

## **Scenario A:**

You have just received a copy of your personal genome sequence. Included in the results is this piece of information:

You have tested positive for Huntington's Disease – a degenerative neurological condition that can strike as early as in one's 30s or 40s. If you have the mutation, it is 100% certain you will get this disease. You are feeling a mix of relief to finally have all the wondering behind you, but are also very upset and distressed to have these fears confirmed. You are seeking support from friends and family.

Your mother knows that you were planning to get your genome sequenced. Your grandfather had Huntington's, and it was a source of confusion and secrecy in your family. She has decided she would rather not know and wants to continue to live her life as she always has. She feels that since there is no treatment or cure for Huntington's, what's the point of learning if she carries the mutation?

Because Huntington's is an autosomal dominant condition, your positive status for the gene mutation implicates your mother: It means she will definitely have the disease. You are 23 years old, she is 47.

### Discussion points:

How do you handle this piece of information with your mother?

If you tell her that you have the mutation for Huntington's, you are also telling her that she has it as well. You know she doesn't want to know – but you need her emotional and financial support. Do you tell her? What are the ethical issues at play?

How might this information change your life? The lives of the other members of your family?

## **Scenario B:**

You have just received a copy of your personal genome sequence. Included in the results is the following piece of information:

You have a slightly lower than average risk for breast cancer, based in part on your status with respect to the so-called “breast cancer genes”, BRCA1 & BRCA2. All people have the BRCA genes, and men also get breast cancer, although less often than women. Sequencing looked at how your BRCA genes were functioning and it appears to be normal.

Your mother and aunt have developed breast cancer and were both diagnosed in their 40s. You are relieved to have learned you are not a carrier of the mutations.

Because of the family history of breast cancer, you have been seeing your doctor more frequently than most people, and paying for the extra screenings out of your own pocket.

### Discussion points:

How confident are you that you are “in the clear”? Will you continue to pay extra for the additional screenings? Do you think your health insurance would have covered the screening if you did have the mutated BRCA genes?

If you were a health insurer, would you think it is smart to cover (pay for) extra screenings if there is a strong family history, but no genetic indicators that suggest an elevated risk?

What do you make of the fact that breast cancer appears to “run in your family”? Would you advise your family members to have their genome sequenced? Are mutations in the BRCA genes the only factors involved with breast cancer?

Can you think of ways that your family might get involved with a research study? What would the scientists be looking for? New genes involved, or other factors such as lifestyle, diet, and environment?

## **Scenario C:**

You have just received a copy of your personal genome sequence. Included in the results is the following piece of information:

You carry a genetic mutation that places you at a much higher risk than average for colon cancer. This is a surprise, as nobody in your family on either your mother or father's side has ever had colon cancer. You read that colon cancer runs in families, so you ask your three siblings to also get sequenced and they agree to do so.

The results of the sequencing prove to be very confusing. It appears that you and your siblings may not share the same mother and father. Your DNA does not match your siblings as might have been expected, and there are too many differences for you all to be full, biological siblings.

### Discussion points:

What could be the explanation? Was there a mistake at the lab? Are you adopted and your parents never told you? Or maybe something there is else going on?

Learning about your genome has proven to be more than you bargained for. How might this sudden question of your genetic lineage change things in your family?

Early and frequent screening for colon cancer is an effective prevention tool. Knowing you carry the mutation is a potentially a life-saving piece of information. Despite the unexpected and possibly distressing information, was it worth it to learn about the colon cancer risk?

How might you seek answers about the fact that you and your siblings appear to not be full biological siblings?

What might be some of the other unexpected or surprising results of personal genome sequencing?

## **Scenario D:**

You have just received a copy of your personal genome sequence. Your personal history was a significant part of why you decided to learn about your genome:

You have always known your mother used an anonymous sperm donor when you were conceived. Shortly after you were born, your mother married and had two more children. You are a family of five, very close knit and the fact that your biological father was a sperm donor is not a big deal.

