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### **AMA CODE SAYS**

#### **The Use of DNA Databanks in Genomic Research**

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Consideration of genetic matters has been entering the *Code of Medical Ethics* over the last 2 decades, often as add-ons or inclusions to existing opinions. In 1993, for example, Opinions 2.04 and 2.05 on artificial insemination from 10 years earlier were amended to cover newly available genetic screening to test sperm before fertilization. (Notably testing for HIV was also added to this and to other Opinions in their 1990s revisions.) Ethical policy on informed consent from research subjects applies, of course, to subjects in gene research; admonitions regarding conflict of interest in research must be heeded by those conducting genetic research, and so on. In many ways, as ethicists have stated again and again, genetic information and technology do not represent a brand new world of ethical, legal, moral, or social concerns.

On the other hand, many people insist that when it comes to issues of consent and confidentiality, genetic information *is* different.<sup>1</sup> There are 2 main reasons for thinking so. First, genetic information, and most especially DNA test results, can reveal more than an ordinary medical record; it can indicate illnesses (breast or colon cancer, for example) and risk conditions (high cholesterol, e.g.) that we might develop in the future. Such information could prejudice insurance companies and employers and could influence one's educational, reproductive, and other lifestyle decisions. The second distinct characteristic of genetic information that complicates standard consent guidelines for physicians is that data obtained about a patient may apply not only to that patient but also to other members of his or her biological family. Such information has, thus, been acquired about that family member without his or her consent and may be unknown to (and perhaps unwanted by) him or her.

For these reasons, the Council on Ethical and Judicial Affairs (CEJA) began considering genetic testing as a separate topic area for opinions and has issued opinions on genetic testing by employers (Opinion 2.132), genetic information and insurance companies (Opinion 2.135), ethical issues in carrier screening (Opinion 2.137), genetic testing of children (Opinion 2.138), and multiplex genetic testing (Opinion 2.139).

At its winter meeting in 1999, CEJA was asked to investigate and report on how physicians should deal with the increasingly thorny issues surrounding test results. Should they document test requests in regular patient records? Should they document test results? If test results affect a patient's family member, what should physicians

do? How should they respond to the rapidly expanding demands from the judicial system for DNA information to be used in adoption and custody cases or for forensic use in criminal cases? CEJA subdivided this large cluster of questions twice, setting the justice issues apart for separate consideration and then subdividing the remaining non-justice-related issues into (1) those issues that concern individual patients and (2) those that concern the special area of population genetics. This subcategory—gene research on populations, called "genomic" research—is the subject of Opinion 2.079.

Genomic research is not clinical research; that is, it does not concern itself with attempts to diagnose or treat illness in individuals. Rather, genomic research looks at specific loci on the genome and notes the similarities and differences at those loci among many people. These horizontal studies reveal that many different forms of a gene (polymorphisms) exist at a given locus; some polymorphisms affect an individual's health or appearance and some do not. Understandably, genomic studies yield more information when conducted among people about whom medical history and family history are available—populations of people, in other words, who are stable, remain in the same place over generations, and marry people on whom medical histories and records are also available. For these reasons, genomic researchers find studies of American Indians, groups such as the Amish, and those who have remained in a distinct geographic location (like the residents of Iceland) particularly attractive. But studying a group that identifies itself or is identified by others as distinct on the basis of its heritage, culture, beliefs, or geographic confines presents consent and confidentiality problems that transcend those found in clinical gene research with individual subjects.

CEJA approached these challenges with greatest concern for protection of informed consent. Because study results will be made public, all group members will have information about aggregate data that may or may not apply to them as individuals and that they may or may not have sought on their own. There is, on average, about 0.1 percent difference between the genomes of any 2 randomly selected individuals; roughly 40 or so of the approximately 40,000 genes scientists currently think *Homo sapiens* possess. But, and here a second risk to consent comes in, as genomic research is attempting to clarify, genes act in conjunction with each other and also with the environment and other lifestyle influences, so that groups that have shared environment, diet, and other customs over many generations and have married others like themselves, may have slightly more genetic similarity than the general 99.9 percent of the world population. And they may have medical histories that show even greater similarities in illness patterns. Having enough subjects to be able to find correlation between genes and health factors is the beauty of population genetics, but it opens the possibility of discrimination or stigmatization of all members of a group on the basis of generalized studies in which individual members did not consent to participate.

Finally, DNA databanks can preserve genetic material so that it can be used over and over again for research purposes through time, and CEJA was aware that, because of

this, subjects who consented to participate in one research protocol might be unwittingly committing their DNA samples to later research projects that they had not consented to and, perhaps, would not approve of.

With these special sensitivities in mind, CEJA determined that policy regarding genomic research must consider both (1) some procedure for consulting the whole group and (2) stringent consent procedures for individuals who chose to participate in the study. The recommendations adopted at the House of Delegates meeting in December 2001 that became Opinion 2.079 laid out these guidelines.

#### **Regarding the group being studied**

". . . investigators should consult with the community to design a study that will minimize harm not only for individual subjects, but also for the community. *When substantial opposition to the research is expressed within the community, investigators should not conduct the study.* When the community supports a proposal, investigators nevertheless should obtain individual consent in the usual manner. The same procedure should be followed whether the investigators intend to collect new samples and data or whether they wish to use previously archived data sets."

#### **Regarding the individuals who consent to participate**

1. Standard informed consent principles apply. In addition, the specific standards of privacy operating in the study must be disclosed. Will the material be "coded (i.e., encrypted so that only the investigator can trace materials back to specific individuals) or . . . completely de-identified (i.e., stripped of identifiers)"?
2. "If data are to be coded, subjects should be told whether they can expect to be contacted in the future to share in findings or to consider participating in additional research."
3. "Individuals should always be free to refuse the use of their biological materials in research, without penalty."
4. "Disclosure should include information about whether investigators or subjects stand to gain financially from research findings."
5. "Subjects should be informed of when, if ever, and how archived information and samples will be discarded."

Finally, Opinion 2.079 advises that "to protect subsets of the population from such harms as stigmatization and discrimination, demographic information not required for the study's purposes should be coded."

The *Code of Medical Ethics'* new Opinion on use of DNA databanks in genomic research takes a strict line on informed consent by groups and individuals who are

subjects of such research. Some procedure for securing group consent must be designed and implemented with the group and individuals must give consent to both the initial and any subsequent use of their genetic material..

### **References**

1. Lagay F. Should genetic information be treated separately? *Virtual Mentor*. 2001;3. Accessed January 7, 2002.

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