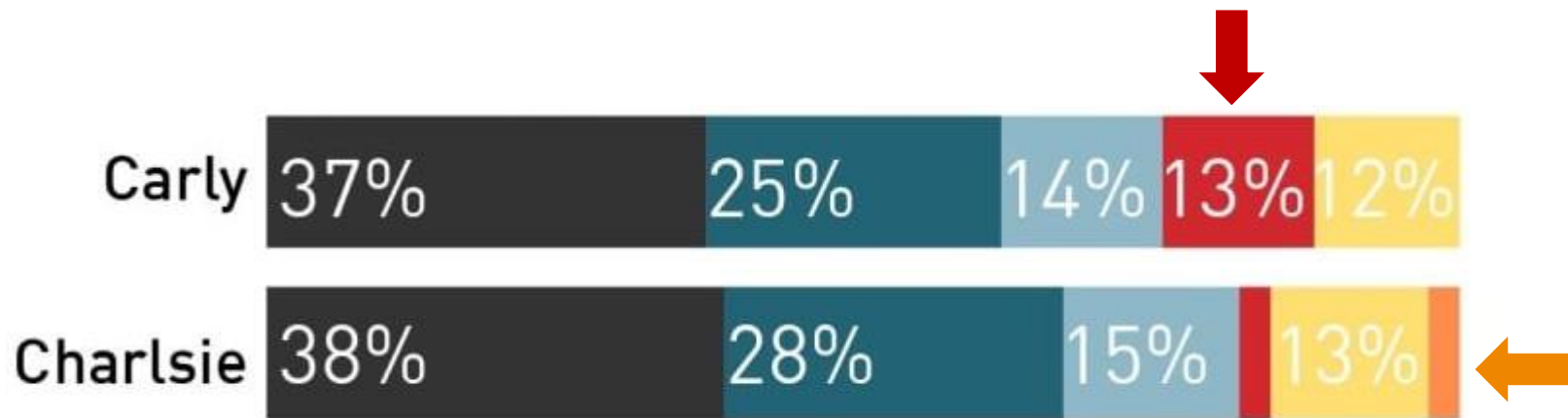


Sicily
Poland
Ukraine



Different results from
different companies

23andMe



Italian

Eastern European

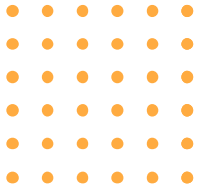
Balkan

Broadly European

Others*

French & German

*Includes: Iberian, Broadly Southern European, Western Asian & North African, North African & Arabian, Western Asian, Broadly Western Asian & North African



What about health?

Can DTC testing give us accurate information about our health?

Occasionally



Types of Genetic Disorders

Monogenic Disorders

Variant in 1 gene



Polygenic Disorders

Variants in many genes





To see the list of approved testing, go to:

<https://www.fda.gov/medical-devices/in-vitro-diagnostics/direct-consumer-tests#list>



HEALTH
+
ANCESTRY

Phenylketonuria

What do we test?

23 variants in the PAH gene.

>500 variants found



BRCA1 & BRCA2

BRCA1



BRCA2



BRCA1: >1,600 variants

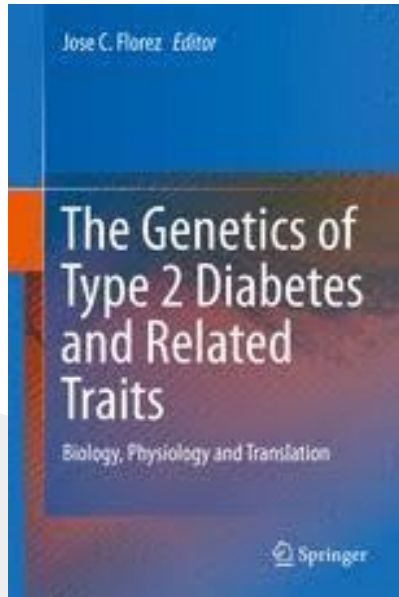
BRCA2: > 1,800 variants

Polygenic



Type 2 Diabetes

>400 susceptibility loci





Jamie, your genetics are associated with an **increased likelihood** of developing type 2 diabetes.

Based on data from 23andMe research participants, people of Hispanic or Latino descent with genetics like yours have an estimated **79% chance** of developing type 2 diabetes at some point **between the ages of 40 (your current age) and 80.**



Your genetic likelihood is higher than typical. But your overall likelihood also depends on factors like weight, diet, and exercise. This means it's important to maintain a healthy lifestyle.

How do they come up with these risks?



What places do they look at?

How do they determine “weight” of variants?

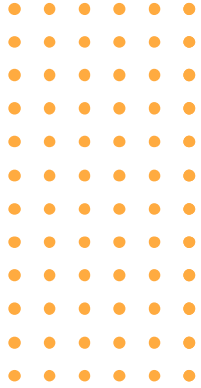
March 8, 2019

23andMe thinks polygenic risk scores are ready for the masses, but experts aren't so sure

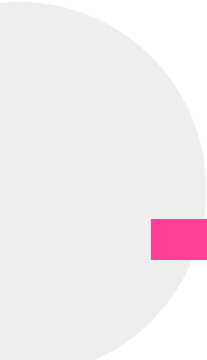
A new genetic test that estimates your risk for diabetes is probably less useful than standing on a scale.

June 10, 2020

23andMe says it's 'part of the problem' on racial inequity. We asked geneticists what the company can do about it

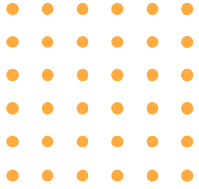


What should
people do?

