

2.7 Case Study

Genetic Diseases: Cystic Fibrosis

Chromosomes carry many genes. Human chromosomes have over 25,000 genes. Some genes carry information for good traits. Some genes carry information for bad traits, such as genetic diseases.

Some genetic diseases can be caused by a dominant allele. If a genetic disease is represented by the letter “A,” a child needs only one allele, the dominant one (genotype AA or Aa) to have the disease. Only people with the genotype “aa” would not be affected by the disease.

But most genetic diseases in humans are recessive. This means that both the chromosomes a baby receives from its parents must contain the recessive allele for the disease. If a genetic disease is represented by the letter “A,” a normal person without the disease would have the genotype “AA.” A normal person who carries the disease would be “Aa.” A person with the disease would be “aa.”

Since the chromosomes move randomly into the offspring during reproduction, there is no way to control which alleles a child will receive. Normal people can be tested for some genetic diseases, to see if they are a carrier of the recessive allele.

Both plants and animals can carry the genes for genetic diseases. Information about genetic diseases is important when developing a new rice plant.



cystic fibrosis:

an inherited disease that causes the body to produce thick, sticky mucus in the lungs and digestive system.

mucus:

a secretion of the body.

chest**physiotherapy:**

a treatment used for removing the thick mucus that forms in the lungs of a person with CF.



Derek has cystic fibrosis and is able to do most of the things every middle school student does.

Hello,

My name is Derek. I am 10 years old, and I like to toss the Frisbee high in the sky and catch it. I enjoy playing a lot of games that all kids play. I have **cystic fibrosis** (CF), a disease that affects the lungs and digestive system. Living with cystic fibrosis is one of my greatest challenges. Because of the thick **mucus** in my lungs, I need at least an hour of breathing treatments and **chest physiotherapy** every day. Chest physiotherapy is a treatment used to remove the mucus. To digest food, avoid lung infections, and thin the mucus in my lungs, I take over 30 pills each day. I also get fed overnight with a tube that carries food directly to my stomach. It's a pain. Sometimes people wonder why I cough so much and eat so much. I would destroy CF if I could, but at least all these therapies help me to grow, play, and learn.

Your pal, Derek

mutation:
changes to
genetic material
of an organism.

*My son, Derek, has cystic fibrosis. We found out he had it when he was only 3 months old. Cystic fibrosis is a genetic condition that affects over 30,000 people in the United States. CF is a recessive trait. One out of every 30 Americans is a carrier of the defective cystic fibrosis allele but do not have any symptoms of the disease. CF results from a **mutation**, a change in the information in a gene. A person having one normal CF allele and one mutated CF allele has no health problems. When a baby has two defective alleles, one from each parent, the child has cystic fibrosis.*

People who are affected by CF have a variety of symptoms. These can include difficulty breathing and digesting food. Various therapies treat the complications in the lungs and digestive tract caused by cystic fibrosis. The therapies do not relieve the CF patient of his or her symptoms.

Researchers continue to seek the cure for cystic fibrosis. They have identified the gene that causes cystic fibrosis. It is called the CFTR gene. The CFTR gene is large and complex. For this one gene, there are over 1000 different mutations. As researchers get closer to understanding the gene and how it works, they will be more likely to find a way to cure this disease.

Derek and his mom frequently give presentations about cystic fibrosis. They teach others about how Derek lives with cystic fibrosis.



Huntington's disease: a fatal genetic disease caused by a dominant allele, which affects the nervous system.

neurofibromatosis: a genetic condition that causes changes in skin coloring (pigmentation) and the growth of tumors along nerves in the skin, brain, and other parts of the body.

adult leukemia: a genetic disease that causes an increased risk of cancer in young adults.

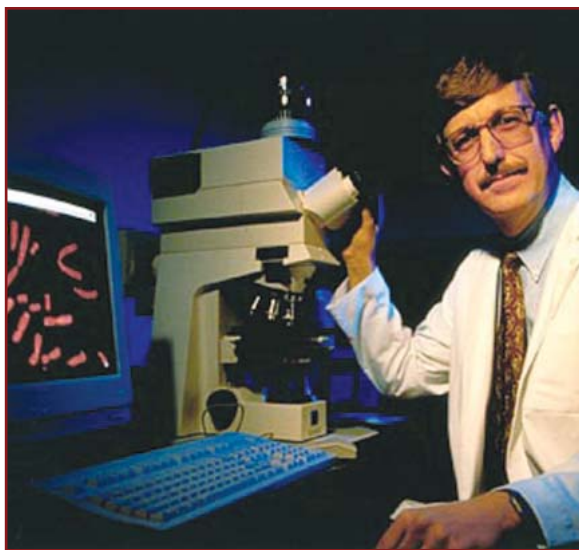
mapping (genes): finding out the exact genes and their locations on a chromosome.

genomics: the science of mapping genes.

