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

Carly 37% 25% 14% 13% 12%

Charlsie 38% 28% 15% 13%

Italian  
Eastern European  
Balkan

Broadly European  
Others*  
French & German

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
Phenylketonuria

Gene: PAH

What do we test?
23 variants in the PAH gene.

>500 variants found

Liang et al., (2014)
BRCA1 & BRCA2

BRCA1: >1,600 variants
BRCA2: > 1,800 variants

Godet & Gilkes (2017)
Polygenic
Type 2 Diabetes

>400 susceptibility loci

Krentz & Gloyn (2020)
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?

The 23andMe Type 2 Diabetes Health Predisposition report estimates your chances of developing type 2 diabetes by looking at more than 1,000 places in your DNA.

What places do they look at?

How do they determine “weight” of variants?
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.

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!

- Confirmed variants: 60%
- False Positives: 40%

Tandy-Connor et al. (2018)
Confirm Results!

False Positives 40%

Tandy-Connor et al. (2018)
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<th>DTC Testing</th>
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<td>Determine at risk disorders</td>
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<td>Explanation of results</td>
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<td>Counseling and support</td>
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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/
• Orig3n: shop.orig3n.com/products/superhero
• Heights, public speaking, sounds: 23andMe from https://blog.23andme.com/health-traits/23andme-adds-four-new-trait-reports/

REFERENCES:

• Tandy-Connor, S., Guiltinan, J., Krempely, K., LaDuca, H., Reineke, P., Gutierrez, S., Gray, P., & Davis, BT. (2018). False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate care. Genetics in Medicine, 20(12):1515-1521
• Krentz & Gloyn (2020) Insights into pancreatic islet cell dysfunction from T2D. Endocrinology, 16:202–212
THANKS

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