# AM I A CARRIER FOR A HEALTH CONDITION?

Exploring Carrier Screening Through Cystic Fibrosis

MINI-LESSON & TEACHER'S GUIDE

PERSONAL GENETICS EDUCATION & DIALOGUE

# STUDENT MATERIALS

Keith and Nyah are considering having a baby. They are feeling excited, especially since this would be their first child. They also have questions and concerns about how their genes might impact the health of their child. Although Keith is unaffected, his sister was born with cystic fibrosis. People with cystic fibrosis experience difficulty breathing and persistent lung infections, among other symptoms. Keith and Nyah wonder if their child might be at risk for developing cystic fibrosis. They read online that, for some people, their risk for passing on particular health conditions to their children can be detected before getting pregnant through a genetic testing process called carrier screening. But what exactly is carrier screening? And what are the potential outcomes?

## What Causes a Person to Develop Cystic Fibrosis?

Cystic fibrosis (CF) is a genetic condition caused by the inheritance of a gene variation that affects a person's **CFTR** protein. The CFTR protein is found in many places in the body, such as the heart and immune system. CFTR is most well-known for its role in managing a healthy layer of mucus and fluid in organs such as the lungs, digestive system, and sweat glands. The function of the CFTR protein can be affected by differences in the DNA of the gene that codes for this protein - the CFTR gene. We call these differences in the DNA code "**variants**". Some variants are more common than others, and some can affect a person's health.

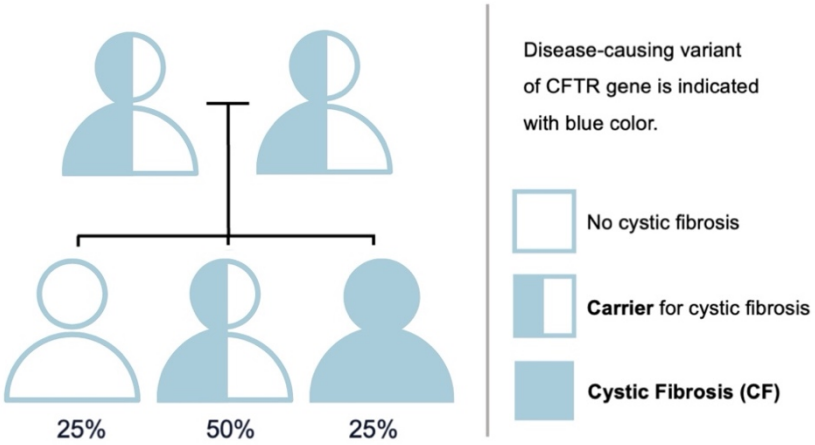


Figure 1: The Genetics of Cystic Fibrosis



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There are close to 2,000 known variants of the CFTR gene. These variants can be divided into two categories: those that cause disease ("disease-causing variant", indicated in blue in Fig. 1) and those that do not (indicated in white in Fig. 1). People have two copies of the CFTR gene, one inherited from each biological parent. With two copies of a gene, there are 3 different combinations of variants that a person may carry:

1. A person might carry 2 variants of the CFTR gene that do not cause disease. In this case, the body's CFTR protein can function correctly.

2. A person might carry two disease-causing variants of the CFTR gene. In this case, the body's CFTR protein does not work correctly. This affects many organs in the body, including causing thick and sticky mucus to develop in the lungs, resulting in the genetic condition we know as cystic fibrosis (CF).

3. A person might carry 1 disease-causing variant of the CFTR gene, and 1 variant that does not cause disease. People with this combination of CFTR variants typically do not have the symptoms of CF. However, because they do have 1 disease-causing variant in their DNA, these people are known as CF **carriers**.

Cystic fibrosis is an example of a **recessive** health condition. Recessive health conditions only develop in people who carry 2 disease-causing variants of the gene related to that condition. Whereas people who only carry 1 disease-causing gene variant of a recessive health condition typically do not have any symptoms and are referred to as 'carriers' of the trait.

Note: There are many different types of health conditions affected by genetics. For example, in contrast to recessive health conditions, there are **dominant** health conditions. For a dominant health condition, a person must carry only 1 disease-causing gene variant to experience the symptoms of that condition.

## What is Carrier Screening?

Since carriers of recessive health conditions generally do not experience symptoms, they are often not aware that they carry a disease-causing variant in their DNA. For example, when two CF carriers decide to have a child, there is a chance that their child will inherit 2 disease-causing variants - one from each parent - and will therefore develop CF (even though neither of the parents has CF, themselves) (Fig. 1).

**Carrier screening** is a type of genetic test that is performed to identify whether a person carries recessive disease-causing variants in their DNA. People might do carrier testing once they have decided to have children, to better understand their risk of having a child with a genetic health condition. People who have a known family history of a recessive health condition might be especially motivated to pursue carrier screening before trying to get pregnant.

Once a couple has decided that they wish to pursue carrier screening, they will meet with a healthcare provider to initiate the process. The provider will consult the couple before they proceed with the screening to ensure that they understand the meaning of potential results, including risks and benefits, and that they are equipped to use the information they receive in a way that best suits their needs.

