**Chapter 11 – Single-Gene Inheritance and Meiosis**

***Rock for a Cause: From patient to performer shining the spotlight on a genetic disease***

**Driving Questions**

1. How does the organization of chromosomes, genes, and their alleles contribute to human traits?
2. How does meiosis produce gametes?
3. Why do different traits have different inheritance patterns?
4. What are some practical applications of understanding the genetic basis of human disease?

**Story Abstract & Additional Information**

This chapter follows Emily Schaller, a young woman with cystic fibrosis. Using her story and cystic fibrosis as its framework, the chapter discusses gene inheritance and how two outwardly healthy people can carry and pass on an allele for disease.

Here are some of the key points in the story for this chapter.

**Cystic Fibrosis**

* Cystic fibrosis (CF) has many symptoms, the most dangerous of which is mucus that clogs airways in the lungs and makes it difficult to breathe. People with CF also can’t digest food well—mucus blocks the passageways through which the necessary enzymes travel to the intestines. So they must swallow enzymes before each meal to ensure their body gets enough nutrients.

<http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001167/>

* Approximately 2,500 babies are born with CF every year, making it the most common fatal genetic disease in the United States. In 1989, a team of scientists led by Lap Chee Tsui, at Toronto’s Hospital for Sick Children, and Francis Collins, then at the University of Michigan, discovered that the disease is caused by genetic mutations in a specific gene that sits on chromosome 7.
* Today, scientists understand the disease better, and this has led to better drugs and therapies to treat symptoms; victims of CF are living longer than ever. But despite scientific advances, there is still much to learn. One aspect of the disease that scientists are studying intensively is that people with identical CF alleles vary in the course of their disease—some have worse symptoms and live shorter lives than others. In recent years, scientists have discovered that there are other genes that contribute to a patient’s overall health—so-called modifier genes. That discovery is leading to exciting new therapies that may extend the lives of thousands of people with CF.

**The Risk of Passing on a Genetic Disease**

* Couples who carry disease genes need not feel that having children is a roll of the dice. There are ways to ensure that their children won’t develop the diseases they could otherwise inherit. Many couples in this situation use a technology called pre-implantation genetic diagnosis to detect and select embryos that do not carry defective alleles. Through in vitro fertilization, a man’s sperm can fertilize a woman’s egg outside the body. The genes of each resulting embryo are then examined for specific alleles, and then only embryos that don’t contain defective alleles are implanted into the mother. Hundreds of thousands of babies have already been born by this technique.
* Some couples, however, may choose not to undergo assisted reproduction because of religious or other reasons. In the case of CF, there are new treatments in the pipeline that could help Emily, and her children and grandchildren, too. Furthest along are a class of medications that, when inhaled, can restore the balance of ions inside affected cells. Scientists are presently testing at least six different experimental drugs in humans.

**Emily Schaller**

* Emily is the founding president of the Rock CF Foundation, a nonprofit company dedicated to helping people with CF. The Foundation uses the arts, entertainment, fashion, and fitness to support research initiatives and heighten public awareness in the fight against CF.

<http://www.letsrockcf.org/>

* Hellen was the Detroit-based punk/classic rock band that Emily Schaller and four of her friends founded: Emily played the drums. In 2004, she and her brother organized the first benefit concert for CF. Called “Just Let Me Breathe,” it featured four Detroit bands. The concert sold out and raised about $9,000. She now has a $20,000 sponsorship from New York-based *Spin* magazine for Rock CF Foundation’s annual benefit concert. To date, the concert series has raised over $60,000, and she has raised over $150,000 through all of her fundraising activities.

**Additional information about other topics from this story and chapter includes:**

Read more about cystic fibrosis by visiting the below links.

<http://www.cff.org/>

<http://www.nhlbi.nih.gov/health/dci/Diseases/cf/cf_what.html>

<http://www.mayoclinic.com/health/cystic-fibrosis/DS00287>