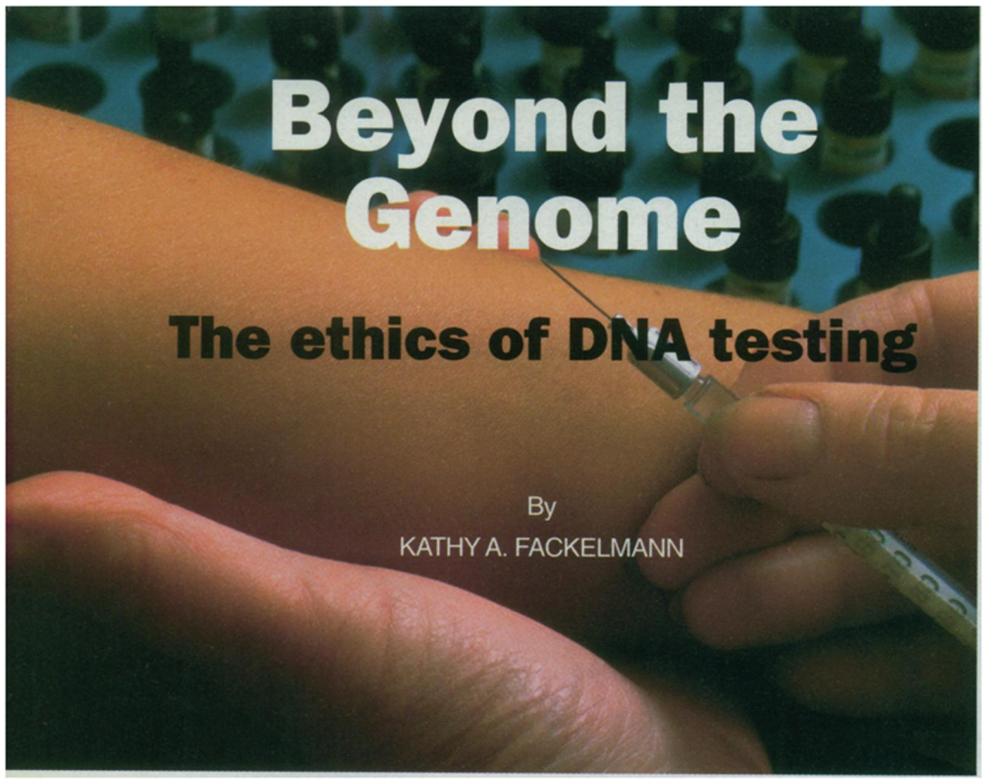

Beyond the Genome

The ethics of DNA testing

By
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Imagine you're playing a board game called GENES-R-US. You roll the dice and pick a card. It says:

You're a 29-year-old pregnant woman. You've just been told you have a gene that almost always causes breast cancer in early adulthood. Your daughter-to-be has inherited this gene. Do you:

(a) abort the fetus to avoid a disease that won't strike for decades?

(b) carry the pregnancy to term and hope your daughter will not develop cancer until a cure has been found?

If you think such a scenario is science fiction, think again.

In September, the news broke that researchers had isolated the long-sought gene responsible for one inherited form of breast cancer. The flawed version of this gene, dubbed BRCA1, is thought to put women at extraordinarily high risk of developing breast cancer (SN: 9/24/94, p.197). Within 2 years, scientists expect to market a blood test to identify those who have inherited the mutant gene.

As geneticists bag their quarry with increasing speed, unresolved medical and ethical issues continue to surface. Society has yet to settle, or in some cases even address, the complex questions that abound in this brave new world. And the road ahead surely contains many more conundrums created by newfound knowledge.

The virtual explosion in genetics research during the last 10 years has led to the identification of genes that underlie heart disease, colon cancer, cystic fibrosis, Duchenne muscular dystrophy, Huntington's disease, and a variety of other ailments that have plagued human beings for millennia.

Researchers around the world continue their feverish hunt for other elusive disease-causing genes, including another gene for familial breast cancer known as BRCA2.

An international team of investigators is closing in on a gene responsible for one type of deafness. Another team has identified the DNA region thought to house a gene for dyslexia, a condition that causes reading difficulties (SN: 10/22/94, p.271).

This fall, researchers reported finding the location of two genes that may contribute to type I diabetes, the most severe form of this sugar-processing disorder (SN: 9/10/94, p.164).

And in an explosive break with tradition, geneticists are beginning to link genes to complex human behaviors, including sexual orientation. Just last year, National Institutes of Health researchers zoomed in on a region of chromosome X believed to contain a gene that may play a role in some homosexual behavior (SN: 7/17/93, p.37).

Such advances bring many tangible benefits to society and to people who suffer from genetic disorders. For example, the discovery of the malfunctioning cystic fibrosis (CF) gene led to the identification of a protein that produces the abnormally thick mucus that clogs the lungs of people with this disorder. Researchers have used that information to fashion new therapies for this disorder. Recently, researchers took a historic first step by inserting a healthy version of the CF gene into the lung of a patient with cystic fibrosis (SN: 9/3/94, p.149).

These examples represent a new frontier in genetic discoveries. The Human Genome Project, a 20-year, multi-billion-dollar effort, aims to chart the estimated 100,000 genes that orchestrate human life. This ambitious undertaking, if successful, means that people of the future may obtain a computerized printout of their genetic code. Couples in such a world may ask for — and get — a detailed DNA profile of an embryo floating in a petri dish.

Such knowledge can prove potent. Insurance companies may refuse to offer coverage to a healthy infant whose DNA foretells a heart attack 40 years later. Indeed, some of the thorny aspects of genetic testing have already surfaced. Geneticists offer a blood test for Huntington's disease, yet this illness has no cure. Thus, people who opt for the test may spend years waiting for the first signs of their deterioration.