Since you don't know anything about the medical history of your biological father or his family, you decide to get your genome sequenced, as the resulting information could inform you of your possible risks for certain diseases and conditions.

When you receive your sequence, you learn you have a lower than average risk for certain types of heart disease, have dry (as opposed to wet) earwax, can't taste the bitterness of certain foods like Brussels sprouts, and have a much higher than average risk for adult-onset diabetes and that you are a carrier for Cystic Fibrosis.

Sequencing has been around a few years at this point, so the cost of sequencing your genome was covered as one of your health insurance benefits.

### Discussion points:

What if your curiosity is piqued and you decide you want to learn more about your biological father – who is your donor? How might you go about finding him? What are his personality, career, and medical history like? Do you have other half-siblings you've never met?

Consider how this might impact you and your family – put yourself in the shoes of your mother, your father who has raised you, and your siblings. How might they feel about the medical information you learned? Could your mother or father feel nervous that the family dynamic might change somehow?

Now, consider the perspective of the sperm donor – at the time he was a college student and frequently donated at the local clinic. He hasn't really thought about this time in his life for decades. Now he is 51, married with 2 teenage children of his own, and lives approximately 18 miles from you. How might he react to a request for contact with a teenage child he never knew he had?

## **Scenario E:**

You have just received a copy of your personal genome sequence. Included in the results is the following piece of information:

You are a “fast metabolizer” in terms of a prescription drug that you take to prevent potentially fatal blood clots. This piece of information will help you choose the right dosage, making it much more effective. You also learned you are at well above average risk for a heritable form of eye disease that can lead to blindness – macular degeneration. This is particularly distressing as you have paid for college via ROTC and plan to enter the Air Force as a pilot.

After receiving your results, you notice a “personal genomes” group on Facebook. There are some “fast-metabolizers” as well as “macular degenerators” who are donating their DNA to various research projects at a number of hospitals and labs.

There seems to be a group for almost every kind of disease, condition, or preference that has a genetic component. Many are support groups, others are more activism/advocacy focused, such as the “Cure Breast Cancer” group. Some people share information their personal treatment plans, and review doctors and hospitals – a sort of [www.ratemyprofessor.com](http://www.ratemyprofessor.com) for the medical world.

### Discussion points:

Would you join a Facebook group for “fast metabolizers” or “macular degenerators”? Do you have concerns about the impact of sharing your sequence data for each of these conditions? Is one riskier than the other to disclose? Why or why not?

How might your potential employers or health insurers use this information, which is easily found via a quick internet search? How do you balance the importance of social networking, research and activism with the importance of personal privacy?

Some people have posted their full sequence online, along with their names, medical history, health records, and photographs. They hope to contribute to basic research and feel that making their information freely available will enable the scientific community to make new connections between genes and traits. Would you consider this option for yourself?

## **Scenario F:**

You have just received a copy of your personal genome sequence. Included in the results is following piece of information:

You have a mutation on your APOE gene that is strongly associated with an elevated risk of Alzheimer's Disease. It is a progressive, degenerative disorder of the nervous system, and at present there are no clinically accepted treatments or cures. Carrying the APOE mutation does not 100% guarantee you will actually develop this typically late-onset disease.

You watched your grandmother suffer for many years with Alzheimer's, and it seemed that your family struggled to make sure she had the right medical attention and was living in a place where she could be safe and comfortable.

### Discussion points:

Will knowing about your risk for Alzheimer's change how you think about your future? How might it impact your plans for career, family, or travel? Maybe, or not at all?

You have decided to get sequenced right after college – you are 23 years old. Are you glad to have learned this information so young? How might it have been different if you were 63 when you learned about your genome sequence, as opposed to being in your early twenties?

Knowing this information now, do you have an obligation to share this information with family? What about someone you are seriously dating?

What are the reasons for wanting to know, versus never wanting to learn this kind of information?