

When consumer genetic tests disagree on critical mutations

[Matt Fender] wasn't worried last December when he clicked a button to dump all the raw data from his 23andMe genetic test into a DNA search engine called Promethease, which sorts through data for gene variants that have received a mention in the medical literature.

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Mr. Fender's update included something new: the terms "PSEN1" and "pathogenic." Mr. Fender is a coder, not a geneticist, but he had spent enough time scrolling through his 23andMe results to know he had gotten some bad news.

The PSEN1 mutation is associated with an early-onset form of Alzheimer's, and it is often described as "100 percent penetrant," which he quickly came to understand meant no exceptions — everyone with the variant gets the disease.

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Meanwhile, he happened to see a holiday special — \$69 — for Ancestry's genetic risk test. He realized he could use it to, in effect, get a second opinion about his PSEN1 variant.

Five weeks later, the results were ready. He downloaded his raw data and returned to Promethease. An hour later, he had a new report. He looked for PSEN1 at the top of the list. It wasn't there.

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[Matt] eventually persuaded his doctor to order a clinical test of the PSEN1 gene. It was negative.

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A person with fewer resources or different inclinations might have lived for years under that cloud, waiting to get sick.

Read full, original post: [23andMe Said He Would Lose His Mind. Ancestry Said the Opposite. Which Was Right?](#)