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Protecting individuals and promoting science

Last month, Lord Falconer of Thorton and Julian Peto, head of epidemiology at the Institute of Cancer Research near London, argued in the pages of *The Times* (London) the value of legislation designed to protect patients' medical information. According to Lord Falconer, British law requires that patients be given a clear explanation of how their data are used and consent to the disclosure of their medical information; but the Lord dismissed concerns that the law prevents researchers from having access to such data. Peto, however, says that research, including his own, is being hampered because doctors, often unclear about what the law actually permits, are afraid to give information to researchers for fear of prosecution. "Clearly worded new legislation is needed to restore the long-established principle that individuals need not be contacted when their civil or clinical records are used for bona fide medical research that does not affect them and has been approved by an accredited research ethics committee," writes Peto.

The arguments highlight one of the most challenging issues facing governments as they try to protect individuals from abuse in the post-genome era: how to determine the right balance between ensuring the privacy of medical information and its fair use. Should individuals know who is looking at their personal information? Can researchers who wish to carry out large population studies be reasonably expected to contact every individual whose information is they would like to use? (See page 207 for discussion of these issues in the context of pharmacogenetics.)

Geneticists, industry representatives, and lawmakers agree that policies protecting confidentiality and privacy in research and strong anti-discrimination laws are essential not only to protect individuals but also to ensure the advancement of science. A case in point is the growing number of reports indicating that about one-third of people invited to participate in genetic research studies refuse because they fear discrimination. However, few agree on the kinds of policies and regulations that are needed.

Many countries, including the United States, lack clear and comprehensive laws on genetic privacy and confidentiality. In the United States, federally funded research projects are reviewed by institutional review boards (IRBs). In most cases, IRBs determine whether the project's informed consent document also describes the extent to which confidentiality of records that identify the subject will be maintained—and whether the mechanism(s) for doing so are appropriate. But practices vary and there are no specific requirements for even the most basic level of privacy and confidentiality protection. For this and other reasons, IRBs have come under

fire in the past couple of years and several groups and organizations have called for a complete overhaul of the system that regulates human research.

For now, however, regulations on the privacy of medical information, including genetic information, are fragmented across many federal agencies and by a patchwork of state laws that vary widely depending on the state and the type of information. In April of this year the US Department of Health and Human Services introduced another layer of complexity through its issue of new federal privacy regulations that will go into effect in two years. The regulations require most health care providers to obtain their patients' consent for use and disclosure—even routine disclosures—of health records. Patients will have the right to review their own records and to request amendments and corrections and to view a disclosure history listing all entities that have received medical information on them.

Advocates of strong privacy regulations point out that the regulations have several loopholes. For one, while they apply to group health plans, health care providers, and health care clearinghouses, they do not cover many entities that hold medical records, such as life insurers and workers' compensation programs. Also, information obtained from the analysis of tissue or DNA through research that has no medical ramifications is likely to be exempt.

Although the new regulations do not go far in strengthening privacy protection, some are concerned that they might have a negative effect on research. Industry representatives think that the complex nature of the regulations, the steep sanctions against those who break them, and the short time frame for compliance by the health establishment will serve as disincentives to provide medical information to researchers.

Moving forward on federal anti-discrimination laws in the United States has been just as problematic. Although genetic anti-discrimination bills are introduced every year and there seems to be bipartisan support for such bills, the complexity of the subject, difficulty in getting all interested parties to agree, and, to some extent, lack of interest (genetic privacy does not seem to be considered a major problem), have made it difficult to move the proposed legislations through government.

Prominent researchers have voiced strong support for anti-discrimination laws, and indeed these laws might go a long way to satisfy public concerns over participating in genetic research studies. Representatives of the health insurance industry, on the other hand, say that such laws are unnecessary because genetic information is not used to set premiums or make decisions on coverage, and employers say that laws are already in place to protect individuals from discrimination.

The existing anti-discrimination laws (which are more limited in scope than the newly proposed ones) have never been tested in court—although one recent case came close. Last year the Burlington Northern Santa Fe Railroad began testing workers (who had reported wrist injuries for a genetic polymorphism associated with carpal tunnel syndrome). The Equal Employment Opportunity Commission sued the railroad following complaints by the workers' union: apparently the tests were carried out for discriminatory purposes and without the knowledge of those being tested. The company denied the charge but said it would abandon the testing policy—and so the suit was dropped a few months ago.

Since the announcement of the draft sequence of the human genome, there has been a steady flurry of media reports speculating on its implications for human health. Improved diagnostics, individualized therapy and targeted screening of susceptible populations are a few of the benefits promised in the post-genome era, and indeed we are starting to see the first glimpses of them. It would be disappointing if the biggest hurdles to bringing these benefits to society were not technical ones but the failure to convince the public to trust that their participation in science would be to their benefit, and the inability of scientists to work with legislators to develop clear guidelines that strike the right balance between timely promotion of research and protection of people.