8.1: Case Study: Genes and Inheritance

Case Study: Cancer in the Family

People tend to look similar to their biological parents, as illustrated by the family tree in Figure \(\PageIndex{1}\). But, you can also inherit traits from your parents that you can’t see. Rebecca becomes very aware of this fact when she visits her new doctor for a physical exam. Her doctor asks several questions about her family’s medical history, including whether Rebecca has or had relatives with cancer. Rebecca tells her that her grandmother, aunt, and uncle, who have all passed away, all had cancer. They all had breast cancer, including her uncle, and her aunt additionally had ovarian cancer. Her doctor asks how old they were when they were diagnosed with cancer. Rebecca is not sure exactly, but she knows that her grandmother was fairly young at the time, probably in her forties.

Rebecca’s doctor explains that while the vast majority of cancers are not due to inherited factors, a cluster of cancers...
within a family may indicate that there are mutations in certain genes that increase the risk of getting certain types of cancer, particularly breast and ovarian cancer. Some signs that cancers may be due to these genetic factors are present in Rebecca’s family, such as cancer with an early age of onset (e.g. breast cancer before age 50), breast cancer in men, and breast cancer and ovarian cancer within the same person or family.

Based on her family medical history, Rebecca’s doctor recommends that she see a genetic counselor because these professionals can help determine whether the high incidence of cancers in her family could be due to inherited mutations in their genes. If so, they can test Rebecca to find out whether she has the particular variations of these genes that would increase her risk of getting cancer.

When Rebecca sees the genetic counselor, he asks how her grandmother, aunt, and uncle with cancer are related to her. She says that these relatives are all on her mother’s side — they are her mother’s mother and siblings. The genetic counselor records this information in the form of a specific type of family tree, called a pedigree, indicating which relatives had which type of cancer and how they are related to each other and to Rebecca. He also asks her ethnicity. Rebecca says that her family, on both sides, are Ashkenazi Jews, meaning Jews whose ancestors came from central and eastern Europe. “But what does that have to do with anything?” she asks. The counselor tells Rebecca that mutations in two tumor-suppressor genes called BRCA1 and BRCA2, located on chromosome 17 and 13, respectively, are particularly prevalent in people of Ashkenazi Jewish descent and greatly increase the risk of getting cancer. About 1 in 40 Ashkenazi Jewish people have one of these mutations, compared to about 1 in 800 in the general population. Her ethnicity, along with the types of cancer, age of onset, and the specific relationships between her family members who had cancer indicate to the counselor that she is a good candidate for genetic testing for the presence of these mutations.

Rebecca says that her 72-year-old mother never had cancer, and nor had many other relatives on that side of the family, so how could the cancers be genetic? The genetic counselor explains that the mutations in the BRCA1 and BRCA2 genes, although dominant, are not inherited by everyone in a family. Also, even people with mutations in these genes do not necessarily get cancer — the mutations simply increase their risk of getting cancer. For instance, 55 to 65% of women with a harmful mutation in the BRCA1 gene will get breast cancer before age 70, compared to 12% of women in the general population who will get breast cancer sometime over the course of their lives.

Rebecca is not sure she wants to know whether she has a higher risk of cancer. The genetic counselor understands her apprehension but explains that if she knows that she has harmful mutations in either of these genes, her doctor will screen her for cancer more often and at earlier ages. Therefore, any cancers she may develop are likely to be caught earlier when they are often much more treatable. Rebecca decides to go through with the testing, which involves taking a blood sample, and nervously waits for her results.

Chapter Overview: Genetics

At the end of this chapter, you will find out Rebecca’s test results. By then, you will have learned how mutations in genes such as BRCA1 and BRCA2 can be passed down and cause disease. Especially, you will learn about:

- How Gregor Mendel discovered the laws of inheritance for certain types of traits.
- The science of heredity, known as genetics, and the relationship between genes and traits.
- Simple Mendelian and more complex non-Mendelian inheritance of some human traits.
- How biotechnology is used in medicine and agriculture, how it works, and some of the ethical issues it may raise.
- The human genome, how it was sequenced, and how it is contributing to discoveries in science and medicine.

As you read this chapter, keep Rebecca’s situation in mind and think about the following questions:

1. What do the BRCA1 and BRCA2 genes normally do? How can they cause cancer?

2. Are BRCA1 and BRCA2 considered linked genes? And are they on autosomes or sex chromosomes?

3. After learning more about pedigrees, draw the pedigree for cancer in Rebecca’s family. Use the pedigree to help you think about why it is possible that her mother does not have one of the BRCA gene mutations, even if her grandmother, aunt, and uncle did have it.

4. Why do you think certain gene mutations are prevalent in certain ethnic groups?