Am I a Carrier for Cystic Fibrosis?

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Primary Learning Outcomes
After completing this lesson, students will have a better grasp of genetic terminology such as genotype, phenotype, carrier, homozygous, heterozygous, autosomal, dominant, and recessive. Students will also become more competent in using Punnett squares to predict traits in offspring (cystic fibrosis (CF) status in this case). Finally, after experiencing the difficulty of making a genetic decision, students will become more aware of the complexity of such bioethical decision making.

Additional Learning Outcomes
Students will increase their awareness of such birth defects as CF.

Materials
1. For Simulated CF Testing:
    a. Containers such as small test tubes, plastic medicine cups, or small drinking cups.
    b. Distilled water
    c. 0.1 M NaOH or other weak base
    d. Phenolphthalein
2. Handouts
3. Computers with Internet access (optional)
4. Teacher presentation station

Technology Connection
Students may use the Internet to research CF individually or as a whole group by going to the sites during class and using an LCD projector to project the information on a screen for all to see.

Total Duration
1 hour, 20 minutes

Procedures

Step 1 **Duration: 40 minutes**

Teacher Preparation
View the U.S. Army Medcom Quality Management Office's PowerPoint in the Web resource to become familiar with the genetic disorder, Cystic Fibrosis (CF), how CF is inherited, and details on the carrier testing that is being offered to couples planning to have children.

Prepare for simulated testing by filling small test tubes, plastic medicine cups, or small drinking cups with distilled water (only need a small amount of liquid). Add a few drops of a weak base such as 0.1M NaOH to half of the containers (these will test "positive"). Place a new label over a dropper bottle of phenolphthalein that says "Cystic Fibrosis Carrier Testing Solution."

Download and print the "Cystic Fibrosis Fact Sheet" in Step 2. Make a classroom set of copies

as they can be reused for multiple class periods.

Download and print the "Pretesting Survey" in Step 3 and the "Post Test Form" in Step 4. You will need one copy per two students. Also download the rubric attached in the assessment field (one per student).

Web Resource
Title: U.S. Army Medcom Quality Management Office Uncomplicated Pregnancy Page
URL: http://www.qmo.amedd.army.mil/pregnancy/uncompreg.htm
Description: Click on "Patient Education PowerPoint Presentation with Audio Voice-Over" in the right hand column to view the information on CF. This is a very detailed PowerPoint created by the U.S. Army Medcom Quality Management Office to be used for patient education. It covers all aspects of CF, focusing on the issue of carrier testing. This could even be shown to the students.

Step 2 **Duration: 20 minutes**

Introduction

Announce and/or display the following message: The State Department of Disease Control and Prevention has asked that all adolescents be tested to determine whether or not they are a carrier for CF, a genetic disorder.

Discuss the following questions with students:

1. **What do you already know about Cystic Fibrosis (CF)?**
   Expect students to share a range of responses from knowing nothing to knowing quite a bit.

2. **What would you like to know?**
   After they share this with you, provide students with the "Cystic Fibrosis Fact Sheet" provided by the National Heart, Lung, and Blood Institute and allow them time to skim through the information. If you would like a shorter fact sheet, use the March of Dimes Web resource "Facts about Cystic Fibrosis." If your students have easy access to the Internet, you could provide them with the Web resources: "Cystic Fibrosis Foundation" homepage and the National Center for Biotechnology Information's "Cystic Fibrosis Reference."

3. **What is CF and is there a cure?**
   CF is a life-long disease of the body's mucous glands. The lungs are affected by thick, clogging mucous, frequent coughing and wheezing, frequent pneumonia and bronchitis. Individuals with CF also have chronic sinus infections, digestion problems, males are infertile and females may be less fertile than women without CF. (Source: PowerPoint from Step 1) Accept any response that is found in the fact sheet. Be sure to discuss that it is a genetic or inherited disorder.

4. **What is a carrier?**
   An individual that does not have the disease but carries one mutated CF gene along with one normal gene.

5. **What is the genotype of a carrier?**
   The genotype of the carrier is Ff (one dominant non-disease gene, F, and one recessive, CF gene, f). Of course, any letter of the alphabet could be used as long as it is expressed as a heterozygous genotype.