Tay-Sachs disease: a fatal genetic disease caused by a dominant allele, which affects the nervous system.

Francis Collins has spent his life working to find the genetic causes of a variety of diseases. He discovered his love of biology after years of being interested in chemistry. While he was working at The University of Michigan, he became known as a gene hunter. In addition to finding the gene for CF, Dr. Collins found the genes for **Huntington's disease**, which affects the nervous system, **neurofibromatosis**, which causes tumors, and **adult leukemia**, which causes cancer. You might want to research some of these genetic diseases.

Dr. Collins left The University of Michigan in 1993 and became the director of the National Center for Human Genome Research. He is the head of the Human Genome Project. His teams are responsible for finding and **mapping** all the genes that make up humans. Mapping involves finding out the exact genes and their locations on a chromosome. Dr. Collins is a leading researcher in a new field of science, begun by the Human Genome Project, called **genomics**, the science of mapping genes. You will learn more about the Human Genome Project later in this Unit.



Other Genetic Diseases

Huntington's disease is a brain disorder that causes uncontrolled movements, mental and emotional problems, and loss of thinking ability. Huntington's disease generally appears in a person's thirties or forties. By then, the person may have already had children and passed the gene on to the next generation. People with Huntington's disease may have trouble walking, speaking, and swallowing. They may also have changes in personality and trouble with thinking and reasoning. The person with this disease usually survives about 15 to 25 years after signs and symptoms begin.

Tay-Sachs disease is a rare inherited disorder that destroys nerve cells in the brain and spinal cord. The most common form of Tay-Sachs disease begins in infants. Infants with this disorder appear normal until the age of 3 to 6 months. Then their development slows and the muscles used for

movement become weak. Infants with Tay-Sachs lose motor skills, such as turning over, sitting, and crawling. Later, children with Tay-Sachs can have seizures, vision and hearing loss, mental retardation, and paralysis.

Sickle-cell anemia affects the blood cells that take oxygen to the body cells. The red blood cells develop a sickle, or crescent, shape. Signs and symptoms of sickle cell disease begin in early childhood. Children have a low number of red blood cells (**anemia**), infections, and pain. The anemia can cause shortness of breath, tiredness, and slow growth and development. The rapid breakdown of red blood cells may also cause yellowing of the eyes and skin, which are signs of **jaundice**. Pain can occur when sickled red blood cells, which are stiff, get stuck in small blood vessels. Sickle-cell anemia can cause high blood pressure and can lead to heart failure.

Reflect

1. Cystic fibrosis is a recessive trait. A person with CF would have two recessive alleles for this trait (cc). Using a Punnett square, show what would happen if two parents with the recessive trait but no CF symptoms (Cc) had offspring. What percent of their offspring are likely to get cystic fibrosis?
2. Using the Punnett square again, what would happen if one parent with cystic fibrosis (cc) and a parent with the recessive trait but no symptoms (Cc) were to have offspring? What percent of the offspring would be likely to get CF? What percent of the offspring would not get CF?
3. Derek has cystic fibrosis. Even though he is sick and has to have chest physiotherapy and medication every day, he still does the things other kids do. If you met Derek, what would you ask him about living with CF? Would you ask him what sports he likes to play? What other things might you ask him?

Update the Project Board

You know that reproduction can produce genetic variation. You also learned how genes can cause disease. Record the new science you learned in the *What are we learning?* column of your *Project Board*. Support what you have learned with evidence in the *What is our evidence?* column. The activity you carried out in the last section and the case study you read may have made you think you know more about genes, chromosomes, and how traits are passed from parents to offspring. Record what you think you know in the *What do we think we know?* column. You may have ideas about new investigations you would like to conduct to explore reproduction and

sickle-cell anemia: a genetic disease, carried by a recessive allele, that affects the ability of the blood cells to carry oxygen.

anemia: a low number of red blood cells, which carry oxygen in the blood to the body cells.

jaundice: a condition when pigments from the gall bladder invade the blood. The skin and eyes become yellow.

variation. Record your ideas for further investigations in the *What do we need to investigate?* column. Your teacher or a student will update the class *Project Board*.



What's the Point?

You read about genetic diseases in a case study on cystic fibrosis. You learned that genetic disease can be caused by a recessive allele on a chromosome or a dominant allele on a chromosome. It is impossible to control which chromosomes a child receives during reproduction, because the chromosomes move randomly into the new organism.

Since both plants and animals can carry the genes for genetic diseases, you must keep this information in mind as you develop a new rice plant.