Upon giving informed consent, the patient can provide a sample for testing. This can be a blood, saliva, or tissue sample from their inner cheek. The person's DNA can then be extracted from the sample to identify their genetic variants. Different options for carrier testing are available, from tests that just look at a single recessive health condition (such as CF), to ones that test for hundreds of different conditions. Healthcare providers can help a parent-to-be decide on the right test for them and/or their partner. After receiving the test results, the provider will meet again with the couple to discuss the results and their next steps.

## What are the Outcomes of Carrier Screening?

Once the parents' gene variants are known, the likelihood of having a child who is affected by, unaffected by, or a carrier of, a genetic health condition can be determined. For example, when both parents are CF carriers, they have a 25% chance



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of having a child who is unaffected by CF, a 25% chance of having a child with CF, and a 50% chance of having a child who is also a CF carrier (Fig. 1). There are many different ways in which people can use this information to make health and family-planning decisions.

Some people do carrier screening before they are pregnant to make informed decisions about their plans to get pregnant. Depending on the carrier screening test results, people may decide to not have children, to have children without medical assistance, or to have children using medical assistance that allows them to lower their chances of having a child with a genetic health condition. Other people choose to do carrier screening during their pregnancy. Obtaining this information during pregnancy enables couples to make preparations in the case that their child is born with a genetic health condition. This may include preparing for special medical care and social service needs. If one or both parents are found to be carriers, they might choose to pursue follow-up testing to determine if they did indeed pass on one or two disease-causing variants to their future child. Choosing when to be tested and what to do with the results are individualized decisions. Couples should make the decision that best suits their unique needs.

# STUDENT WORKSHEET

**Name:**_____ **Date:**_____

Answer the following reflection questions to demonstrate your understanding of what it means to be a carrier for a health condition and the process of carrier screening.

1. Cystic fibrosis is an example of a recessive health condition. In your own words, explain what a recessive health condition is.

2. In the context of a recessive health condition, explain what a 'carrier' is.

3. Why are people often unaware that they are carriers for certain genetic health conditions?

4. Before initiating the carrier screening process, a patient will discuss the risks and benefits of screening with a healthcare provider. With this knowledge, a patient can decide to give their informed consent to move forward with finding out whether they carry any disease-causing variants. Please describe what happens next. How are these carrier screening results obtained?

5. Give two examples of a health and/or family planning-related decision that a person might make based on their carrier screening test results.

# TEACHER'S GUIDE

## Related PGED Resources

PGED offers several educational resources that can be utilized in a classroom setting to foster learning about genetics and health. You can find them in the resource hub at www.pged.org. Here are some highlighted resources:

- *Reproductive Genetic Testing: Technology, Access, and Decision-Making*: This lesson addresses major innovations in reproductive genetic technologies that are being used by individuals who, for a variety of reasons, wish to know and/or make decisions related to the genetic makeup of their children. The goal of this lesson is to allow students to discuss many aspects of reproductive genetic testing such that they become aware of the diversity of opinions surrounding the use of these technologies.

- *The Many Faces of Sickle Cell Disease*: Sickle Cell Disease (SCD) is a recessive genetic condition caused by a common gene variant that affects a person's red blood cells. This mini-lesson explores why the variant is more common in people with ancestry from certain parts of the world, causing some populations to have larger numbers of carriers and individuals affected by the condition.

## Additional Resources for Teachers

The Washington University Physicians Fertility & Reproductive Medicine Center's Genetic Carrier Screening resource provides answers to commonly asked questions about the decision to pursue carrier screening, the screening process, and the potential outcomes.

This resource may be especially useful if you are looking for information about carrier screening that goes beyond what is discussed in this mini-lesson, such as a more in-depth discussion of the potential downsides to carrier screening and the health implications that the results might reveal for the person being tested.



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# STUDENT WORKSHEET: TEACHER NOTES

1. Recessive health conditions only develop in people who carry 2 disease-causing variants of the gene related to that condition.

2. People who only carry 1 disease-causing gene variant of a recessive health condition are referred to as 'carriers' of the trait.

3. Because carriers of recessive health conditions typically do not have symptoms, they are often not aware that they carry a disease-causing variant in their DNA.

4. Upon giving informed consent, the patient can provide a sample for testing. This can be a blood, saliva, or tissue sample from their inner cheek. The person's DNA can then be extracted from the sample to identify their genetic variants.

5. There are many different ways in which people can use this information to make health and family-planning decisions. Some people do carrier screening before they are pregnant to make informed decisions about their plans to get pregnant. Depending on the carrier screening test results, people may decide to not have children, to have children without medical assistance, or to have children using medical assistance that allows them to lower their chances of having a child with a genetic health condition. Other people choose to do carrier screening during their pregnancy. Obtaining this information during pregnancy enables couples to make preparations in the case that their child is born with a genetic health condition. This may include preparing for special medical care and social service needs. If one or both parents are found to be carriers, they might choose to pursue follow-up testing to determine if they did indeed pass on one or two disease-causing variants to their future child. Choosing when to be tested and what to do with the results are individualized decisions. Couples should make the decision that best suits their unique needs.

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