ETHICAL, LEGAL, AND SOCIAL IMPLICATIONS

42. What is known about stigmatization, discrimination, health disparities, privacy/confidentiality and person/family/social issues?

Potential risks of being identified as having an inherited susceptibility to colorectal cancer are listed in Table 9.

Table 9. Potential burdens of genetic testing for hereditary colorectal cancer (modified from Trimbath and Giardello, 2002)

If result is gene positive (disease-causing mutation detected):

- Psychological distress, including anxiety, depression, anger, or denial
- Changes in family psychosocial dynamics
- Stigmatization
- Increased fear about surgery or death
- Guilt/worry about transmission to children
- Worry about genetic discrimination re insurability and employability
- Colon surgery and possible lifestyle changes

If result is gene negative (truly negative for a gene mutation identified in the family)

- Survivor guilt (guilt about being unaffected when other family members are affected)
- Changes in family psychosocial dynamics

If result is inconclusive (no family mutation found or variant of unknown significance)

- False sense of security
- Need for continued screening
- Confusion/anxiety

Nearly half of individuals at 50 percent risk of inheriting an HNPCC mutation are not tested (Hadley *et al*., 2003). Explanations proposed are that these individuals: (1) do not perceive themselves as at increased risk of cancer, (2) are not aware of the availability of genetic testing, (3) experience financial or time constraints that limit their ability to pursue genetic counseling and testing, (4) perceive no benefit from testing, (5) fear the potential consequences of genetic testing, e.g. discrimination by insurers and employers, stigmatization, and the psychological effect on themselves or family members, or (6) are too depressed to pursue it.

When genetic counselors were asked what they would do if at 50 percent risk of having HNPCC, 68 percent would not bill their insurance companies for genetic testing because of concern about discrimination and 26 percent would even use an alias, even though discrimination has rarely been reported (Matloff *et al*., 2000).

A 1993 survey of medical directors of 27 U.S. life insurance companies indicated that familial colon or breast cancer constituted sufficient grounds to deny insurance by only one company and to charge higher premiums by only six (Wong *et al*., 2001).

Of those offered genetic testing for colon cancer who decline, concern about discrimination is one of the leading causes. However, the actual risk of discrimination is very low; few cases have been reported. In spite of this, media coverage has raised sensitivity about this possibility, and the comprehensive Genetic Nondiscrimination in Health Insurance and Employment Act is currently in process by the U.S. Congress. The recently instituted Health Insurance Protection and Accountability Act (HIPAA), intended to secure confidentiality of medical records, may reassure some patients concerned about discrimination, but may also make more difficult the transfer of medical information between the providers of willing family members.

43. Are there legal issues regarding consent, ownership of data and/or samples, patents, licensing, proprietary testing, obligation to disclose, or reporting requirements?

Consent issues are the same with or without a large-scale program. While some patents related to colon cancer susceptibility testing have been issued, licenses to permit testing have been granted, as is routine in the testing industry.

If a large-scale program to systematically test incident cases for HNPCC was accomplished entirely through current channels, i.e. involving a patient's customary providers and payment mechanisms, no legal problems are anticipated. If other personnel were involved, such as agents of the government or a private entity contracted to carry out the program, then more persons would presumably have access to a given patient's result. Such a funding mechanism might carry with it a reporting requirement in terms of numbers of subjects tested and outcome, but there is no justification for disclosure of personal identity in this connection.