Web Resources
Title: Cystic Fibrosis Foundation
URL: http://www.cff.org/
Description: This homepage for the Cystic Fibrosis Foundation includes links to information, research, case studies, and testing.

Title: Facts of Cystic Fibrosis
URL: http://www.marchofdimes.com/professionals/681_1213.asp
Description: This March of Dimes site is an excellent overview of Cystic Fibrosis as well as information about the availability of carrier testing.

Title: Cystic Fibrosis Reference
URL: http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.section.168
Description: This site provides a brief summary with genetics links from National Center for Biotechnology Information "Genes and Diseases" site.

Supplemental Document
Title: Cystic Fibrosis Fact Sheet
File Name: Cystic_Fibrosis_Fact_Sheet.pdf
Description: This document is a detailed six-page fact sheet with up-to-date information about Cystic Fibrosis, including carrier and prenatal testing. It is provided by the National Heart Lung and Blood Institute.

Step 3 **Duration: 30 minutes**
Testing
Partner students and have one be the husband and one be the wife. If there are equal numbers of girls and boys you can partner one girl with every boy; however, this is usually not the situation and so any two students can partner. A random way of assigning partners is to take a deck of cards and choose enough hearts to equal half of your class. Then choose the matching clubs, mix them up, and have students draw a card. They will find the other student with the same number and that will be their partner.

Invite students to make predictions about whether they will be carriers or not. Ask each pair of students to complete the "Pretesting Survey and Genetic Inheritance Review".

When the survey is completed, the students may go to the testing station to find out if they carry one copy of the CF gene. Students will randomly choose a small test tube of clear fluid (all have distilled water, half of them with weak NaOH added to it) to represent their blood plasma and add one drop of CF Carrier Testing Solution (phenolphthalein) to it. If the solution turns pink, the student is a carrier. If it stays clear, the student does not carry the gene. Be sure to let students know that this test is simulated and not the same procedure that is performed for genetic testing in real life.

Students will then return to their desks to record their results and discuss the results with their partner. Answers for the worksheet are found in the “Prettesting Survey and Genetic Inheritance Review – Answer Key”.

Supplemental Documents
Title: Pretesting Survey and Genetic Inheritance Review
File Name: Pretesting Survey and Genetic Inheritance Review.doc
Description: This form calls on students to review genotype and inheritance information

related to Cystic Fibrosis, an autosomal recessive disorder, prior to the simulated testing to see if they are a carrier for it.

Title: Pretesting Survey and Genetic Inheritance Review – Answer Key
File Name: Pretesting Survey and Genetic Inheritance Review – Answer Key.doc
Description: This teacher answer sheet contains an answer to each question in the Pretesting Survey, including completed Punnett squares.

Step 4 **Duration: 20-30 minutes**

Posttest Discussion

Record results on the attached "Posttest Form" (both genotype and phenotype)and launch a large group discussion about what this means for couples who are "at risk" for having a child with CF. The "Posttest Form" includes most of the options to guide the discussion. Be sure to reinforce to the students that actual carrier testing requires a blood sample from which the DNA is tested.

After discussing the many options available to a couple at risk, have the partners assume that they too are at risk (despite testing results) and complete the decision making matrix on the "Posttest Form" to arrive at a decision. When all couples have completed this, invite students to share their thoughts about the difficulty involved with reaching this decision. To save on class time, copies of the "Posttest Form" could be made available to each student to complete individually as homework. Then, the students could be given a few minutes in class the following day to compare notes and agree on a final decision.

Discussion with students

1. **Why would couples want to pursue CF screening?**
   For reproductive options. For example, a couple with at least one affected (CF) child together may want to know if a subsequent pregnancy is similarly affected.

2. **Does it truly make sense to screen everyone for CF as we are doing in our classroom?**
   It probably does not make sense because the prevalence is low. Screening could be expensive and time consuming, No test is 100 percent correct and thus with large screenings there is a greater chance for inaccurate tests.

3. **What would make more sense?**
   One possibility is testing women wanting to have children and then, if they are carriers, testing their partners.

4. **As biotechnology develops, new options become available. Ask the students if they would change their decision if gene therapy was made widely available as a cure for CF.**
   Answers will vary.

5. **End by asking students to brainstorm other situations in which difficult decisions, one for which there is no right answer, must be made.**
   Answers will vary.

