

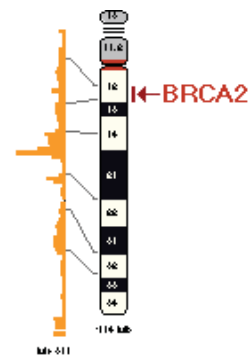
Genetic Testing and Breast Cancer: Is a Little Knowledge a Dangerous Thing?

by

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Kathy was late for her book club meeting. She had had her yearly mammogram and the results had been negative—all was well. However, she had been asked to wait because Dr. Benjamin, the radiologist, wanted to talk to her.

She raced into the restaurant where the book club was meeting, sat down with her friends, and burst into tears. Her friends gathered around her, concerned.

Mary spoke first. “Kathy, why are you so upset?”

Kathy calmed herself and looked at her friends. “I’ve just come from Dr. Benjamin’s office.”

Sarah interjected, “Please don’t tell us that he found a lump.”

“No, no,” said Kathy. “My mammogram was negative. But we had the most upsetting conversation afterwards. He wants me to get tested for the breast cancer gene. He says that I should find out if I’m at risk.”

Peter jumped in. “Of course you’re at risk. Your daughter, Rachel, survived breast cancer—you know you are at risk. What’s the matter with him?”

John added, “I guess he figures that since Rachel had breast cancer, she might have the gene, and she might have gotten it from you.”

“Dr. Benjamin was explaining the gene to me, but I really don’t get it. You know, the whole thing scares me,” said Kathy. “I can’t see what good can come from knowing if I have it.”

John interjected, “Susan and I had ourselves tested for Tay Sachs. Knowledge is power, Kathy. Why wouldn’t you get tested? The more you know, the better informed decisions you can make.”

“Knowledge may not always be an ally,” said Sarah. “Some years ago, when I was fired from my job, I suspected it was because I mentioned that my Dad had Huntington’s disease.”

“I’ll tell you something good you can do,” said Peter. “Eat healthy and exercise. That’ll do you more good than all the worry and stress about testing.”

Martha jumped in. “The knowledge from the Human Genome Project is exciting, Kathy. Every day at work we move closer to finding the causes of disease by our knowledge of genes.”

“My parishioners are often telling me of good things accomplished by genetic testing,” said Mary.

Kathy started crying again. “This is just too confusing for me.”

Martha put her arm around Kathy. “We’re here for you, Kathy. We’ll sort this out. It sounds like each of us has some information that may help. Let’s think about what we know that might be useful, and then we can share that with you. I’m sure there’s a good answer for you.”



Credit: The BCRA1 and BCRA2 genes map, respectively, to chromosomes 17 and 13. Illustrations from the National Center for Biotechnology Information, <http://www.ncbi.nlm.nih.gov/>. Case copyright held by the **National Center for Case Study Teaching in Science**, University at Buffalo, State University of New York. Originally published July 31, 2002; last revised February 19, 2012.. Please see our **usage guidelines**, which outline our policy concerning permissible reproduction of this work.

Role-Play Sheet for Martha

Bioinformatics Researcher at a Pharmaceutical Company (Expertise—genetic and DNA analysis)

Bioinformatics is a new field of study that combines biology and information technology. It is used to analyze large databases of information, such as those resulting from the Human Genome Project.

- Breast cancer facts
 - 180,000 women a year are diagnosed with breast cancer.
 - 5%-10% of these are hereditary (caused by known breast cancer genes).
- Two breast cancer genes have been identified: BRCA1 and BRCA2.
- The normal functioning of both genes is to inhibit the development of cancerous cells. They are tumor suppressor genes.
 - A mutation in either gene raises a woman's lifetime chance of getting cancer from 12% to 85%.
 - A mutation in either gene also raises the risk for ovarian cancer.
 - A BRCA2 mutated gene raises the risk for breast cancer in males.
 - More than 600 different mutations that would increase the risk of breast cancer have been found in each gene.
 - There are many more mutations that have been found and are being studied. They may be harmless, and their connection to breast cancer is not yet known.
 - Every person has two copies of each gene. A mutation in one copy will increase the risk of breast cancer.
 - A person with one good copy and one mutated copy has a 50-50 chance of passing the mutated gene on to his/her children.
 - Virtually all individuals with a mutation in BRCA1 or BRCA2 have inherited it from a parent.
- Benefits of genetic information to research
 - The Cancer Family Registry for Breast Cancer Studies has information and lab specimens from more than 5,000 families with breast and/or ovarian cancer.
 - The Registry makes this information available to researchers.
 - The Registry provides information that would take researchers years to gather by themselves.
 - The information in the Registry is key to a speedy solution to breast cancer. The greater the number of families that provide information to the Registry, the more we will know about detecting and treating breast cancer and the faster we will learn how to prevent it.