Scientists and ethicists routinely discuss the ethical difficulties raised by the project to map the human genome. Yet such discourse must move out of the realm of academia. Society must weigh in on the debate, as it touches nearly every aspect of what it will mean to be human in the next century.

What do you think?

Last summer, at the Short Course in Medical and Experimental Mammalian Genetics held at the Jackson Laboratory in Bar Harbor, Maine, researchers met to discuss scenarios that illustrate the many facets of genetic counseling. All are real cases.

SCIENCE NEWS invites you to read through the following cases and come to your own conclusions. How would you handle the situation outlined? What should society do when such difficulties arise?

We will report on reader responses — and the reactions of the Bar Harbor participants and leading geneticists and ethicists — in the Dec. 17 SCIENCE NEWS. Please keep your responses brief and send them by Nov. 28 to:

Cases
SCIENCE NEWS
1719 N Street, N.W.
Washington, D.C. 20036

or fax them to:
1-202-467-5039.

These cases were described by the organizers of the Bar Harbor ethics discussion: Clair A. Francomano of the Johns Hopkins University School of Medicine in Baltimore, Judith G. Hall of the University of British Columbia in Vancouver, Laird G. Jackson of the Thomas Jefferson Medical College in Philadelphia, and John A. Phillips III of the Vanderbilt University School of Medicine in Nashville.

A Case of Dwarfism

A husband and his pregnant wife seek genetic counseling. Each carries one flawed copy of the gene responsible for achondroplasia; thus both are dwarfs. Recently, a California research team described the mutation in a gene on chromosome 4 that causes achondroplasia.

The counselor explains that genetic testing can determine whether the fetus has inherited the achondroplasia mutation. In the discussion, the couple inform the counselor that they will abort any fetus that carries two mutant genes. That's not surprising, since children born with two such genes rarely survive beyond infancy. Indeed, the couple had had such a child.

This time around, they say, they want a baby who is heterozygous for the achondroplasia trait. Such a child inherits a flawed gene from one parent and a healthy gene from the other parent. That genetic combination means the child will be a dwarf — just like the parents.

At the same time, the parents say, they will abort any fetus that does *not* inherit one copy of the mutant gene.

Should the counseling center perform the test, knowing that the couple plan to abort a healthy fetus?

Some things to consider include the fact that achondroplasia is a fairly serious disorder. The bones can be abnormal in structure, sometimes requiring the use of a wheelchair.

Yet many dwarfs live long, healthy lives and don't regard their condition as a disability. In addition, some couples with achondroplasia worry about the problems involved in raising a normal-size child.

Under *Roe v. Wade*, women in the United States have the right to obtain an abortion during early pregnancy for a variety of personal reasons.

A Case of Paternity

A husband and wife have a child who suffers from cystic fibrosis, an incurable, fatal hereditary disease that results in frequent infections and difficulty breathing. The couple want to determine their risk of having another child with this disorder.

Because CF is a recessive disorder, a child usually must inherit the CF gene from both parents to get the disease. A child with just one CF gene is a carrier: Such a person doesn't have the disorder but can pass the trait on to the next generation.

The DNA test revealed that the mother of the child carried the CF trait. However, her husband did not. Indeed, the DNA tests showed that he was not the biological father of the child.

That fact significantly decreased this couple's chance of having another child with CF. But the test has put the counselor in a difficult situation. Should the counselor tell the couple about the nonpaternity findings? Should the mother be told privately? If so, is the center colluding with the mother to withhold information from the husband?

In addition, this case brings up issues concerning the biological father of the child. This man has not contracted with the genetics center for the tests, yet the counselor now knows that this man is probably a carrier of the mutant gene for CF. Should the genetic counselor call this man and tell him about his risk?

A Question of Privacy

A 30-year-old woman has been diagnosed with familial adenomatous polyposis, an inherited condition that puts her at extremely high risk of colon cancer. Indeed, most people with this condition, if untreated, will develop the cancer by age 40.

The patient's own mother died of colon cancer at age 32. Despite this grim history, the woman refuses to share the diagnosis with her family, including her four siblings and her husband. In addition, she does not want her two children to be tested for the genetic flaw. Researchers know that the mutant gene responsible for this disorder lies on the long arm of chromosome 5. This flaw results in hundreds or thousands of polyps, small wartlike growths, carpeting the lining of the colon. Some of the polyps will become malignant.

The woman's refusal to tell her family puts the genetic counseling center in a quandary. To honor this patient's request might harm the rest of the family. Without appropriate medical intervention, family members with the mutant gene remain at extremely high risk of dying of colon cancer. People who carry this flawed gene undergo frequent screening for suspicious growths in the colon. That aggressive approach slashes their risk of dying of the disease.

Should the counselor disclose the results of the test to the rest of the family despite the woman's objections?

Who "owns" the information encoded in a person's genes? Bear in mind the fact that the rest of the family shares a common heritage with this woman.

A Case of Who Decides

A 37-year-old pregnant woman worries about her family's history of mental retardation. Genetic testing shows this woman is a carrier of the mutant gene for fragile X syndrome, an inherited form of mental retardation. U.S. and Dutch researchers have demonstrated that carriers and people with this condition show repetitive DNA segments in a gene called FMR-1 (SN: 6/8/91, p.359).

The woman is offered prenatal testing and says she will abort any affected fetus. She also indicates that she will abort a female fetus that is a carrier of the flawed gene. She states that she wants her legacy of this inherited disorder to end.

This case raises the issue of whether the woman should have the right to abort a healthy fetus that happens to be a carrier of a genetic mutation. In a sense, the mother is making her future daughter's decision for her. By choosing an abortion, the patient has ensured that she will not have grandchildren who carry the flawed gene.