The “Cystic Fibrosis Carrier Testing Rubric” is designed to evaluate student performance on both the "Posttest Form" and meaningful participation in class discussion about carrier testing.

Supplemental Documents
Title: Posttest Form
File Name: Posttest Form.doc
Description: This form has an area to record the genotype and phenotype results of the testing for both the husband and wife. This is followed by the available options and a decision making matrix for arriving at a decision.

Title: Cystic Fibrosis Carrier Testing Rubric
File Name: Cystic Fibrosis Carrier Testing Rubric.doc
Description: This is a detailed rubric to assess the whole carrier testing experience from the time of receiving the test results to the decision about what to do with the new information. This rubric assesses the completion of the "Post Test Form" as well as student participation in class discussion.

Assessment

Students should be assessed on the quality/accuracy of their "Pretesting Surveys" (an answer key is provided in Step 1). Due to its divergent nature, there is no actual key for assessing the "Posttest Form." However, a "Cystic Fibrosis Carrier Testing Rubric" is included in Step 5 that is designed to evaluate student performance on both the "Post Test Form" and meaningful participation in class discussion about the whole carrier testing issue. The rubric emphasizes that any decision made by the "couple" is acceptable as long as both individuals agree upon it and can clearly explain their reasons. In addition, student discussion and written explanations should provide evidence that the students have struggled with their decision and realize that such decisions are not "cut and dried" or easy to make.

Modifications

Extension
Further research ideas:

1. What protein is coded for by the CF gene and how does it result in the observed characteristics of CF disease?

2. Research another genetic disorder in which genetic screening or testing is available. Create a pamphlet that could be used to educate the general public about the availability and issues surrounding being tested.

The web resource provides a place for students to start pursuing both of these questions.

Web Resource
Title: National Center for Biotechnology Information's Genes and Disease Search Page
URL: http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowSection&rid=gnd.preface.91
Description: This site provides a great starting point to researching genetic diseases.

Remediation

1. Have students partner with students that understand the important concepts, encouraging peer teaching.
2. Have students visit an online tutorial (see Web resource) for help and practice with genetics concepts.

Web Resource
Title: The Biology Project Mendelian Genetics Tutorial
URL: http://www.biology.arizona.edu/mendelian_genetics/problem_sets/monohybrid_cross/01t.html
Description: This tutorial provides practice in monohybrid cross problems.

Education Standards

National Science Education Standards
LIFE SCIENCE, CONTENT STANDARD C:
As a result of their activities in grades 9-12, all students should develop understanding of

- The cell
- **Molecular basis of heredity**
- Biological evolution
- Interdependence of organisms
- Matter, energy, and organization in living systems
- Behavior of organisms

SCIENCE IN PERSONAL AND SOCIAL PERSPECTIVES, CONTENT STANDARD F:
As a result of activities in grades 9-12, all students should develop understanding of

- **Personal and community health**
- Population growth
- Natural resources
- Environmental quality
- Natural and human-induced hazards
- Science and technology in local, national, and global challenges

Georgia State Science Standards
Grade: 9-12, Science, Biology 10
Topic: Genetics (Mendelian Genetics)
Standard: Explains and uses the basic Mendelian genetic principles.

Grade: 9-12, Science, Biology 11
Topic: Genetics (Patterns of Inheritance)
Standard: Describes patterns of inheritance and genetic engineering.

Grade: 9-12, Healthcare Science Technology, Introduction to Public Health 81
Topic: Prevention
Standard: Explains the difference between primary, secondary, and tertiary prevention of disease and provide examples of each.

Grade: 9-12, Healthcare Science Technology, Introduction to Public Health 84
Topic: Prevention
Standard: Explains the various approaches for controlling the incidence of non-communicable diseases.

Pretesting Survey and Review of Genetic Inheritance

Am I a Carrier of Cystic Fibrosis?
Julia Koble, CDC's 2004 Science Ambassador Program

Name of Wife: _____

Name of Husband: _____

Genotypes

1. If we use **f** to represent the recessive gene for Cystic Fibrosis (CF), what is the *genotype* of individuals that do **not** have CF? _____

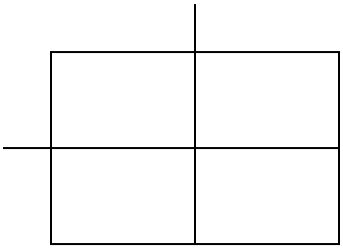
2. What is the *genotype* of a carrier? _____

3. What is the *genotype* of an individual that has CF? _____

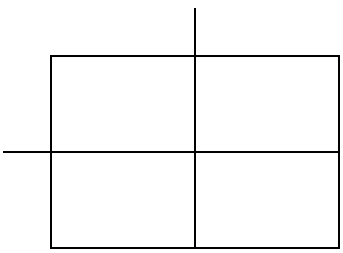
Probability

Please complete the Punnett squares to support your answer.

