NOVA
science
NOW

Risky Genetics

Can genetic testing predict if you will get a particular disease? Sometimes. Only a small number of diseases are caused by a defect in a single gene. Most result from a combination of multiple genes and other factors, such as pollution, eating habits, and alcohol, tobacco, and drug use. Today, you and a partner will play the role of genetic counselors. Two patients have asked you to interpret their genetic test results and make recommendations.

Procedure

1 Before beginning your role as genetic counselors, you and your partner will each create a character with unique genes and lifestyle habits. To do this, fill in Sections 1–3 of the Patient Record. (NOTE: The defective gene for breast cancer is called BRCA2. The defective gene for lung disease is SERPINA1, and the one for Alzheimer's is PSEN1.)

2 You and your partner need to be briefed about each disease. Use the videos and Web sites below to fill in Section 4 the Patient Record.

Breast Cancer videos (2 minutes each)

pbs.org/pov/inthefamily/video_classroom1.php

pbs.org/pov/inthefamily/video_classroom2.php

Lung Disease Web sites

learn.genetics.utah.edu/content/disorders/whataregd/a1ad/

nlm.nih.gov/medlineplus/ency/article/000120.htm

Alzheimer's video (3 minutes)

teachersdomain.org/resource/tdc02.sci.life.gen.alzheimers/

3 With your partner, discuss how responsive each disease is to preventive treatments or changes in lifestyle. Now that you know the genetic profile of your two characters, consider how important it is that they be tested for each of the diseases. Summarize your recommendations in the space below.

Patient Record

1 **Gender:** ☐ Female ☐ Male *(Decide with your partner.)*

2 **Lifestyle:** ☐ Very Healthy ☐ Average ☐ Unhealthy *(Roll the die. If the number is: 1 or 2 = Very healthy; 3 or 4 = Average health; 5 or 6 = Unhealthy—patient eats junk food, smokes, doesn't exercise, and works in a dusty factory.)*

3 **Gene Profile** (For each cell, roll the die. Even = dominant; Odd = recessive)

	Breast Cancer (BRCA2)	Lung Disease (SERPINA1)	Alzheimer's (PSEN1)
Allele 1			
Allele 2			

4 **Disease briefing**

	Breast Cancer (BRCA2)	Lung Disease (SERPINA1)	Alzheimer's (PSEN1)
Genotype (heterozygous, homozygous recessive, or homozygous dominant)			
Genetic risk (high or low)			
Lifestyle effect on risk (increase, decrease, no effect)			

4 The two characters (yours and your partner's) are considering having children together. Complete Punnett squares for the three genes to show the possible genotypes of future children. Then, write the probability of each genotype and draw a star next to genotypes with an increased risk of disease.

Punnett Squares

BRCA2		

SERPINA1		

PSEN1		

Probabilities

	BRCA2	SERPINA1	PSEN1
Homozygous dominant			
Heterozygous			
Homozygous recessive			

5 Punnett squares show a child's theoretical probability of having a defective gene. But how does theory compare to what actually happens? To find out, you need to look at a child's pedigree, or family history. Get a Pedigree Strip from your teacher. On a separate piece of paper, draw the child's pedigree, using the following symbols.

Unaffected male   Affected male   Unaffected female   Affected female   Marriage

6 For each disease, look at the pedigree and make a prediction about whether a child is very likely, moderately likely, or unlikely to acquire it.

Pedigree # _____ Gender(s): _____ Environment: _____

	BRCA2	SERPINA1	PSEN1
Prediction for getting the disease			
Recommend testing? (Yes/No)			

Questions
Write your answers on a separate sheet of paper.

1 What is the difference between a Punnett square and a pedigree?

2 Can doctors predict who will get a particular disease based on a genotype for one gene alone? Why or why not?

3 Why might factors, such as food choices, pollution, or smoking habits, not have the same effect on all people?

4 List some of the advantages and disadvantages of genetic testing.

5 Do the advantages of genetic testing outweigh the disadvantages? Form an opinion and support your response with facts and examples.