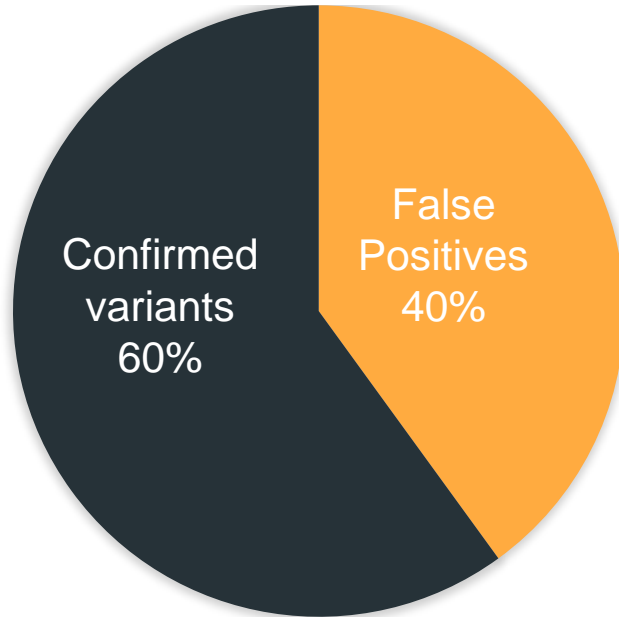


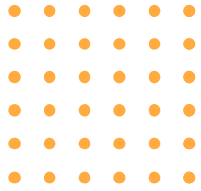
Think about...

1. Why are you doing the test?
2. What will you do with the results?
3. How much are you going to believe the results?
4. How important is privacy to you?

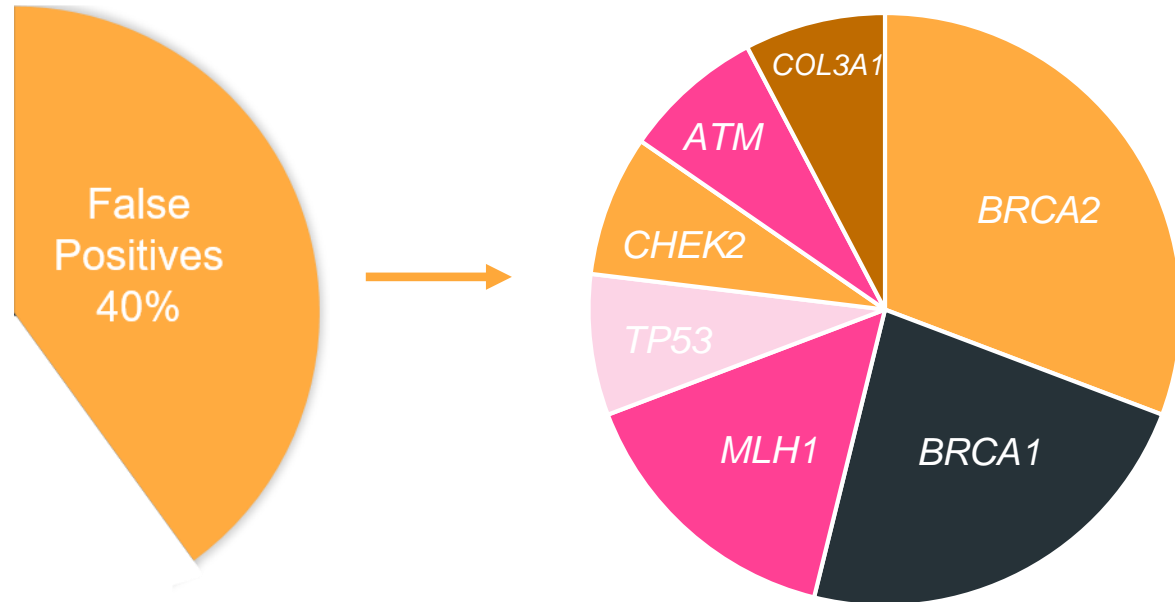


Confirm Results!





Confirm Results!



	DTC Testing	Genetics
Determine at risk disorders		✓
Explanation of results		✓
Genetic counseling		



RESOURCES

IMAGES:

- Factors that contribute to diabetes: netmeds.com/health-library/post/type-2-diabetes-mellitus-causes-symptoms-and-treatment
- Ashkenazi Jewish map: Jewish Genetic Disease Consortium at jewishgeneticdiseases.org/jewish-genetic-heritage/
- Marketing funnel: neilpatel.com/blog/how-marketing-funnels-work/
- Carly & Charlsie – twins: geneticliteracyproject.org/2019/01/31/ancestry-results-for-identical-twins-illustrate-flaws-in-consumer-genetic-testing/
- Orig3n: shop.orig3n.com/products/superhero
- Heights, public speaking, sounds: 23andMe from <https://blog.23andme.com/health-traits/23andme-adds-four-new-trait-reports/>

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- Godet, I & Gilkes, DM. (2017). BRCA1 and BRCA2 mutations and treatment strategies for breast cancer. *Integr Cancer Sci Ther*, 4(1). doi: 10.15661/ICST.1000228
- Krentz & Gloyn (2020) Insights into pancreatic islet cell dysfunction from T2D. *Endocrinology*, 16:202–212
- Liang, Y., Huang, M., Cheng, C., Chao, H., Fwu, V., Chiang, S., Hsiao, K., Niu, D., Su, T. (2014). The mutation spectrum of the *phenylalanine hydroxylase (PAH)* gene and associated haplotypes reveal ethnic heterogeneity in the Taiwanese population. *Journal of Human Genetics*, 59: 145–152



THANKS

Tiffany Lepard Tassin

tlepard@uams.edu

501-526-7700