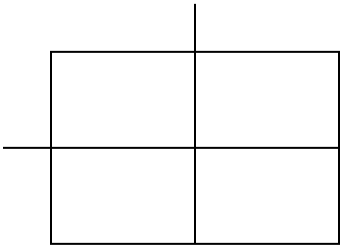
4. Do you and your spouse need to be concerned about the possibility of having a child with CF if neither of you carry the gene for the disease?_____ Explain.



5. Do you need to be concerned if just one of you carries the gene? _____ Explain.



6. Do you need to be concerned if both of you carry the gene? _____ Explain.



7. What is the chance of having a child with CF if both are carriers? _____

Pretesting Survey and Review of Genetic Inheritance – Answer Key

Am I a Carrier of Cystic Fibrosis?
Julia Koble, CDC's 2004 Science Ambassador Program

Name of Wife: _____

Name of Husband: _____

Genotypes

1. If we use **f** to represent the recessive gene for Cystic Fibrosis (CF), what is the *genotype* of individuals that do **not** have CF? **FF**

2. What is the *genotype* of a carrier? **Ff**

3. What is the *genotype* of an individual that has CF? **ff**

Probability

Please complete the Punnett squares to support your answer.

5. Do you and your spouse need to be concerned about the possibility of having a child with CF if neither of you carry the gene for the disease? **No.** Explain.

	F	**F**
F	**FF**	**FF**
F	**FF**	**FF**

The parents only have dominant, non-CF genes to pass on and thus 100% of the offspring will have non-CF genes.

7. Do you need to be concerned if just one of you carries the gene? **No.**

Explain.

	F	**F**
F	**FF**	**FF**
f	**Ff**	**Ff**

There is no change of having a child with CF; however, each child has a 50% change of being a carrier.

8. Do you need to be concerned if both of you carry the gene? **Yes.** Explain.

	F	f
F	FF	Ff
f	Ff	ff

Each carrier has a 50/50 chance of passing their CF gene onto their offspring which results in ¼ chance that one child will get both CF genes.

9. What is the chance of having a child with CF if both are carriers? **¼ or 25%**

Posttest Form

Am I a Carrier for Cystic Fibrosis?
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Name of **Wife**: _____

Genotype of Wife (please circle one): FF Ff ff

Phenotype of Wife (please circle one): Homozygous Dominant Carrier CF

Name of **Husband**: _____

Genotype of Husband (please circle one): FF Ff ff

Phenotype of Husband (please circle one): Homozygous Dominant Carrier CF

Discussion Questions

1. Are the two of you at risk for having a child with Cystic Fibrosis (CF)? _____ Please Explain.

2. If you are at risk, what is the chance that your child will be born with CF?

Note: Testing cannot say 100% you are not a carrier; Testing itself does not reduce your change of being a carrier. It gives you knowledge you might want to make a decision. (1)

Both Parents are Carriers, Now What?

Assume that both you and your spouse are at risk for having a child born with CF. What choices do you now have in regards to having children?

1. Choose not to have a biological child
    a. Remain childless
    b. Adoption
2. Choose to have a child
    a. Take your chances that the child will not have CF (75%)
    b. Donor sperm or egg from a non-carrier
    c. **Pregnancy and Prenatal testing**: Test the developing fetus to see if the child has CF; if it does, take the time to prepare for such a child or potentially terminate the pregnancy (a very difficult moral decision).

