# The Potential Perils of Pauline: Personal Genotyping for BRCA 1 and 2

*by*
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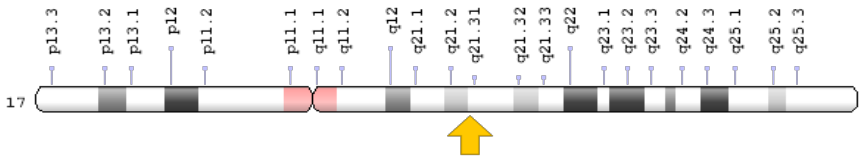


"Hey Brad, I just got my results back from 23andMe!" Pauline had always been interested in her ancestry, but as a college student she hadn't felt she could justify spending so much money on those advertised tests she had seen. Her uncle, however, had thought it would make for a perfect 20th brithday gift since she was currently taking a course in general biology.

Brad, who was also in her class, looked up from his laptop. "Great, what did you find out?"

"Well, my family background is mostly northwestern European—we knew that—but I also have some East Asian and northern African ancestry."

"Did you also get your health risks tested? I heard that they're now able to test if you'll get breast cancer."

"Yes," Pauline nodded. "I have no chance for breast cancer or Alzheimer's disease or Parkinson's disease."

"Are you sure? How do they know that you have no risk? Do they sequence your genes?"

"No, I'm not sure," Pauline answered. "But I got a readout that said something about not finding gene variants or mutants or something."

"Didn't you tell me that your grandmother had breast cancer?" Brad asked.

"Yes, but apparently she didn't have whatever gene 23andMe tested for," Pauline replied.

"But couldn't you have inherited the gene your grandmother had? Wouldn't that put you at risk?"

"All I know is that 23andMe said I didn't have the gene variant. I'm pretty sure that means I don't have to worry."

Brad wasn't convinced. "What about your age? You're 20, right? If you have a family history of breast cancer, shouldn't you take some precautions? How did 23andMe test for breast cancer? Did they have to get approval from the government? Do doctors have all the information they need to declare someone risk-free from cancer?"

"You sure have a lot of questions, Brad," Pauline said. "I think you care more about this test than I do."

"I'm just concerned about you," he replied. "I don't want you to ignore potentially harmful symptoms just because a company said you don't have anything to worry about. I'll tell you what: let's examine this test together and get a better idea of what it really means."

"Thanks, Brad," said Pauline. "You're a great friend!"

*Note: For an example of a 23andMe report on health/disease risks see: <https://permalinks.23andme.com/pdf/samplereport_geneticheath.pdf>.*

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Image shows cytogenetic location of BRCA1 gene at 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31. Image courtesy of NCBI, https://ghr.nlm.nih.gov/gene/BRCA1#location.

## *Questions*

1. What do genes do? Briefly describe how genes are responsible for health-related traits. How do people inherit genes?

2. What are mutations? How do mutations affect traits? Are mutations always harmful or can they be beneficial?

3. In general, what is cancer? At the cellular level, what causes cancer? How might carcinogens (such as cigarette smoke or ultraviolet light) bring about these cellular changes?

4. What do the genes BRCA 1 and BRCA 2 do in human cells? How might they be responsible for cancer?

5. What environmental risk factors exist for breast cancer? How might they affect genes?

6. What is the difference between genotyping and gene sequencing? Where does the name 23andMe come from? How does 23andMe collect customers' samples? Briefly, how do they determine the data they supply in their reports, such as ancestry and health risks?

7. Did 23andMe have to go through testing by the government? If not, should they have been required to go through testing, why, and which agency should they have gone through? If they have gone through testing, how was that testing done, what organization was responsible for conducting the testing, and what were the criteria for approving the test?

8. If a variant (or mutation) is found on a report, what steps should the customer take? Are these steps recommended by 23andMe or are they common sense?

9. In the United States, does health insurance generally pay for tests by 23andMe? Does health insurance pay for other genetic tests? If so, under what conditions will they pay?

10. For what diseases does 23andMe test? If a customer is found to be at risk for one or more of these diseases, how is this information protected? What would happen if the customer's employer were to obtain these results?

11. If you were in Pauline's position, how would you proceed? Would you (a) relax, confident that you are at no (or very low) risk for breast cancer; (b) worry that 23andMe may have gotten the results wrong or that the results are meaningless; (c) be proactive in the area of health by avoiding environmental risk factors (from Question 5) and consider a mastectomy (surgical removal of one or both breasts) before cancer is detected; or (d) consider another course of action? Please give reasons for your decision.