Questions

1. Would Martha encourage Kathy to get the test? Why or why not?
2. What other advice would Martha give Kathy?
3. How might these decisions affect Kathy cognitively, psychologically, emotionally?

Role-Play Sheet for John

Father of Three Adopted Children (Expertise—genetic testing as a means to information)

John and his wife, Barbara, decided to be tested for the Tay Sachs gene. John and Barbara both tested positive for the mutated form of the Tay Sachs gene. John and Barbara decided to marry anyway, but to adopt children rather than run the risk of having a child with Tay Sachs disease. (The book club members do not know this story—you may tell it to them.)

- Genetic testing
 - The test only requires a blood sample to be drawn.
 - The cost of genetic testing for breast cancer varies from several hundred to several thousand dollars, depending on the type and levels of testing required.
- Tests are available that can screen for cystic fibrosis, Duchenne muscular dystrophy, fragile X syndrome, Gaucher's disease, Huntington's disease, Lou Gehrig's disease, Marfan syndrome, Tay Sachs, and many others.
- Tay Sachs disease—a genetic disease that can be identified by genetic testing.
 - Children born with Tay Sachs, a genetic disease, die by the age of two.
 - Tay Sachs is a recessive disease—you need two copies, one from each parent, to get the disease. Each natural child of two carriers has a one in four chance of having the disease.
 - A massive government genetic screening program during the 1970s reduced the number of babies dying per year from 50-100 to less than 15.
- Law
 - The Americans with Disabilities Act (ADA) forbids employers from discriminating against disabled individuals who can do their job.
 - In March 1995, the Equal Employment Opportunities Commission (EEOC) said that people with genetic predisposition to disease are perceived as disabled and protected by the ADA.
 - The Health Insurance Portability and Accountability Act (HIPAA) of 1996 prohibits group health plans from denying coverage based on health status. An individual cannot be excluded just because of genetic information.
- News—Government genetic screening program recommendations address cystic fibrosis.
 - Cystic fibrosis (CF) is a genetic disease that usually affects the lungs and digestive system. How sick a person gets varies widely. There is no cure; most die by age 30.
 - CF is a recessive disease. A person who is a carrier has no symptoms.
 - While over 900 mutations have been found, about 25 are the major contributors.
 - The presence of the disease varies widely among ethnic groups.
 - Screening is recommended for individuals with a family history of CF, for partners of people with CF, for couples planning a pregnancy, and for couples seeking prenatal care. Testing for the general population is not recommended.
 - Screening for the 25 mutations is more cost effective than for 900 mutations.
 - Screening for the 25 mutations will identify carriers more accurately for ethnic groups that have the 25 mutations, and less accurately for ethnic groups with rare mutations.

Questions

1. Would John encourage Kathy to get the test? Why or why not?
2. What other advice would John give Kathy?
3. How might these decisions affect Kathy cognitively, psychologically, emotionally?

Role-Play Sheet for Sarah

Daughter of a Man who had Huntington's Disease (Expertise—insurance/employment risks as a result of genetic testing)

Sarah's father died of Huntington's disease. She has a 50% chance of having inherited the disease. She mentioned at a staff meeting that she was the primary caretaker of her father who had Huntington's disease. One week later, she was fired, although her previous evaluations showed outstanding performance on the job.

- Employment risk example
 - Burlington Northern Santa Fe Railroad story—In 2000, this company drew blood samples during a physical exam from employees who had developed carpal tunnel syndrome on the job. The blood was sent for genetic testing, without employee knowledge or consent, in an attempt to find a gene that is in some way connected to carpal tunnel syndrome, so the employers could absolve themselves of their responsibility. One employee refused testing and was threatened with termination.
- Health insurance risk example
 - Danny's story—Danny is 7 years old and in perfect health. Genetic tests revealed that he has a gene predisposing him to a heart disorder. He takes medication to lower his risk of heart attack. His insurance company canceled his health insurance, stating that since he had the gene from birth, it was a pre-existing medical condition.
- Testing implications
 - Many surveys of physicians and patients have been conducted and revealed hundreds of cases of genetic discrimination. Examples:
 - In 1996 Georgetown University polled 332 families with perceived genetic risks:
 - † 22% were refused health insurance.
 - † 13% were fired from their jobs based on their risk.
 - A 1999 Yale Center survey of 296 genetic counselors, when asked how they would deal with genetic screening for themselves, reported:
 - † 68% said they would not bill their insurance company for the cost of genetic screening for fear of discrimination.
 - † 26% said they would give a false name.
 - † 57% said they would seek psychological counseling to cope with the results.
- Legislation
 - President Clinton signed an executive order prohibiting the federal government from using genetic testing results in any decision to hire, fire, or promote its employees. This bill covers 2 million federal employees. It does not apply to the private sector.
 - In 2007, a bill sponsored in the House of Representatives (H.R. 493) by Slaughter, and in the Senate (S. 358) by Snow, would prohibit discrimination on the basis of genetic information with respect to health insurance and employment. After 13 years of legislative proposals, the Genetic Information Nondiscrimination Act (GINA) was signed into law in May 2008.
 - As of 2008, some states already had their own laws about genetic discrimination, pertaining to:
 - health insurance: 45 states
 - employment: 35 states
 - disability insurance: 15 states
 - life insurance: 14 states
 - consent required to disclose genetic information: 27 states

Questions

1. Would Sarah encourage Kathy to get the test? Why or why not?
2. What other advice would Sarah give Kathy?
3. How might these decisions affect Kathy cognitively, psychologically, emotionally?

Role-Play Sheet for Peter

Yoga Instructor (Expertise—healthy eating, aerobic exercise, stress reduction)

More than 90% of women who get breast cancer do not have the gene. Every woman has a 12% chance of getting cancer.

Things you can do to prevent cancer whether or not you have the gene:

- Diet
 - Alcohol increases the risk of cancer, including breast cancer. A few drinks a week is related to an increased risk.
 - Vegetables and fruits reduce cancer risk.
 - Normal metabolism can damage tissues, and this damage is related to an increased risk for cancer. Vegetables and fruits contain antioxidants that may protect tissues.
 - Antioxidants include: vitamin C, vitamin E, selenium, carotenoids, and phytochemicals.
 - Obesity is related to increased cancer risk, including cancer of the breast among postmenopausal women.
 - Saturated fats may increase cancer risk.
 - Fats containing omega-3 fatty acids may reduce cancer risk.
 - Fish (such as salmon) are high in omega-3 fatty acids.
 - Too little folic acid increases risk of cancer, including breast cancer.
- Exercise
 - Physical activity, weight control, and diet may delay or prevent cancer in people with genetic risk factors.
 - Moderate to vigorous levels of physical activity lowers risk of cancer, including breast cancer.
- Life style
 - Use of hormone replacement therapy after menopause for more than 5 years increases the risk of breast cancer.
 - Smoking is linked to cancer. Teenage smoking increases the risk of breast cancer.
- Psychological factors
 - When genetic testing reveals having the gene, psychological outcomes vary, and sometimes depend on the personality of the person tested.
 - Carriers expressed anger and worry.
 - Persons who had cancer reacted with anger, worry, discouragement, and various concerns upon finding that they had positive genetic test results.
 - Persons who underestimated the impact of testing were still distressed six months later.
 - Some persons wanted to join support groups.
 - Some no longer exhibited six months after testing.
 - When genetic testing reveals not having the gene:
 - There is psychological relief.
 - False negative results are possible and can lead to complacency.
- Medical support includes: regular mammograms, monthly self examination, and annual gynecological exam.

Questions

1. Would Peter encourage Kathy to get the test? Why or why not?
2. What other advice would Peter give Kathy?
3. How might these decisions affect Kathy cognitively, psychologically, emotionally?

Role-Play Sheet for Mary

Minister (Expertise—making ethical decisions)

- Ethical responsibilities
 - Genetic testing allows you to give useful information to other family members.
 - All blood relatives have similar genetic inheritances.
 - If you are tested, you may feel responsible to share the information with other family members.
 - Some family members may want information, some may not.
 - Genetic testing now provides data for future studies that may help future generations, such as Kathy’s grandchildren.
- Ethical worries
 - There is no guarantee that test results will remain private.
- Psychological factors
 - How does the possibility that a person has breast cancer affect their outlook?
 - How do you see yourself? How do you see the world?
 - Which aspects of your health do you regard with personal guilt?
- Questions, when a test for a disease is available:
 - Who should be screened?
 - At what age?
 - Who will explain the test beforehand?
 - Who will explain the results?
 - What about results of a test that is known to not be 100% accurate?
- Quote and comment from Dr. Francis Collins, Director of the National Center for Human Genome Research:
 - “It is not inconceivable that every woman in America may want to be screened for this [breast cancer] gene. The economic, ethical, and counseling issues will be very daunting.”
 - In the near future, physical examinations for 18 year olds will include DNA testing for diseases with genetic components, and that physicians in the interest of preventive medicine will make risk-based recommendations for a healthy life style.

Questions

1. Would Mary encourage Kathy to get the test? Why or why not?
2. What other advice would Mary give Kathy?
3. How might these decisions affect Kathy cognitively, psychologically, emotionally?