i. **Amniocentesis:** Under ultrasound guidance, a thin needle is inserted through the mother's abdomen to take cells from the fluid surrounding the baby and test them for the presence of the CF gene. Usually performed in the 2nd trimester of pregnancy (15-20 weeks from the first day of the last menstrual period). The test brings with it a rare risk for miscarriage (one in 200-300 cases). (1)

ii. **Chorionic Villi Sampling (CVS):** Under ultrasound guidance, a thin needle is inserted through the mother's abdomen or a thin tube is inserted through the vagina to take cells from that tissue that will eventually form the placenta and test them for the presence of the CF gene. Is generally performed 10-13 weeks after the first day of the pregnant woman's last menstrual period. The test itself brings the risk (one in 100-200 cases) of miscarriage. (1)

d. Preimplantation Genetic Diagnosis (PGD): Embryos are created through in vitro fertilization techniques. A biopsy is conducted on a single cell per embryo for genetic diagnosis. If the embryo does not have CF, it can be implanted in the mother's uterus. (2)

References

1. Olney, Richard, and Cheng, Sabrina. 2004. *Genetic Screening* PowerPoint presentation. Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities.

2. ReproGenetics. *What is PGD?* Cited 2004 July 7. Available at URL: http://www.reprogenetics.com/patientstudies.html

Additional Resources

Army Medcom Quality Management Office for Patient Education. *Genetic Screening for Cystic Fibrosis: A New Choice for You and Your Pregnancy.* Cited 2004 July 7. Available at URL: http://www.qmo.amedd.army.mil/pregnancy/uncompreg.htm.

March of Dimes. *Facts about Cystic Fibrosis.* Cited 2004 July 7. Available at URL: http://www.marchofdimes.com/professionals/681_1213.asp.

Final Decision
Use the decision-making matrix on the next page to discuss the pros and cons of the above options with your class and/or partner. Then, please agree on a decision. In other words, what will you and your partner choose to do? Record your decision below the matrix.

Options for Couples when both are Carriers for Cystic Fibrosis

Choice/Option	Pros	Cons	Why acceptable/unacceptable to us
Remain Childless			
Adoption			
Take your chances			
Donor sperm or egg			
Pregnancy and amniocentesis			
Pregnancy and CVS			
Preimplantation Genetic Diagnosis			

Our Decision

_____
_____

Why?

_____
_____
_____

Rubric for Cystic Fibrosis Carrier Testing

Am I a Carrier for Cystic Fibrosis?
Julia Koble, CDC's 2004 Science Ambassador Program

(insert name of class here) **Name**_____
Hour 1 2 3 4 5 6 7

	10 8	6 4	3 2
Test Results *Did you correctly identify your CF status on the Post Test Form?*	Student accurately identifies his/her genotype and phenotype on the *Post Test Form*.	Either the genotype or phenotype is incorrect.	Neither the genotype or phenotype is identified correctly.
	10 8	6 4	3 2
Meaning of Results *Are you and your partner at risk for having a child with CF?*	Student correctly identifies whether his/her partnership is at risk for having a child with CF and, where applicable, expresses the actual chance of having a baby with CF.	Student correctly identifies whether his/her partnership is at risk for having a child with CF but, where applicable, incorrectly expresses the actual chance of having a baby with CF.	Incorrectly identifies risk.
	10 8	6 4	3 2
Decision Making Matrix *What are our options if we are both carriers?*	Student completes the decision making matrix completely. The pros and cons listed are accurate/realistic and 1 or 2 reasons per option are included for "Why unacceptable/ acceptable to us." There are no correct reasons here, but responses should include facts and/or individual values.	Decision making matrix is not complete and/or inaccurate information was included.	Little or no attempt is made to complete the decision making matrix.
	10 8	6 4	3 2
Final Decision *If we had to make the decision, what would it be and why?*	Student chooses just one of the available options and the decision made shows evidence of collaboration and consensus between partners. The explanation as to why this was the final choice is clear and easy to understand.	Final decision is not clear OR is not limited to just one option OR it lacks a reasonable explanation as to why this was the final choice.	No clear final decision is made and no explanation is included.
	10 8	6 4	3 2
Participation in Class Discussions *Am I respectfully engaged in the conversations related to carrier testing?*	Student shares his/her own views with classmates and respectfully listens to the views of others. There is evidence that student is aware of the difficulty of making such decisions--that there is no one right answer and individual values play a large part in this process.	Student participation in class discussion is lacking or student does not show respect for the views of others.	Little or no attempt is made to participate in class discussion.

Student self-assessment: _____/ 50 **Teacher assessment: _____/ 50**
Extra Comments: