Conference Abstracts

Section 1: Clinical Practice and Policy

"Genetic Exceptionalism" and the Paradigm of Risk in U.S. Biomedicine

Nancy Press, Wylie Burke

Genetic exceptionalism refers to the view that genetic information is qualitatively different from other medical information in its potential to be used in a discriminatory manner and to be perceived as definitive, and in its implications for family members, and thus deserves special protections. We suggest that much of what is supposedly unique about genetic information is actually part of a paradigm of risk in U.S. biomedicine. The diagram below represents a conceptual and historical change within U.S. biomedicine from a tight linkage of medical diagnosis with troubling symptoms toward an increasing emphasis on the delineation of risk states.

At the left, a patient seeks care because of symptoms. Further along the continuum, a patient sees the physician feeling well but with the possibility of having a disease diagnosed. Moving right, less is known about the natural history of the disease detected or even the necessity of medical interventions. Still, prior to actual genetic testing, a patient is classified at risk on the basis of family history; although this risk prediction is imperfect, it presages the possibility that an individual without disease may consider herself unwell. It is only at the far right that risk is based on DNA testing.

The ability to identify disease early can lead to health advantages if effective treatment is available; this is the preventive health model. But the same risk identification carries significant individual burden to a growing class of the worried pre-ill and may generate various types of costs for health care providers and society when risk factors are poorly defined or preventive measures are of uncertain benefit. Our analysis indicates that this issue applies broadly in U.S. medicine. While genetics is not the source or special location of this problem, it may best be through the careful delineation and study of the ethical, legal, and social implications of genetic information that the problem is confronted and addressed.

The Meaning of Autonomy in Genetic Testing: Lessons From Qualitative Research About the Role of Partnership in Decision-Making

Gail Geller, Barbara A. Bernhardt, Neil A. Holtzman, Teresa Doksum, Ellen S. Tambor

Over the last decade, we have studied, both quantitatively and qualitatively, consumers' and providers' attitudes and behaviors regarding informed consent for genetic testing. In this talk, we will focus on the relationship between provider directiveness, patient autonomy, and shared decision-making, which is of particular interest as genetic testing moves into primary care. In our national physician survey, 44 percent of primary care physicians would give a recommendation to a cystic fibrosis carrier couple about undergoing prenatal diagnosis. In follow-up focus groups, many physicians stated that, because patients seek their guidance, nondirectiveness is an impossible and undesirable goal in primary care. In a second provider survey, 68 percent of physicians would make a recommendation about BRCA1/2 testing. In our survey of consumer attitudes toward BRCA1/2 testing, 78 percent of at-risk women would want their providers' recommendations about testing. In focus groups, we found

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that most women wanted a recommendation from a knowledgeable and trusted provider, but they still wanted the freedom to make their own decisions. Such recommendations may not be viewed by patients as directive, since patients do not feel obligated to follow them. When we offered BRCA1/2 testing to at-risk women with a nurse or genetic counselor providing education and counseling, 91 percent of subjects reported establishing a partnership with the provider, 100 percent felt that the provider listened to them, and 98 percent perceived complete autonomy in testing decisions. Patients seem to prefer a model of shared decision-making. A similar pattern emerged in our qualitative study of parents' and children's attitudes toward enrolling minors in genetic susceptibility research. Most children wanted to make decisions together with their parents, and most parents would involve their children in decision-making, although the child's age and the risk of the research would shift the balance of control. Our findings underscore the view that informed consent should be a process of shared decision-making that acknowledges the importance of partnership, whether between patient and provider or parent and child. Qualitative input helps to identify subtle variations in attitudes and practices regarding autonomy that facilitates the achievement of truly informed consent in the context of genetic testing.

BRCA1 Testing and Insurance Issues: Survey Evidence From Tested and Non-Tested Individuals

Cathleen D. Zick, PhD, Ken R. Smith, PhD, Robert N. Mayer, PhD, Jeffery R. Botkin, MD, PhD

Over the past decade, the development of genetic tests for inherited diseases has raised questions about whether insurers should have access to these test results. Consumers fear that insurers will use such information to deny coverage or raise rates to individuals who carry serious gene mutations. Insurance companies worry that if consumers are allowed to keep the test results private, then known carriers may obtain higher levels of insurance protection at prices that are below actuarially justified values. Both sides of the insurance and genetic testing debate have made assertions without extensive supporting data.

In our study, we surveyed two groups to assess the impact of genetic testing on insurance behaviors and attitudes. The first group includes individuals in a large kindred who are at risk of carrying a BRCA1 gene mutation that dramatically increases the female members' lifetime risk of developing breast and/or ovarian cancer. These individuals have been tested for this mutation and they know their test results. The second group consists of women in the general public who have not undergone testing.

We address the following questions: (1) Does testing positive for the BRCA1 gene mutation lead individuals to increase their life insurance and/or cancer insurance coverage above levels held by otherwise similar non-tested individuals? (2) Are those who test negative for the BRCA1 gene mutation more likely to share their test results voluntarily with health and/or life insurers than those who test positive? (3) Do tested and non-tested individuals differ in their opinions regarding how public policy should be structured to balance genetic privacy concerns against the insurance industry's concerns?

This paper highlights our major findings. Specifically, individuals who test positive do not purchase more life insurance than non-tested individuals, but they are more likely to have recently considered purchasing supplemental cancer insurance. Those who tested negative were significantly more likely than those who tested positive to communicate their test results to insurers. Finally, an overwhelming majority of tested and non-tested individuals support public policy that would emphasize the right to genetic privacy over an insurer's need to know genetic test results.

Categorizing Genetic Tests To Identify Ethical, Legal, and Social Implications

Wylie Burke, L. Pinsky, Nancy Press

Practice standards in medical genetics provide an implicit guide to the ethical, legal, and social implications (ELSI) of genetic tests. The common use of nondirective counseling reflects the principle that many testing decisions should be determined by personal values. Yet geneticists make testing recommendations in some settings, e.g., RET mutation testing when clinical findings suggest MEN2 and newborn screening for PKU. Conversely, many geneticists recommend against testing APOE alleles to predict Alzheimer disease (AD) risk. Taken together, these examples suggest that genetic tests can be categorized by a joint consideration of accuracy (i.e., clinical validity) and availability of effective treatment for persons who test positive. For genetic tests with high accuracy/no treatment (e.g., presymptomatic testing for Huntington disease), the predominant ELSI concern is adequate nondirective counseling prior to testing to ensure an informed, autonomous decision. By contrast, the predominant ELSI concern for tests with high accuracy/effective treatment (e.g., PKU) is ensuring access to treatment for eligible persons. APOE represents tests with limited accuracy/no treatment; these involve potential harm from adverse labeling but little if any benefit; the predominant ELSI concern is consumer protection. A fourth category raises more complex ELSI issues: tests with limited accuracy/effective treatment (e.g., HFE mutation testing for hereditary hemochromatosis). Here, net benefit is the issue: the balance between effectiveness of treatment (including safety) and potential harms from genetic labeling. Careful evaluation of all outcomes, including social outcomes, is needed before the appropriate use of testing can be determined. Where uncertainty exists concerning both accuracy and effectiveness of treatment, as in the case of BRCA1/2 testing, the

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analysis must take into account all potential outcomes of testing, including misassignment of risk status and exposure to unnecessary or unproven treatment; the concern for adequate pre-test counseling is similar to that for high accuracy/no treatment tests. This approach to test categorization, derived from implicit principles of medical genetics practice, allows a rapid determination of the predominant ELSI concerns for different kinds of genetic tests and identifies those tests for which outcome studies are most urgent.

Section 2: Research Practice and Policy

1999 NHANES: Sociodemographic Factors Associated with Consent to Future Research and to Genetic Research Among Adult Participants

Maria Agelli, Kathryn S. Porter, Geraldine M. McQuillan, Raynard S. Kington

The developments in molecular genetics and cloning of cancer-susceptible genes have increased the interest of professionals and the public in genetic testing. Many chronic diseases are influenced by heredity. Genetic research will help to identify those at risk and to prevent the development of diseases. However, perceptions and concerns on genetic research are largely unknown. How public health researchers and policymakers address these concerns will affect attitudes towards genetic research and health care. It is important to understand who consents to future research, including genetic research. NHANES surveys a nationally representative sample of the U.S. household population. African-Americans and Mexican-Americans are oversampled. Participants in the 1999 NHANES aged 20 years or older were asked permission for urine and blood samples to be kept for future and/or genetic research. The aim of this study is to evaluate the sociodemographic factors associated with consent. Of 1,947 adults, 84 percent gave consent to future and genetic research. Preliminary results (unweighted data) indicate that consent is not related to age ($p=.35$), education ($p=.19$), or spoken language ($p=.29$), but is related to ethnicity ($p<.0001$), with 74 percent of African-Americans giving consent compared with 86 percent of whites and Mexican-Americans, and to gender ($p=.03$), with 86 percent of men giving consent compared with 83 percent of women. The gender-related difference in giving consent was independent of age and ethnicity. Few studies have addressed either statements of intent or actual consent to genetic testing. These studies were limited to relatives of patients with familial cancers. This is the first report on actual consent to future genetic research in a general population representative of the United States. These data provide insight into the willingness of the general population to participate in future research, including genetic research.

Creating a Process to Collect Human Biological Materials and Medical Records for Research from Patients in Teaching Hospitals

Ellen Wright Clayton

For the last year, leadership of the Meharry-Vanderbilt Alliance has been developing a strategy to seek permission from patients at our institutions to use their biological materials and medical records for research. To this end, focus groups were conducted with Caucasian and African-American individuals to obtain greater insight into how the public might respond to such a process and to identify areas of concern that need to be addressed. Based in part on these results as well as the requirements of the Federal regulations for the protection of human subjects and the guidance provided by the National Bioethics Advisory Committee as well as other commentators, we are developing a process that includes public education and outreach, attention to informed consent, internal and community-based review processes for protocols, and the development of bioinformatics tools that permit exploration of the medical record while permitting investigators to receive only coded samples and information. We will present the results of the focus groups and the considerations that led us to create this process, which we will compare with some of the developments occurring elsewhere in the country, such as Web-based DNA solicitation, the creation of a commercial entity to explore the Framingham data and samples, and the reported arrangement between Duke, the Beth Israel Deaconess, and Ardais.

Lay Advocacy Group's Blood and Tissue Bank Accelerates Research

S.F. Terry, P.F. Terry, A.S. Marais, I.P. Ronchetti, C. Boyd, E. Johnson, L.G. Bercovitch

Lay advocacy groups can accelerate and enhance genetics research. Representing research participants, ethical and legal issues are of prime concern to these groups. A limited pool of willing participants, inadequate funding, and a competitive research environment can lead to redundant collections, poor confidentiality protections, and a variable informed consent process.

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Already isolated by diagnosis, participants feel "mined" for their biological samples and sense little regard for familial, social, or cultural issues.

Lay advocacy group involvement in sample collection offers a unique solution. With advice from ELSI, PXE International, the lay advocacy group for pseudoxanthoma elasticum (PXE), established the PXE International Blood and Tissue Bank. PXE International recruits individuals, engages them in a culturally sensitive, comprehensive, informed consent process, and then sends them a coded blood donation kit. Identifiers reside in a centralized database maintained by PXE International. A number of blood and tissue repositories, in various countries and communities, empower local groups to participate fully according to particular cultural needs.

Researchers interested in using archived DNA apply directly to PXE International. Requirements include institutional IRB approval, agreement to provide regular scientific and lay progress reports, assurance that samples will not be shared with other laboratories, and no contact with DNA donors or reporting of individual donor results. In addition, the researcher shares any intellectual property arising from the use of samples with PXE International. PXE International is a primary collaborator, and clinicians who contribute substantial amounts of DNA and tissue coauthor resulting papers. Because of regular reporting and discussion, PXE International contributes in a substantial and ongoing way to the intellectual and scientific discovery processes.

This method of distribution effectively separates identifiers from samples, provides a meaningful informed consent process, and ensures compliance with all oversight and regulatory guidelines. It provides access to a comprehensive archive of DNA for all interested, competent researchers. It maintains the integrity of various world and local communities, resulting in increased trust and collaboration internationally. It also empowers the affected individuals and the lay organization, and, most importantly, it helps keep the research focused on the patient.

Privacy and Confidentiality in the Publication of Pedigrees

Jeffrey R. Botkin, MD, William McMahon, MD, Ken Smith, PhD, Jean Wylie (Nash)

The publication of pedigrees in biomedical journals poses a threat to the privacy and confidentiality of family members. Although professional publications are intended to communicate research results to scientific colleagues, family members often read the medical literature and can identify their pedigree diagrams. These reports may reveal experimental genetic test results and personal health information within the family and to research subjects themselves. Our research published in 1998 (Botkin JR, McMahon W, Smith K, Nash J. Privacy and Confidentiality in the Publication of Pedigrees: A Survey of Investigators and Biomedical Journals. JAMA 1998; 279: 1808-1812) contributed to the assessment of this problem. We found that 36 percent of responding genetics investigators stated that family members were not informed that their pedigree would be published. Further, 78 percent do not obtain informed consent specifically for pedigree publication, and, when consent is obtained, it is often not from all family members depicted. In response to this problem, 19 percent of the investigators report the alteration of published pedigrees, and 45 percent who have altered pedigrees state that alterations are not disclosed to the publishing journal. Further, current practices by many biomedical journals are not in conformity with professional editorial standards. Improved informed consent or adherence to editorial standards are not adequate solutions to this complex problem. This presentation will expand on the current literature by highlighting existing problems and providing recommendations to investigators and journal editors to enhance the privacy and confidentiality of participants in pedigree research.

Section 3: Outreach and Education

The Science and Issues of Human DNA Polymorphisms: An ELSI Training Program for High School Biology Teachers

David Micklos, Scott Bronson

This DOE ELSI Project was a nationwide training program to introduce high school biology teachers to the key uses and societal implications of human DNA polymorphisms. During the term of the project, 256 high school faculty attended 12 2.5-day workshops conducted in 10 States. Each workshop mixed theoretical, laboratory, and computer work with practical and ethical implications. Program participants learned simplified PCR techniques for examining three types of human DNA polymorphismsCa VNTR repeat, an Alu insertion, and mitochondrial (mt) point mutations. These polymorphisms illustrate the use of DNA variations in disease diagnosis, forensic biology, and identity testing and provide a starting point for discussing the uses and potential abuses of genetic technology.

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These experiments are unique, in that they allow students to use their own DNA polymorphisms as the starting point of far-ranging inquiries into human population genetics and evolution. Alu insertion polymorphisms are analyzed entirely in the classroom, while mt DNA samples are sent to the DNA Learning Center for sequencing. In spring 2000, Applied Biosystems provided us with a 377 DNA Sequencer and reagents to support a free sequencing service for student mt DNA samples submitted from around the country. To our knowledge, this is the only shared DNA sequencing facility dedicated entirely to education.

Laboratory protocols, reagent recipes, supporting animations, and video interviews with molecular anthropologists are all available at our Genetic Origins World Wide Web site (vector.cshl.org/geneticorigins/). Student Alu and mt sequence data are posted to online databases at the site, which include reference data sets from world populations. Students then use online analysis tools, including chi-square, genetic distance, and multiple sequence alignment, to answer their own questions about the relatedness of world populations. By analyzing the worldwide distribution of Alu insertions, students consider competing theories of the emergence of modern humans. By analyzing mt mutations, students determine Neanderthal's position in the hominid family tree. Students may also use their own DNA sequences to identify related sequences in Genbank, providing a direct link to the world of genomics research.

Cultural and Linguistic Considerations in Development of Genetic Educational Materials in a Predominately Mexican-American Population in South Texas

Maricela C. Aguilar, MSN, RN, Peggy Visio, MS, RD, LD, Sara Kolb, PhD, RN, Judith Livingston, M.Ed., Cynthia Aguirre, BA, Celia Kaye, MD, PhD

Problem: Children in South Texas, of whom 76 percent are Mexican-American (MA), fail to comply with recommendations for genetic services. Over 63 percent of families of children with spina bifida (SB) are not compliant with recommendations for urinary care; 65 percent of children with SB are obese; and 53 percent of children with metabolic disorders and 72 percent of children with diabetes mellitus are in poor dietary control.

Sample Population: A sample of 27 MA families was selected on the basis of clinic attendance rate of 80 percent and poor compliance with medical recommendations.

Methodology: Each family was assessed for Spanish/English health literacy, values/attitudes, and preferred learning methods. The Test of Functional Health Literacy in Adults (TOFHLA) was used to assess literacy. A Family Learning Preferences (FLP) tool inquired about previous teaching received by the family, and preferred ways of learning. Family-specific culturally based health factors were assessed by use of the Health Beliefs Questionnaire - Revised (HBQ-R).

Results: The TOFHLA scores indicated that 31 percent of the subjects interviewed had an inadequate to marginal functional health literacy level, while 69 percent of subjects had an adequate functional health literacy level. The FLP indicated that the best liked way of learning was in small groups (4-5) and by videotapes and demonstration. The least liked way of learning was in large groups (10 or more), by written materials, and by teaching done by other parents. Use of the HBQ-R Likert Scale indicated statistically significant positive correlations between acculturation and education ($r=.648$) and between literacy and education ($r=.460$) and a negative correlation between folk beliefs and education ($r=-.570$).

Conclusions: (1) Although literacy was correlated with educational achievement, 20 percent of individuals with 10th grade or higher education level demonstrated low literacy, (2) although acceptance of folk beliefs was negatively correlated with educational level, 50 percent of individuals with 10th grade or higher education level demonstrated a folk belief score of 3 or more on a scale of 1 to 5, and (3) preliminary findings suggest that attention to reading level and preferred learning styles is needed for development of culturally/linguistically appropriate genetic educational materials to ensure successful outcomes.

This project is supported by MCHG Project 97-11.

A Practice-Based Genetics Curriculum for Nurse Educators

Dale Halsey Lea, RN, MPH

Project Overview: Effects of the Human Genome Project are already having a major influence on health care. Primary care providers, including nurses, will be increasingly challenged to integrate new genetics knowledge into their practice in order to ensure that those patients and families affected with genetic-related health conditions receive quality genetic health services. Nurse educators must be prepared with knowledge and skills to educate and support nurses in managing challenges in the new era of genetic health care.

Research Focus: A 3-year educational research project was designed to address an identified lack of knowledge of genetic principles and their application to nursing practice, and barriers to the inclusion of genetic principles into nursing curricula. During year 1, four teacher-assisted modules that focus on practical, case-based genetics materials were developed to assist

nurse educators to integrate the principles of genetics and health promotion into existing nursing curricula. Topics of the modules include ethical, legal, and social issues of genetic testing, high-risk pregnancy and prenatal diagnosis, preconception prevention and prenatal testing, and presymptomatic diagnosis using breast cancer as an exemplar. Each of the modules includes didactic information, case studies, genetic and family history, risk assessment tools, handouts, factsheets, PowerPoint materials, Internet resources, other professional and client resources, and evaluation questions and activities. Educational materials are leveled for undergraduate and advanced practice students.

Field-Testing: During year 2 nurse educators from four schools of nursing within northern New England and 23 schools of nursing that serve a broader population of urban and minority students were enlisted to participate in field-testing the modules. The development and evaluation of the practice-based genetics modules, education and resource materials and evaluation methods included in the modules, and results of the education and training seminar held to educate and support participating field-testers were evaluated.

Evaluation: Evaluation of year 1 focused on module development and the education and training program for nurse educators. Evaluation of year 2 included results of faculty interviews and student response from field-testing. Faculty response to the genetics modules has been overwhelmingly positive. Field-testers have found the practice-based genetics materials to be well organized, informative, easy to use, easy to integrate into existing curricula, and well received by students. The curriculum is now available for distribution.

Developing a Case-Based Ethics Curriculum for the Biotechnology Industry

Margaret Eaton, JD, PharmD, David Brady, PhD, Barbara A. Koenig, PhD

Biotechnology business managers will bring the discoveries of human genome research into the marketplace. Without their efforts, the benefits of research will not be realized. However, developing and marketing the medical products of biotechnology have not and will not be easy. On the one hand, corporate managers have a duty to the company and its shareholders to maximize corporate value. On the other hand, biotechnology products, such as genetic tests, may create profound ethical dilemmas for patients and for society as a whole. This project — a collaboration between Stanford's Center for Biomedical Ethics and Graduate School of Business — is developing a case-based ethics curriculum targeted to industry.

Currently, many corporations deal with ethical issues only when management is forced to revise or rescind actions that have harmed or offended patients, health care providers, government regulators, or the public. When a company deals with ethical issues after the fact, it loses opportunities to prevent problems. Another way of addressing these issues is to include them prospectively in corporate decision-making.

The purpose of this project, then, is to teach the fundamentals of business ethics to managers, to encourage them to incorporate ethical analysis into decision-making, and to teach them how to do so. The primary mechanism of teaching is detailed case analysis, a standard approach in business education. Our project is developing 10 studies (see below), which were refined during pilot-testing in a course offered through Stanford's Graduate School of Business. The case studies — which will be published via the Internet and included in a book — were selected to cover the full spectrum of new product development in biotechnology, from basic research through marketing. Courses based on the project will be used for business school education and for executive education programs.

Examples of questions addressed in the cases include the following: Is it appropriate to conduct human research in countries with few human subject protections? Is it premature to market a gene test when no preventive or treatment measures are available? What kind of patient information or labeling materials should be marketed with a product; is direct-to-consumer marketing acceptable? The presentation will highlight one case: marketing genetic tests for breast cancer susceptibility.

Section 4: Genetic Privacy and Discrimination

Experiences, Attitudes, and Beliefs of Persons with Genetic and Other Serious Illnesses Concerning Privacy, Confidentiality, and Access to Health Insurance

Nancy Kass, Sara Hull, Marvin Natowicz, Ruth Faden, Larry Gostin, Julia Slutsman, Neil Holtzman

Background: Despite policy attention to medical privacy and access to health insurance, little empirical work exists in these areas documenting and comparing experiences of persons with genetic and other illness.

Purpose: The goal of this cross-sectional interview study was to document and compare the experiences, attitudes, and beliefs of persons with strictly genetic conditions — cystic fibrosis (CF) or sickle cell disease (SCD) — and persons with other

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chronic conditions — HIV infection, diabetes, breast cancer, and colon cancer — in terms of privacy, confidentiality, and health insurance.

Methods: Interviews were conducted once with 597 affected adults, parents of affected children, or persons at risk for hereditary cancers. Quantitative and qualitative data were collected.

Results: Respondents with HIV infection were less likely than others to disclose their condition to others. Among all respondents, 13 percent regretted telling at least someone, while 75 percent reported being glad. One-quarter of respondents had discussed confidentiality with their physicians, although 72 percent believed physicians would protect their confidentiality; 6 percent of respondents overall had not told a health care provider about their condition, compared with 20 percent of those with HIV ($p<.001$). The majority believed that most people have little power to stop health insurance companies from getting their medical information (64 percent).

Respondents with HIV infection were less likely to have health insurance (80.0 v. 95.0 percent overall, $p<.001$). Those with CF and SCD were more likely to have been denied insurance or had been offered it at an unaffordable rate (40.8 and 32.3 percent v. 26.6 percent overall, $p<.001$). Almost one-half (43 percent) have felt they could not leave their jobs because they would lose their insurance; this was particularly true for those with HIV infection (68 percent) or CF (43 percent) (overall, $p<.001$).

Conclusions: Persons with serious illnesses trust providers with medical information but are less willing to trust insurers. While most were insured, many have been offered policies at unaffordable rates, and almost one-half feel locked into their jobs because of insurance.

A Pilot Study of Genetic Privacy, Discrimination, and Informed Decision-Making

Mary Davidson, Lois Lander, Janine Lewis, Sharon Terry, Vicky Whittemore

Recent reports suggest that individuals are facing genetic discrimination in health care, insurance, and employment decision-making because of identification with a genetic condition or predisposition. There is widespread concern that current State and Federal laws do not adequately protect against misuse and unwanted disclosure of personal genetic information and test results. This will be especially true as more tests to identify both common and rare conditions are developed with the continued sequencing of the human genome. On the State and Federal levels, consumer and professional groups are advocating for legislation that will safeguard the privacy of genetic information and provide genetic nondiscrimination protection.

Despite abundant anecdotal reports, limited documentation of genetic discrimination inhibits development of health care and legislative nondiscrimination policies. A discrimination questionnaire was developed and disseminated through the 300 Genetic Alliance member groups and their constituents and on the Alliance Web site. Its purpose is to document incidents of genetic discrimination and privacy abuse.

The questionnaire was also designed to determine whether people are told, before making a decision about genetic testing, about the risks of misuse and unwanted disclosure of test results, as well as the benefits of being tested. In addition, the questionnaire sought to reveal how fear of employers, insurance companies, and other entities accessing and using test results affects the public's decisions to participate in genetic testing and research.

An overwhelming response to the questionnaire illustrates discrimination in areas including school admission, employment, military, and health, disability, and life insurance. Sixty-six percent of these respondents agreed to participate in follow-up telephone interviews conducted by trained Alliance staff and/or genetic counselors. These cases are added to a Story Bank Database without identifying information. Some respondents are willing to share their stories in public arenas, providing an invaluable resource to policymakers. A follow-up survey is ongoing to further understand the extent and nature of health insurance discrimination.

Validation of the survey instrument and research will be conducted as a result of the data from this study.

Genetics, Ethics, and the Law: Results from Case Studies in Maryland, Michigan, and California

Lawrence O. Gostin, James G. Hodge, Jr., Cheye M. Calvo

The existing and future proliferation of individually identifiable genetic data in society raises complex legal and ethical issues, many of which center on the dual interests of protecting individual privacy and preventing discrimination. Whereas the Federal Government has failed to enact comprehensive genetic-specific laws to date, many State legislators and executive policymakers have created genetic-specific privacy and antidiscrimination laws and regulations (often focused on the uses of identifiable genetic data by government, employers, insurers [health, life, and disability], and medical providers and researchers). These laws vary widely among States according to their subject matter, framework, intent, and sophistication.

What do State policymakers perceive as their role in protecting individual genetic privacy? Why have legislators and

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policymakers focused extensively on genetic privacy concerns? How have States differed historically and in modern times in their approach to regulating genetic privacy?

To attempt to answer these and other questions, we conducted comprehensive case studies in three States (Maryland, Michigan, and California) with divergent approaches to regulating genetic data and technologies. These case studies involved systematic reviews of State genetics laws, interviews with key legislators and high-ranking executive branch officials, and comprehensive analysis. The results of these studies present a fascinating look into the motivation and intent of State genetics policymakers from legal, ethical, and public policy perspectives. This presentation will examine these study results through narrative and statistical information explaining the legal approaches of these States.

A State Model for Public Policy in Genetic Testing and Screening: A Report from the New York State Task Force on Life and the Law

Ann M. Carroll, Aytan Y. Bellin, Dana H.C. Lee, Carl H. Coleman

State government is a primary locus for legislative, regulatory, and policy initiatives that address concerns associated with predictive genetic testing. The New York State Task Force on Life and the Law, a State bioethical policy commission that considers issues that arise from technological advances that affect health care, recently completed a 3-year study of States' roles in genetic testing and screening issues. The study, funded in part by the National Human Genome Research Institute's ELSI program, will result in the release of a comprehensive report, scheduled for winter 2000. The Task Force report focuses on the predictive uses of clinical genetic testing to detect future health risks for apparently healthy individuals and their offspring. It examines a broad range of issues including confidentiality of genetic information, informed consent for genetic testing, clinical integration of predictive genetic testing and screening, genetic testing of children, use of clinical tissue samples for genetic research, use of genetic information in insurance underwriting, the role of health care providers in the delivery of genetic services, and the State's role in newborn screening and oversight of genetic test approval and laboratory quality assurance. The report makes definitive practice, regulatory, and legislative recommendations on these issues, including recommendations to amend existing New York State legislation for confidentiality and informed consent for predictive genetic testing. Two major and parallel themes emerged from the project. The first theme is the enormous potential that predictive genetic testing poses for health care. This led the Task Force to identify barriers to appropriate testing and research, including concerns about potential misuses of genetic information against individuals, and to recommend approaches to reduce these barriers. The second theme is that, despite the potential promise of predictive genetic testing, there are potentially dangerous misunderstandings and misperceptions within the general public about the role of genes in health, disease, and behavior. This led the Task Force to call for measures to improve genetics education, ensure appropriate genetic counseling, limit the use of genetic screening, enhance oversight of genetic research, and increase community involvement in policy decisions.

Section 5: Genetic Screening and Testing

Factors Influencing the Diffusion of Genetic Tests

N.A. Holtzman, B.A. Bernhardt, T. Doksum, G. Geller, E. Mountcastle-Shah, M.M. Schoonmaker, E.S. Tambor

Over the last decade, we have conducted several studies on the diffusion of new genetic tests, focusing on practitioners' interest in offering the technology, patients' interest in using it, and insurers' willingness to cover it. In an early study, we found that recent medical graduates and those in specialties more likely to deal with genetic problems had greater knowledge of genetics and genetic tests than others. However, this study could not assess the relationship between an individual physician's knowledge and his or her adoption of new genetic tests. Using logistic regression, we subsequently assessed factors that affect obstetrician-gynecologists' (OB-GYN) adoption of cystic fibrosis carrier screening (CFCS) and OB-GYNs' and oncologists' adoption of BRCA1/2 testing. Adoption was measured by whether physicians discussed the test with their patients or ordered the test. Physicians' knowledge was associated with OB-GYNs ordering CFCS and oncologists ordering BRCA1/2 testing. No association was found between physicians' knowledge and their discussing either test with their patients. No other variable was consistently associated with adoption. We also interviewed 60 physicians. Their most frequently mentioned reasons for not adopting new predictive genetic tests were uncertainty about the tests' clinical validity and utility.

We also studied factors influencing patients' adoption of new technologies. In studies in which we offered free CF carrier testing and breast cancer susceptibility testing, uptake was low (7 and 13 percent, respectively). Convenience appeared to influence uptake in both cases. Tolerance for test uncertainty and fear of stigma were also significant predictors of uptake of CF

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carrier screening. The need to involve an affected relative in BRCA1/2 testing and the questionable validity of the test were barriers.

Our survey of health insurers indicated that the validity of genetic tests and the safety and effectiveness of new therapies were important influences on their coverage of hypothetical genetic technologies. The findings were somewhat discrepant from insurers' actual coverage decisions.

We will present an innovative model of diffusion of new genetic technologies that was generated by these data.

Expanded Newborn Screening for Metabolic Disorders: A Giant Leap Forward in Preventive Medicine or Pandora's Box?

Susan Waisbren, PhD, Simone Albers, MD, Deborah Marsden, MD, Isabel Bailey, LICSW, Laura Finkelson, PhD, Debby Lobbregt, Mark Korson, MD, Harvey Levy, MD, and the New England Consortium of Metabolic Programs

The technology of tandem mass spectrometry (MS/MS), recently adapted to the dried blood specimen collected for newborn screening, allows for the detection of many biochemical genetic disorders heretofore not identifiable in the newborn specimen. In 1999, within the New England Screening Program, Massachusetts instituted a pilot program using MS/MS, adding 20 biochemical genetic disorders to the traditional screening panel. Maine added only medium chain Acyl-CoA dehydrogenase deficiency (MCADD). The other New England States are considering expanding their screening mandate. With technology as the driving force, there is the possibility of expanding screening for an even larger number of genetic disorders. It is important to study the ethical, social, and legal implications of these programs now when they are new and still amenable to change.

The New England Consortium of Metabolic Programs, a regional organization of health care professionals at all levels involved in identifying and treating biochemical genetic disorders, is examining the impact of expanded newborn screening. The research is in collaboration with Neo Gen Screening, which conducts expanded newborn screening in Pennsylvania. Four naturally occurring study groups exist: children identified through MS/MS, children whose disorders are diagnosed clinically, children for whom the initial screening diagnosis was false positive, and a control group of children for whom the newborn screen was normal. Parent interviews take place 6 months after the diagnosis is made and then again a year later, and, for the children with metabolic disorders, medical examinations and developmental testing are performed. Parents report on their experiences at the time of diagnosis in terms of how they responded to the information received, their subsequent interactions with health care systems, and their family's adjustment.

Enrollment to date includes 16 patients identified by MS/MS, 5 siblings of screened patients, 12 patients identified clinically, 4 false positives, and 22 controls for whom the newborn screen was normal.

This study assesses not only the health outcome of children, but also the impact of the diagnostic process in terms of parental stress and experiences with the health care system. Potentially, this study could be a model for addressing the ethical, legal, and social concerns about newborn genetic screening.

On the Restriction of HFE Testing for Hemochromatosis: Results of a Nationwide Survey of Clinical Laboratories

Jon F. Merz, Antigone Kriss, Debra G.B. Leonard, Mildred K. Cho

The HFE gene was discovered in 1995 by Mercator Genetics, and patents covering diagnostic tests for the gene were issued beginning in early 1998. The patents were licensed exclusively for clinical testing purposes to SmithKline Beecham (SB) in mid-1998. SB began enforcement that summer by notifying laboratories that SB held exclusive rights to the patents, and offering sublicenses. In summer 1999, we ran an IRB-approved survey of clinical laboratories throughout the United States. We identified 117 laboratories in the Genetests database and AMP Test Directory and used snowball sampling to increase our sample to 128 laboratories that could be offering clinical HFE testing. We (AK) contacted 121 of these laboratories by phone, had 9 refusals, and completed surveys with 112 respondents (87.5 percent). We find that 60 percent of laboratories reported performing the HFE genetic test at that time; 19 percent of respondents reported not developing the test and 4 percent reported that they had stopped doing the test after developing and validating it in their laboratory, at least in part because of the patents. These data regarding first discovery, publication, patenting, and time to test adoption are consistent with the model proposed in Merz, Clinical Chemistry 1999; 45: 324-330. We find a mean time to adoption of the test of 14 months from first publication, and more than half the laboratories reported adopting the test before the first patent issued. The implications of these findings are discussed.

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Risk Estimation and Value-of-Information Analysis for Three Proposed Genetic Screening Programs for Chronic Beryllium Disease Prevention

Scott M. Bartell, Rafael A. Ponce, Tim K. Takaro, Richard O. Zerbe, Gilbert S. Omenn, Elaine M. Faustman

Genetic differences (polymorphisms) among members of a population are thought to influence susceptibility to various environmental exposures. In practice, however, incorporation of this information into quantitative risk assessment and risk management is rarely accomplished. We describe an analytic framework for predicting the risk reduction and value of information (VOI) resulting from specific risk management applications of genetic biomarkers and apply the framework to the example of occupational chronic beryllium disease (CBD), an immune-mediated pulmonary granulomatous disease. One described human leukocyte antigen gene variant, HLA-DPb1*0201, contains a substitution of glutamate for lysine at position 69 that appears to have high sensitivity (~95 percent) but low specificity (~70 percent) with respect to CBD among individuals occupationally exposed to respirable beryllium. The expected post-intervention CBD prevalence rates for using the genetic variant (1) as a required job placement screen, (2) as a medical screen for semiannual in place of annual lymphocyte proliferation testing, or (3) as a voluntary job placement screen are 0.08 percent, 0.8 percent, and 0.6 percent, respectively, in a cohort with 1 percent baseline CBD prevalence. VOI analysis is used to examine the reduction in total social cost, calculated as the net value of disease reduction and financial expenditures, expected for proposed CBD intervention programs based on the genetic susceptibility test. For the example cohort, the expected net VOI per beryllium worker for genetically based testing and intervention is $13,000, $1,800, and $5,100, respectively, based on a health valuation of $1.45 million per CBD case avoided. VOI results for alternative CBD valuations are also presented. Despite large parameter uncertainty, probabilistic analysis predicts generally positive utility for each of the three evaluated programs when avoidance of a CBD case is valued at $1 million or higher. Although the utility of a proposed risk management program may be evaluated solely in terms of risk reduction and financial costs, decisions about genetic testing and program implementation must also take into consideration serious social, legal, and ethical factors.

Quality Assurance Practices in Clinical Molecular Genetic Testing Laboratories

Margaret M. McGovern, Marta Benach, Sylvan Wallenstein, Joe Boone

It has been suggested that specific regulation of clinical molecular genetic testing laboratories is required to ensure personnel standards and quality assurance and to safeguard patients' rights with regard to informed consent and confidentiality. However, an assessment of the current practices of such laboratories, which is important for determining the need for regulation and the impact it would have on access to testing, has not been conducted. To collect and analyze information about the personnel qualification and the clinical and clinical laboratory practices of laboratories that offer molecular genetic testing, a survey study of molecular genetic testing laboratory directors was conducted and a quality assurance score was assigned on the basis of responses to specific testing process items that were derived from the American College of Medical Genetics Laboratory Practice Guidelines. Although all directors had doctoral degrees and the majority were board certified, variability in the qualifications of technical personnel was found. Quality assurance scores were related in a statistically significant manner with test menu size, number of tests performed annually, director's degree, laboratory setting, and participation in a proficiency testing program. Lowest scoring laboratories had an overrepresentation of research-based laboratories and non-board-certified directors. Seventy percent of laboratories provide access to genetic counseling, 69 percent have a policy about confidentiality, and 45 percent require informed consent. The findings of the study suggest that a substantial number of laboratories were practicing at substandard levels, with laboratories in a research setting and those with non-board-certified and/or non-Ph.D.-trained directors having the most serious quality assurance deficiencies. The majority of directors recognize that genetic counseling, informed consent, and confidentiality are integral to the molecular genetic testing process.

Section 6: Neuropsychiatric and Behavioral Genetics

The REVEAL Study: Developing Cumulative Risk Profiles to Provide Susceptibility Genotyping, Risk Assessment, and Counseling for Alzheimer Disease

R.C. Green, N.R. Relkin, P.J. Whitehouse, L.A. Farrer, L.A. Cupples, S.G. Post, A.D. Sadovnick, T.C. Brown, S.A. LaRusse, M.J. Barber

Genes and other markers for complex diseases are rapidly being identified, and presymptomatic risk estimates are now possible. The REVEAL Study (Risk Evaluation and Education for Alzheimer Disease) is an NIH funded project to examine the

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impact of providing risk assessment, including apolipoprotein E (APOE) genotyping, for Alzheimer disease (AD). REVEAL investigators are using the well-established genetic epidemiology of the disease, including the risk attributable to different APOE polymorphisms, to develop risk profiles. Twelve cumulative risk curves have been generated (six for each gender) to portray APOE genotype and age-specific risks for first-degree relatives. Asymptomatic adult children of AD patients will be randomized into one arm, in which risk assessment is based upon age, gender, and APOE genotype, or a control arm, in which risk assessment is based upon only age and gender. Genetic counselors will communicate these risks (including APOE disclosure) and follow subjects to determine the psychological and practical impact of this information. REVEAL will be the first study to explore susceptibility genotyping, risk assessment, and counseling in an age-related degenerative disease, and results will inform policy debates and future guidelines about the advisability of such disclosures. We will present the risk curves and describe our approach to the development of research in this controversial area.

Public Beliefs About the Causes of Mental Illness and Possible Consequences of Adopting More Genetic Views

Jo C. Phelan, Rosangely Cruz Rojas

At this time, families affected by mental illnesses are seriously stigmatized. We are currently undertaking a study to assess how this stigma may change as new discoveries are made about genetic bases of mental illnesses. Will stigma be reduced, because mental illnesses are seen as more biological and thus an illness like any other? Will stigma become magnified, because genetic influences make mental illnesses seem more immutable and inextricably attached to the affected family?

At this point in the study, we have conducted semi-structured interviews with approximately 50 people of varying ethnic backgrounds in New York City and Los Angeles. In the interviews, we assess respondents' understanding of processes of genetic inheritance. We also describe two hypothetical individuals, one suffering from schizophrenia and one from major depressive disorder, and ask a series of questions about the problem, the person with the problem, and the person's family. We then introduce the idea that the illness is influenced by genetic factors and reassess respondents' attitudes in the light of this information.

We will present data on the following questions: (1) To what extent do respondents' understanding of genes, heredity, and how characteristics are passed from parents to children conform with current scientific understanding of genetics? (2) Based on responses to the vignette descriptions, what do respondents believe causes schizophrenia and major depressive disorder, (3) When told that an expert has said that, in the particular case described in the vignette, genetics or heredity probably played a big role in causing the illness, do respondents consider this explanation to be plausible? (4) Do respondents indicate that their attitudes toward the vignette subject are affected by the statement that the illness was strongly influenced by genetic factors?

These data will provide a preliminary indication of the current context, in terms of public knowledge and beliefs, in which emerging information about genetics is arriving. For example, how receptive are people to the notion that mental illnesses are influenced by genetic factors, and what does that mean to them? Further, what are the possible consequences of adopting such a belief for public attitudes toward individuals and families affected by these illnesses?

Expectations for and Anticipated Implications of Genetic Applications for Managing Mental Illness: Voices of Patients and Psychiatrists

E.M. Petty, K. Neudoerffer, A.C. Madeo, L.B. Smith, T.E. Jayaratne, A.E. Cassidy, K.K. Milner

Individuals with chronic persistent mental illness may experience suffering related to psychiatric symptoms and, unfortunately, the stigmatization/discrimination they encounter. Family, twin, biochemical, and genetic analyses support a genetic contribution to mental illness. Some alterations (e.g., del 22q) have been associated with psychiatric problems, but the etiology of most common psychiatric disorders is complex, limiting current knowledge about and management of them. This, along with the disease-associated morbidity, fuels the undeserved fears, stereotypes, and prejudices currently associated with mental illness. At a recent genetics conference (ASHG 2000), one predicted outcome of the genome effort noted by Francis Collins (NHGRI Director) was that the diagnosis and treatment of mental illnesses will be transformed, shifting societal views, in the next few decades. Although providers in Michigan rarely refer clients for genetic services, some clients have independently sought our genetic services — even requesting prenatal and/or predictive genetic tests for specific psychiatric disorders. To better understand the genetic knowledge, related expectations, and anticipated implications that patients, their families/friends, and mental health care providers have regarding genetics, we administered a survey to them (n=206 clients/advocates; n=213 providers). Most felt that genetic and nongenetic factors contributed to mental illness. Many clients/advocates saw potential benefits of increasing genetic knowledge but lacked a clear understanding of genetic concepts and available genetic tests. Providers were generally more knowledgeable but did not anticipate as many potential benefits. Most clients wanted more information about

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genetics from their psychiatrist/therapist, especially on recurrence risk and future therapies. Most providers did not seem to realize that many clients/advocates might be so interested in, and hopeful for, genetic advances. Several individuals in both groups noted that one major implication might be decreased stigmatization. Others anticipated negative implications related to reproductive choices and eugenic policies. If predictions are correct, increased genetic knowledge and technology will enhance diagnostic, prognostic, and therapeutic options in psychiatry within the foreseeable future. Further exploration of the desires and needs of the people who will be directly affected by these applications and related implications is needed when considering what kinds of health care and educational initiatives should be developed to reduce adverse outcomes.

The Fate of Genetic Optimism: Framing Behavior and Genetics in the News

Peter Conrad

The news media are a major source of public understanding of genetics and a strong influence on public discourse. This paper is based on a systematic study of the reporting of genetics in the news in five American newspapers and three news magazines from 1965 to 1995 (funded by ELSI). The paper examines the framing of the news about genetics for two cases: mental illness and homosexuality. The focus is on the structure of what I call the frame of genetic optimism, which dominated the reporting of genetics and mental illness (i.e., bipolar illness and schizophrenia) beginning in the mid-1980s. The structure of the frame comprises three elements: A gene for the disorder exists; it will be found; and it will be good. New discoveries of genes were announced with great fanfare, but the most promising claims could not be replicated or were retracted in short order. Despite these disconfirmations, genetic optimism persisted in subsequent news stories. I contrast this with the case of the American and British press' reporting of Dean Hamer's 1993 and 1995 finding of a gay gene. In this case, the American press presented the science of the research in detail and framed the story with cautious optimism; i.e., there could be problems but this is good science and the naturalization of homosexuality is likely to be good for gay men. The British press framed the story as the perils of the gay gene, emphasizing concerns about potential discrimination, misuse of the research, and the dangers of eugenics. There was considerable skepticism about the science as well. The British presentation contrasted with genetic optimism. These cases are briefly contrasted with the reporting of genetics concerning alcoholism and intelligence. Based on this research, we can assess the implications of the prevalent use of the genetic optimism frame in the American press in terms of public discourse and understanding of genetics. The paper concludes with a discussion of the sources, shifts, and fate of the genetic optimism framing and asks what alternative frames might be considered.

Understanding and Assessing Claims of Genetic Influence on Criminal Behavior

Robert Wachbroit, PhD, and David Wasserman, JD

This presentation will discuss some of the work done under an ELSI-funded project on the meaning and significance of claims of genetic influence on criminal, violent, and antisocial behavior. The apparent success of genetics in understanding human disease suggests that it could be a powerful tool in the scientific investigation of human behavior, including criminal behavior. At the same time, the checkered history of genetics suggests that it can easily be abused, misrepresented, or misunderstood, regardless of the validity of the studies or the motivation of the researchers.

This project brought together scholars from a variety of disciplines, including philosophy, history, ethics, law, and genetics. These scholars met in three working groups to present papers on behavioral genetics, criminality, and moral responsibility. Those papers provided the intellectual framework for a highly visible conference, which brought together leading critics and advocates of genetic research on crime to discuss its scientific value and social impact. The papers were widely cited at the conference, and several were revised in light of the discussion they provoked. The project investigators have edited an anthology, *Genetics and Criminal Behavior: Methods, Meanings, And Morals* (Cambridge University Press 2001), which contains a large subset of those papers, significantly revised and supplemented by additional contributions. This presentation will offer an overview of the two sets of issues on which the anthology focuses, issues central to the appraisal of current research into the genetics of criminal behavior:

1. What are the assumptions about criminal behavior, causation, and scientific explanation underlying research programs that seek genetic factors in criminal behavior, and are they justifiable? Given the widely acknowledged importance of environmental factors, and the fact that crime is a social category, what can a genetic investigation possibly tell us about criminality?
2. How would credible evidence of genetic influence on criminal behavior affect our practices of blaming and punishing? Would such evidence compel us to revise our conceptions of moral and legal responsibility or modify our assessment of particular agents?

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Section 7: Cancer Genetics

Use of an Interactive Computer Program to Educate Women About Genetic Testing for Breast Cancer Susceptibility

M.J. Green, B.B. Biesecker, A.M. McInerney, N. Fost

Purpose: To compare education by an interactive computer program with one-on-one education and counseling by a genetic counselor, assessing their effects on knowledge of breast cancer genetics and intent to undergo genetic testing.

Methods: Women considering genetic testing for breast cancer susceptibility who had a first-degree relative with breast cancer were randomized to undergo education by genetic counselor or interactive computer. Twenty-nine received individualized counseling about breast cancer risk and genetic testing from a genetic counselor, and 29 received education from an interactive computer program, followed by individualized counseling. Knowledge of breast cancer genetics was measured at baseline for 14 controls and after individualized counseling or computer use for experimental groups. Intent to undergo testing was measured at baseline and after the interventions. Preferences for interacting with the counselor or the computer were elicited.

Results: Controls correctly answered 74 percent of knowledge questions, while those educated by a genetic counselor correctly answered 92 percent and those educated by computer, 96 percent. This confirms our hypothesis that both counselor and computer-based interventions can significantly improve knowledge over baseline ($p<.0001$). Further, unadjusted knowledge scores were significantly higher in the computer group than in the counselor group ($p=.047$). Education and counseling affect intent to undergo testing. Before either educational intervention, a majority of participants in both groups (69 percent) indicated they were likely (definitely + most likely) to undergo testing if a genetic test were offered to them that day, whereas following either educational intervention, only 44 percent indicated that they were likely to do so; that is, participants were 2.9 times more likely (95 percent CI, 1.7-4.9) to say they would be tested before education than after ($p=.0002$). Participants preferred interacting with a genetic counselor to help them make decisions and to receive psychosocial support, but they were neutral or preferred the computer for its time efficiency, convenience, and the provision of factual information.

Conclusions: An interactive computer program can successfully educate patients about breast cancer susceptibility and can influence patients' intentions to undergo genetic testing. This can free counselors to use their time providing psychological support and individualized risk assessment.

BRCA1 Mutation Testing and Changes in Psychological Distress: Who Is Most at Risk of Adverse Effects?

Ken R. Smith, Jeffrey Botkin, Robert Croyle, Heidi Hamann, Anna Chan, Antoinette Stroup, Jean Wylie, Lee Ellington

An anticipated consequence of genetic testing for cancer susceptibility has been the effect that test results would have on the psychosocial well-being of tested individuals. Many predicted an increase in psychological distress among persons testing positive and a commensurate decline for members of high-risk families who receive negative results.

Recent studies have concluded that the adverse consequences of genetic testing for cancer susceptibility have been small and short-lived. While differences in mean levels of psychological distress have not proven to be large between those at elevated genetic risk for cancer and those lacking such a risk, far less is known about subgroups that may be especially susceptible to elevated psychological morbidity attributable to genetic testing.

We draw upon the experiences of a large kindred that has undergone genetic testing for a BRCA1 mutation. Subjects have been tested and were interviewed before receiving their test results along with four follow-up interviews, with the last 2 years after the receipt of test results.

The primary research question asked in this analysis is whether certain subgroups demonstrate greater susceptibility to the possible adverse psychological effects of testing positive for a BRCA1 mutation. Using the full Impact of Event Scale (IES) as a measure of distress, we find that mutation carriers have higher initial levels of distress 1 week after receipt of test results. This difference does not change with time. However, individuals who initially (before testing) did not suspect that they were mutation carriers, but later learned that they were, experienced a growing excess level of distress compared with persons who initially suspected that they were at risk. Women (carriers and noncarriers alike) who have had a mastectomy prior to testing experienced rising levels of distress over time. Mutation carrier men with no mutation carrier sisters had higher levels of distress compared with men with no carrier sisters, a difference that persists over time.

These results suggest that distinct subgroups of tested family members may experience elevated distress associated with being a mutation carrier or with the testing experience.

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Cognitive Appraisal, Coping Efforts, and Distress Following Genetic Testing for BRCA1/2 Mutations

Chanita Hughes, PhD, Marc Schwartz, PhD, Lari Wenzel, PhD, Caryn Lerman, PhD

Genetic testing for BRCA1/2 mutations is increasingly being integrated into the clinical management of high-risk individuals. Although the psychosocial impact of genetic testing for BRCA1/2 mutations has received significant attention, there is limited research on the mediational effects of cognitive appraisals and coping efforts. Therefore, the purpose of this longitudinal study was to evaluate the impact of BRCA1/2 test results on genetic testing distress and to determine whether this relationship is mediated by cognitive appraisals and coping efforts. We predicted that receiving positive test results would lead to higher levels of distress immediately following results disclosure and that this effect would be mediated by greater perceptions of stress, decreased perceptions of confidence, and greater utilization of problem-solving coping strategies. Participants were 148 adult women and men, each of whom was the first family member to have genetic testing for BRCA1/2 mutations (probands, n=92) or a relative (n=56). They completed measures of cognitive appraisals, coping efforts, and distress 1 month following test result disclosure. Multiple linear regression was conducted to evaluate the mediational effects of cognitive appraisals and coping on the association between test results and distress. Among relatives, higher primary appraisals of stress and increased utilization of problem-solving coping efforts mediated the relationship between BRCA1/2 test results and distress. Specifically, receiving positive test results had a significant independent association with distress (beta=.42; p=.001); however, when controlling for the effects of primary appraisal and problem-solving coping, the association between BRCA1/2 test results and distress was no longer significant (beta=.13; p=.22). Although the association between BRCA1/2 test results and distress was sizably reduced when primary appraisal and coping efforts were controlled, the effect of test results on distress was marginally significant (beta=.14; p=.10) among probands. The results of this study suggest that the impact of receiving positive BRCA1/2 test results on psychological functioning immediately following results disclosure may at least be attributed to perceptions of stress and coping efforts. Additional research is needed to evaluate whether interventions designed to decrease perceptions of stress and facilitate effective coping improve psychosocial functioning among BRCA1/2 mutation carriers.

Genetic Testing for a BRCA1 Mutation in a Large Kindred: Screening Behavior and Prophylactic Surgery in Women Following Genetic Testing

Jeffrey R. Botkin, MD, MPH, Ken R. Smith, PhD, Robert T. Croyle, PhD, Bonnie J. Baty, MS, Jean E. Nash, Debra Dutson, Anna Chan, PhD, Heidi A. Hamann, MS, Caryn Lerman, PhD, Jamie McDonald, MS, Vickie Venne, MS, John H. Ward, MD, David E. Goldgar, PhD, and Elaine Lyon, PhD

Mutations in the BRCA1 gene are associated with an increased risk of breast and ovarian cancer in female carriers. We conducted a prospective study of 189 women known to be at risk for a BRCA1 mutation in an extended kindred. Participants were offered BRCA1 mutation testing, and surveillance behaviors were assessed at baseline, 1-2 weeks, 4-6 months, and 1 and 2 years after the provision of test results. Mutation carriers, noncarriers, and individuals of unknown mutation status were compared to determine the impact of test results. Utilization of genetic testing for both men and women are reported and, for women, mammography, breast self-examination, clinical breast examination, mastectomy, oophorectomy, transvaginal ultrasound, and CA125 screening were assessed.

Results: Of those fully informed of the opportunity for testing, 55 percent of the women pursued genetic testing and 52 percent of the men. With respect to mammography, 63.4 percent (26/41) of mutation carriers ages 25 and older and 53.5 percent (54/101) of noncarriers ages 25 and older had a mammogram after receiving their test results (p=.35). These rates of mammography represent a significant increase over baseline rates for both mutation carriers (p<.01) and noncarriers (p<.05). At 1 year, 80 percent of the carrier women and 74 percent of the noncarriers reported adherence to recommendations for breast self-examination and greater than 80 percent of both groups had obtained a clinical breast examination following testing. None of the carrier women had obtained a mastectomy by 1 year after testing, although 17.1 percent were considering this procedure. In contrast, of women ages 25 years and older who had at least one intact ovary at the time of testing, 43.3 percent (13/30) of carriers had obtained an oophorectomy 1 year after testing, including 69.3 percent of women ages 40 years and older. The majority of carrier women (72 percent) had discussed their genetic test results with a medical doctor or health care provider.

Conclusions: Our results indicate a relatively high level of utilization of genetic testing. Both carriers and noncarriers demonstrate increased utilization of mammography, breast self-examination, and clinical breast examination following testing. Oophorectomy was obtained by a large proportion of carrier women in contrast with mastectomy, which was not utilized by mutation carriers within the first year following testing. These results have important implications for the relative value of genetic testing for cancer susceptibility in high-risk individuals.

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Attitudes About Hereditary Breast Cancer Among Diverse Breast Cancer Survivors

W.F. Cohn, G. Fraser, S.M. Jones, S. Miesfeldt

Introduction: Much is unknown about the breast cancer genetic counseling and risk assessment process. Until the factors involved in a woman's understanding of the cause and course of hereditary breast cancer (HBC) are known, the clinical utility of major basic scientific advances in this area will not be fully realized. Purpose: To examine the attitudes and beliefs of genetically endangered women from diverse backgrounds concerning the etiology, course and management of HBC.

Methods: All women with breast cancer diagnosed younger than age 50 years and entered in the Virginia Cancer Registry (VCR) in 1995 were eligible for inclusion. Thirty-one hospitals, statewide, entering 370 eligible patients in the VCR, participated in this project. Potential participants received a family history questionnaire and an informed consent document. Responses were received from 132 women: 45 were determined to be at risk for HBC based on their personal and family histories. Eighteen women completed an in-depth qualitative interview, based on an interview guide, that addressed attitudes and beliefs related to the cause and course of HBC. All interviews were audiotaped and transcribed.

Results: Participants included 14 European-Americans, 3 African-Americans, and 1 Native-American from throughout the State. Interviews were content-analyzed by two investigators using an emergent category system. Analysis of the interviews revealed 13 major emergent categories relating to the attitudes, knowledge, and beliefs of study participants regarding the cause and course of HBC. Included in these were factors related to causation, including impact of family history and multifactorial causation; disease risk expectation for self/family; risk reduction strategies; impact of genetic susceptibility on risk management and cancer treatment decisions; information sources, needs, and comprehensibility; and genetic blame. These data provide those directly involved with the care of women at risk for HBC a better understanding of the fears, myths, and beliefs that surround this disease.

Cancer Worry, Distress, and Health Behaviors Among Members of Li-Fraumeni Syndrome Families Undergoing P53 Genetic Testing

Andrea Farkas Patenaude, PhD, Katherine A. Schneider, MPH, Lisa DiGianni, PhD, Frederick P. Li, MD, Judy E. Garber, MD

Background: Li-Fraumeni syndrome (LFS) is a model of genetic cancer predisposition. It is one of the first conditions for which genetic testing made possible identification of those at elevated cancer risk (90 percent lifetime risk).

Methods: Our study of coping and health behavior change among 32 LFS family members (from 9 extended kindreds) undergoing genetic testing investigated the relationship between the result of p53 testing and cancer worry, distress, and adherence to recommendations for health monitoring and health behaviors. Extensive genetic counseling, repeated psychological interviewing and measurement with standard psychological instruments, and meetings with an oncologist occurred over a 12-month period. All families had an identified p53 mutation.

Results: At 1 year post-disclosure, there was no significant difference between carriers of p53 mutations and noncarriers in worry about getting cancer themselves, frequency of worrying about cancer, or in level of worry about their children getting cancer. Risk perception between carriers and noncarriers showed considerable overlap. Initial distress was high; depression in the clinical range on the BSI was seen in 23 percent of subjects at baseline. Even after excluding two subjects for extreme psychological distress, 17 percent of baseline scores were at clinical levels of depression. Distress declined for carriers and some noncarriers following disclosure and only rarely increased into the clinical range for carriers. However, clinical levels of depression remained in 7 percent of subjects 12 months after disclosure, and one subject regretted participating in testing. Among carriers of p53 mutations, reduction of tobacco use, mammogram utilization, and ability to identify a primary care physician improved only slightly in the year following result disclosure.

Conclusions: Results point to a tension between the medical and psychological goals of cancer genetic testing and suggest the complexity of response to knowledge of genetic risk status. Lessons learned from this early quantitative and qualitative study lead to recommendations for future research in cancer genetic testing. These include the need for better understanding of the impact of defensive coping mechanisms on adoption of surveillance and health behavior recommendations among high-risk individuals undergoing genetic testing.

Intentions and Decisions Regarding the Option of Genetic Testing for HNPCC: A Preliminary Look

Don Hadley, Jean Jenkins, Dave Liewehr, Eileen Dimond, Ilan Kirsch

The application of genetic testing to refine cancer risk is most often discussed in the context of families with inherited breast and ovarian cancers. In this ongoing study, we seek to understand factors that affect decisions regarding genetic testing in

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families with hereditary non-polyposis colorectal cancer (HNPCC) and the behavioral correlates of these decisions. Baseline responses to completed questionnaires by the first 104 participants focusing on intentions, thoughts, and decisions regarding genetic testing for HNPCC will be presented.

Consenting adults with colon cancer who have a tumor demonstrating microsatellite instability (MSI+) and a positive family history of colon cancer suggestive of HNPCC complete a baseline questionnaire (ELSI CSC Psychometric Tools Core). Knowledge, expectations, intentions, mood, attitudes, perceived risk, cancer worries, family relationships, spirituality, coping, and health beliefs are assessed through the questionnaire. Participants are then provided with a structured genetics education session. Following the education session, participants are provided counseling to consider personally the implications of HNPCC gene testing. They are then presented with the choice of whether to undergo gene testing. Psychological and behavioral outcomes are reassessed through telephone contact at 6 and 12 months following risk notification or the decision not to undergo testing. Notification of test results occurs in person along with a review of surveillance options. Follow-up counseling/support are provided. First-degree adult relatives of probands with identified HNPCC germ-line mutations are offered participation.

At baseline, 64 percent of participants had heard Almost Nothing or Relatively Little about genetic testing for colon cancer. Despite this relative lack of information, a majority of participants stated intentions to pursue genetic testing prior to education and counseling (69 percent stated they would definitely and 28 percent stated they would probably pursue genetic testing for HNPCC). The majority of participants (51 percent) identified learning about their children's risks of developing cancer as the most important reason to consider genetic testing. Thirty-nine percent identified concerns regarding the potential impact on their insurance coverage as the most important reason not to undergo genetic testing. Further descriptive and statistical data will be presented pertaining to intentions and decisions regarding genetic testing.

Psychosocial Characteristics of Colorectal Cancer Patients at Different Medical Risk of Colorectal Cancer

S.W. Vernon, S.K. Peterson, E.R. Gritz, C.A. Perz, B.G. Watts, S.K. Marani, W.F. Baile, C.I. Amos, P.M. Lynch

Background: Little is known about the psychosocial functioning of colorectal cancer (CRC) patients or whether functioning differs for patients at different medical risk for CRC. We used data from a prospective observational study of CRC patients, who were invited to undergo genetic counseling and testing, to examine the association between objective medical risk and demographic, medical history, and psychosocial variables.

Methods: CRC patients (n=288) completed a baseline interview before receiving genetic test results. Patients were classified as high risk if they met the Amsterdam or Bethesda criteria for hereditary nonpolyposis colon cancer (HNPCC) or as average risk if their cancer history did not meet those criteria.

Results: The two groups did not differ on sex, race/ethnicity, or disease stage. Compared with patients at average medical risk, patients at high risk had higher mean scores on the CES-D Scale (11.8 and 9.7) and on the State-Trait Anxiety Inventory (33.7 and 30.3 on the State subscale; 34.7 and 30.7 on the Trait subscale). Patients at high risk had lower scores on all Ferrans and Power Quality of Life Index subscales and on the Sarason Social Support Questionnaire (SSQ) satisfaction subscale (5.6 and 5.8). There were no differences between the two groups for the SSQ number subscale, the Miller Behavioral Style Scale, and the pros and cons of genetic testing or of informing relatives. Only one of seven items measuring attitudes about HNPCC genetic testing showed group differences; high-risk patients were more likely than average risk patients to agree that learning genetic test results would help them or their family. High-risk patients were not more likely to worry that they carried an altered gene for HNPCC or to feel less able to cope with genetic test results.

Conclusions: Patients at high medical risk of CRC may benefit from counseling that addresses psychological and quality of life issues. Psychological status may affect patients' ability to communicate or interest in communicating with their relatives about their increased risk. Because relatives of high-risk patients could potentially benefit from genetic counseling and testing, it would be useful to know whether psychological status is associated with communicating risk information to relatives.

Impact of Genetic Counseling on Persons at Increased Risk for Colorectal Cancer

Tracy L. Waldeck, Ann-Marie Codori, Jill D. Brensinger, Candace Young, Marijayne T. Bushey, Judith Bacon

One goal of genetic counseling is to modify erroneous perceptions of disease risk, improve disease-related knowledge, and allay excessive fear. This study examines the effect of genetic counseling on risk perception, knowledge, and fears/worries among 101 healthy adults at increased risk for hereditary nonpolyposis colorectal cancer (HNPCC). The subjects were participants in Phase I of a study offering genetic testing for HNPCC. Each participant had one or more first-degree relatives affected with colorectal cancer (CRC) and at least one relative with early onset (<50). Immediately before genetic counseling, participants rated their CRC risk perception with a visual analogue scale, demonstrated HNPCC knowledge with a six-item true/false scale

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and several questions related to CRC prevention, and rated their CRC fear, worry, and stress on 5-point Likert scales. Predictor variables included demographic characteristics, perceived health status, and perceived likelihood of having a gene mutation for HNPCC. At various points after genetic counseling, but before blood draw for genetic testing, participants repeated the baseline measures. The length of the interval, between baseline and the second assessment, was employed as a modifier variable. Preliminary analyses showed that baseline risk perception was significantly associated with age ($p<.0001$; older persons reporting lowered perceived risk), perceived health status ($p=.037$; persons reporting relatively good health reporting lower risk perception), and perception of oneself as a gene mutation carrier ($p<.0001$; persons who strongly believe they have a gene mutation for CRC reporting greater perceived risk). Analyses are currently under way to determine whether and how genetic counseling changes risk perception, CRC knowledge, and CRC-related fears and worries. Additional analyses evaluate the moderating effect of individual coping styles on the outcome variables. We hypothesize that genetic counseling will create more realistic perceived risk (i.e., risk perception will move closer to the counseled risk), increased knowledge about HNPCC, including relevant screening behaviors, and decreased fears and worries about CRC. In addition, we hypothesize that the effect of genetic counseling on the outcome variables will be moderated by coping style, with active copers (i.e., information seekers) showing greater benefits from genetic counseling than denial copers (i.e., problem ignorers).

Psychosocial Impact of HNPCC Genetic Counseling and Testing

E.R. Gritz, S.K. Peterson, S.W. Vernon, S.K. Marani, B.W. Watts, P.A. Ward, W.K. Kohlmann, M.L. Frazier, C.I. Amos, P.M. Lynch

We are conducting a longitudinal descriptive study of behavioral and psychosocial aspects of genetic counseling and testing for HNPCC, in which genetic counseling and testing are first offered to colorectal cancer patients (CRC) and then to first-degree relatives (FDRs) of patients who are carriers of an HNPCC-predisposing mutation. We have recruited patients from clinics at the University of Texas M.D. Anderson Cancer Center, including a consecutive series of patients and those identified as having a possible HNPCC family history. Two hundred and eighty-nine CRC patients who provided a blood sample for genetic testing and 95 FDRs of mutation-positive patients will be profiled. All individuals completed psychosocial questionnaires before undergoing genetic testing. Measures include demographics, psychological distress, coping style, quality of life, social support, knowledge, and attitudes toward testing and CRC screening (among FDRs). To date, genetic test results for 72 CRC patients have been disclosed. HNPCC-predisposing mutations have been found in 33 distinct families. We have offered genetic counseling and testing to 137 FDRs from those families: 93 completed a psychosocial questionnaire; 78 underwent genetic counseling; 73 completed genetic testing; and 69 received genetic test results. This report will describe the psychological impact of HNPCC genetic counseling and testing on 41 CRC patients and 51 FDRs within 2 weeks and at 6 months after receiving their test results.

Section 8: Education and Outreach

Washington University Science Outreach Project

Sarah Elgin, David Kirk, Victoria May, Cynthia Moore

The Washington University Science Outreach curriculum project Modern Genetics for All Students was developed to incorporate basic DNA science within a traditional high school genetics unit, including the social and ethical issues surrounding modern genetics. The project, begun in 1991, features curriculum enhancement materials that combine strong scientific content with human health applications, providing a high level of hands-on involvement for students using wet lab experiments, modeling activities, and information-gathering and analysis. With funding from the National Institutes of Health and the Howard Hughes Medical Institute, this long-term project has succeeded through a strong partnership among scientists and implementation specialists at Washington University, project evaluators, and biology teachers at local high schools. The curriculum unit has been extensively tested and evaluated in more than 200 biology, life science, and applied biology/chemistry classrooms over a 9-year period. Statistical evaluation of both student knowledge gains and improvements in student attitudes toward science and health shows the curriculum to be effective with all student levels, including females and underrepresented minorities. Our field-testing model has identified three factors that are essential for the success of hands-on intensive science in the classroom: teacher content training, provision of materials in classroom-ready form, and strong implementation support during the first year of new curriculum use. Teachers at local high schools indicate that this model has enabled them not only to improve their current teaching strategies, including a higher level of hands-on involvement for students, but also to establish an internal support network to sustain these changes with minimal Washington University-based core support.

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Teaching the Teaching of ELSI Issues: A Progress Report

David J. Bzik, Ronald M. Green, Albert Scherr

Since 1996, Dartmouth College's Ethics Institute has brought three classes of college and university teachers through a Faculty Summer Institute designed to assist them in the preparation and teaching of undergraduate courses on the ethical, legal, and social implications of the Human Genome Project. With two future Institute offerings ahead, this effort remains a work on progress. Here, we describe some of the leading insights we have gained in developing this curriculum project. Because ELSI issues are continuously evolving, we place special emphasis on our outreach, continuing education and dissemination activities, and efforts to enable Institute graduates to serve themselves as teachers of teachers. Drawing on the experiences of the Dartmouth College faculty team and the subsequent field reports submitted by Institute graduates, we also discuss the skills and preparation needed by teachers to assist students to understand and respond effectively to ELSI issues. We conclude that ELSI issues provide an exemplary opportunity for multidisciplinary education. Teaching ELSI issues requires a willingness to comprehend the approaches of other disciplines. It also asks teachers to convey the insights and methods of their own discipline in ways that can be understood and appreciated by nonspecialists. Students with both science and nonscience backgrounds respond enthusiastically when faculty are able to achieve this level of cross-disciplinary conversation.

The Gap Between Practice and Genetics Education of Health Professionals: HuGEM Survey Results

E. Virginia Lapham, PhD, Chahira Kozma, MD, Joan O. Weiss, MSW, Judith L. Benkendorf, MS, CGC, Mary Ann Wilson, BA

A 1998 random sample survey of 3,600 health professionals confirmed the finding of a 1995 exploratory study of 329 health professionals that a variety of health disciplines are already being asked to provide genetic services. Both studies were carried out as part of the Human Genome Education Model (HuGEM) Project of Georgetown University and the Genetic Alliance and funded by the NIH, NHGRI, ELSI Program. The health professionals in the 1995 study practiced in 52 of 71 University Affiliated Programs associated with medical centers around the country. The collaborating associations in the 1998 study included (ADA) dietitians, (AOTA) occupational therapists, (APTA) physical therapists, (APA) psychologists, (ASHA) speech-language-hearing specialists, and (NASW) social workers. Of the 1,958 responses received in the 1998 study, 70 percent have discussed the genetic component of problems with at least a few clients, 19 percent made referrals for genetic counseling, 15 percent referred for genetic testing, and 30 percent provided counseling about genetic concerns. There is a considerable gap between providing genetics services and confidence of health professionals. While 67 percent take family histories, only 1 in 4 is confident about eliciting genetic information as part of the history. Most social workers (87 percent) and psychologists (79 percent) provide counseling; however, only 26 percent and 29 percent, respectively, have high confidence in counseling clients making decisions about genetic testing. Although two-thirds of respondents hold graduate degrees, less than 21 percent have had one or more courses in genetics, 44 percent have had genetic content in course work, and 33 percent have had no formal education in genetics. High confidence is related to the amount of genetics education, independent of overall education. For example, 98 percent of psychologists have doctorates; however, their confidence in providing genetic services is correlated with the amount of genetics education on all eight measures ($p<.05$). Nearly two-thirds of respondents would like to have continuing education in genetics. Priority topics identified were (1) role of genetics in common disorders, (2) overview of human genetics, (3) identifying genetic resources, (4) helping clients cope with new genetic diagnoses, and (5) genetic information and racial/ethnic concerns. The survey supports the need for incorporating genetics into the practice and teaching of health professionals as essential for these professionals to confidently meet the needs of their clients. It will take a coordinated effort among genetics professionals, professional associations, academic institutions, and funding agencies to ensure that health professionals receive the genetics education needed early in the 21st century.

Partnership for Genetic Services Pilot Program: Health Care Provider Education in Genetics in Managed Care Organizations

Vicky Whittemore, Nancy Hanson, Ute Ochs, Jacob Reiss, Jessica Kushner, Nisha Isaac, Bowie Little, Nachama Wilker, Ann Smith, Cindy Holmes, Mary Davidson

The focus of professional education efforts by the Genetic Alliance has always been on providing health care providers with the information, resources, and tools they need to deliver quality genetic services. The involvement of consumer educators (individuals with genetic conditions and/or their families) in the education process highlights the ethical, legal, and social issues in genetics. The Partnership in Genetic Services Pilot Program has worked directly with providers-in-practice in two managed care organizations (MCOs) to foster family-centered, culturally competent, and consumer-informed service delivery to individuals and families with genetic conditions.

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A needs assessment was performed by interviewing MCO plan members, medical administrators, and plan providers (family practice, neurologists, and pediatricians). Baseline information about genetic services delivery in the two MCO pilot sites was obtained. These studies produced an understanding of (1) how and where providers access information and support resources for their patients, (2) what types of resources they accessed, (3) how consumers access and experience services, and (4) which services were most/least helpful to the consumers.

The results of the assessment provided guidance on provider preferences about learning tools, methods, and the time they had available for educational interventions. These measures shaped the interventions and produced a valuable snapshot of genetic services delivery from inside a managed care system.

Case-based workshops were developed and presented to both of the MCO pilot sites in the form of departmental meetings, 1 hour, half day, or full day CME sessions, and online computer resources. These workshops combined the clinical expertise of MCO genetics professionals with consumer expertise in presenting the ethical, legal, social, and family aspects and introducing high-quality resource materials. The insights, lessons learned, and results of the baseline delivery assessment, interventions, and evaluations will be presented.

This project was funded by the National Human Genome Research Institute - Ethical, Legal, and Social Implications Research Program and the Genetic Services Branch of the Maternal and Child Health Bureau (Title V, Social Security Act, Health Resources and Services Administration).

Getting the Word Out on the Human Genome Project: A Multimedia Course for Physicians

Sara L. Tobin, Ann Boughton, L. Caron, B. Koenig

The rapid progress of the Human Genome Project and the exponential identification of genes that are correlated with genetic conditions have the potential to change the face of medical care. An increasing number of persons will be faced with the prospect of making informed personal choices based on this emerging technology. Because the field has emerged so recently, most practicing physicians have not been trained in molecular genetic testing and its implications for patients and their families. We are addressing this discontinuity with the development of an interactive, multimedia CD-ROM-based course for the continuing education of physicians. This courseware is entitled The New Genetics: Courseware for Physicians. Molecular Concepts, Applications, and Ramifications. The courseware will provide training in four areas: (1) genetics, including DNA as a molecular blueprint and patterns of inheritance, (2) recombinant techniques, stressing cloning and analytical tools and techniques applied to medical case studies, (3) current and future clinical applications, encompassing disease diagnosis and prognosis, as well as technical advances, and (4) societal implications, focusing on issues such as privacy and impact on the family.

This choice of educational technology confers the ability to incorporate a wide variety of media approaches. The courseware is designed for ease of navigation and will be capable of seamless retrieval of supplements and updates from the Internet. In the first stage of this project, a prototype was developed and evaluated by physicians in isolated practice environments to validate our approach. This courseware may also be useful for undergraduates, medical students, and the general public, especially in distance learning situations. We believe that multimedia education will be practical and effective because interactive technology encourages involvement, and the use of varied media enhances user understanding of complex concepts. Physicians will be able to obtain continuing medical education credits through Stanford University. The courseware is designed to provide a powerful tool for the education of physicians and the public about the potential of the Human Genome Project to benefit human health. The courseware will be demonstrated at the poster session.

This project is supported by grant number DE-FG_ER67128 from the Office of Biological and Environmental Research, U.S. Department of Energy.

Genetic Education for Native Americans: An Update and Preliminary Evaluation Data

L. Burhansstipanov, L. Bemis, M.B. Dignan, C. Poodry, F. Romero

Genetic Education for Native Americans (GENA) is being implemented by Native American Cancer Research (an Indian owned and operated nonprofit organization) and is funded by the Ethical, Legal and Social Implications (ELSI) of the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH). The primary goal of this 3-year study is to provide culturally competent education about genetic research and genetic testing to Native American college and university students. The curriculum is implemented as customized modules within science-based conferences that have a significant number of Native American college and university students or as a 16-hour comprehensive preconference workshop. This presentation will provide a brief update of the project, with special emphasis on the preliminary evaluation data. The workshop provides a balanced approach to the scientific and cultural information and provides recommendations for ways

to improve the cultural competence of contemporary genetic research, education, and careers. Participants who complete the workshop are eligible for individualized mentor-supervised internship opportunities in diverse regions of the United States.

Information Conferences: The Human Genome Project — The Challenges and Impact of Human Genome Research for Minority Communities

Issie L. Jenkins, Esq., Dr. Rosalind P. Hale, Dr. Kathryn T. Malvern

The Zeta Phi Beta Sorority National Educational Foundation planned and presented two conferences targeted to minorities, one in New Orleans, Louisiana, April 16-17, 1999, and one in Philadelphia, Pennsylvania, July 7-8, 2000, funded with the assistance of government grants. The conferences reached 400 members of the minority communities. Local community involvement was promoted with the assistance of advisory committees comprising representatives from all segments of the communities involved.

Conference objectives included disseminating information to African-Americans, Asian-Americans, Native-Americans, Hispanics, and others about the Human Genome Project and related genetic research; encouraging greater inquiry and involvement of minorities; and providing an appreciation of the ethical, legal, and social issues involved as well as a forum for discussion of community concerns.

Conference presenters and workshop leaders included research scientists, anthropologists, government health officials, social scientists, educators, religious leaders, attorneys, and representatives of the medical profession and the criminal justice system.

The Foundation proposes that its presentation and/or poster emphasize what it has learned about the state of information in the minority communities on the Human Genome Project and the status of genetic research; how best to get the information out to these communities; what issues concern the minority communities; and the recommendations of the minority communities for promoting greater minority involvement. The presentation and/or poster will be based on the Foundation's experience in planning and implementing the two conferences referred to above, on the recommendations that these conferences generated, and on a statistical analysis of the conference evaluations.

The Einstein Institute for Science, Health & the Courts

Franklin Zweig

The Einstein Institute for Science, Health & the Courts (EINSHAC) is the DOE and NIEHS grantee for the Genetics Adjudication Resource Project (GARP), an ELSI-genetics education and publications effort affiliated with the Judicial Branch of Government. By the January 2001 date of the ELSI, Decade of Research conference, the GARP will have provided education in molecular biology, biotechnology and genetics to 2001 Federal and State court judges in the United States.

Our calling is to make science accessible to the instruments of justice. Our mission is to provide judges, courts, and court-related personnel with knowledge tools related to criminal and civil justice proceedings involving evidence from the genetic sciences — genetics, molecular biology, biotechnology, and molecular medicine — and from new discoveries and technologies in the environmental and neurosciences. In sum, we emphasize the science and impacts of life technologies in judicial system proceedings.

EINSHAC operates three divisions: (1) the Genetics Adjudication Resource Project, (2) the Law and Science Academy (LSA), and (3) Professional Education Services.

GARP's objective is to assist the Judicial Branch with respect to new case varieties emanating from the Human Genome Project's identification of all human genes. LSA's objective is to peer into the future of science and technology-based legal controversies in semiannual judges' seminars. The Professional Education Services Division will make its debut in 2001 and offer case-related life science and technology workshops, courses, seminars, and laboratories to professional and graduate schools and continuing education entities.

We apply educational technologies in three genetics-related conference series: (1) genetics orientation (the ELSI-genetics basic boot camp series), (2) specialized subject matters (the advanced series), and (3) the leadership development series. Each series strives to promote case management and appellate review.

Public Education Project to Foster Awareness of Science and Policy Issues in Environmental Genetics and Genetic Testing

Susan Vandale, Eula Bingham, LaVerne Mayfield, Gregory Oakley

In 1998, the Center for Environmental Genetics, Department of Environmental Health, University of Cincinnati (DEH-UC),

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began a science education project known as LEGENDS (Learning Exchange for Genetic and Environmental Disease Solutions). LEGENDS fosters public awareness regarding environmental genetics and genetic testing, with a focus on the ethical, legal, and social implications (ELSI) of advances in these areas.

The project has developed a modular curriculum, highlighting topics on environmental health, genetics and cell biology, human genetics, environmental genetics, genetic testing, and human genome research. DEH-UC faculty and staff, other scientists, and a community partner (Greater Cincinnati Occupational Health Center) collaborate to present the curriculum.

The factual knowledge in the curriculum is organized into 24 thematic modules, that form the outline for lesson plans. Project-sponsored workshops feature brief, instructor-led presentations to communicate scientific concepts and project-developed exercises, such as games, video skits, and role-plays, to reinforce the science and evoke discussion of ELSI. Before each workshop, a planning meeting is held with representatives of groups to choose topics and adapt the lesson plan to meet group requirements.

To date, 185 individuals have participated in the workshops, each ranging from 2 to 4 hours in length and covering two to five modules. Approximately 225 more people have attended short focus group discussions on curriculum-related themes. Attendees at these sessions included worker health and safety trainers, environmental advocates, labor representatives, science teachers, health department sanitation inspectors, and other concerned citizens, many of whom represent minority and underserved populations as well as health and education professionals interested in community outreach.

Workshop evaluation currently shows the following outcomes: participants indicating personal utility of session, 92 percent; participants rating workshop good or excellent, 80 percent; conceptual knowledge gain, measured as simple percentage change (post-test mean minus pre-test mean divided by pre-test mean), 11 percent, and as relative percentage change (post-test mean minus pre-test mean divided by top score minus pre-test mean), 19 percent.

Evaluation data suggest that the LEGENDS curriculum is a useful resource for developing public awareness on science and ELSI-related topics in environmental genetics and genetic testing.

A Statewide Project to Educate and Raise Awareness About "The New Genetics": The CGEP Project in Vermont

David W. Yandell, Rachel Wallace-Brodeur, Karen Richardson-Nassif, Leah Burke, Edward Mahoney

The Community Genetics and Ethics Project (CGEP) is an NHGRI-ELSI-funded educational project targeted broadly at all Vermont residents. The statewide project has partnered with community organizations and existing State networks to facilitate access to the broadest possible audience including professional, lay, and special-interest groups. CGEP is organized in a "concentric circles" model, with initial contact occurring in intensive 2- to 4-day retreats. Retreat participation is voluntary. This self-selected and motivated group of retreat participants is next called upon to facilitate interaction with broader audiences via local, community-based discussion groups and public "town meeting" style forums.

To date, CGEP has coordinated 16 multi-day retreats for groups including clergy, mental health professionals, primary care health care providers including M.D., NP, and midwives; social service workers; public policymakers (elected and appointed officials, lobbyists, and SIGs); book discussion group leaders; students in health care professions (M.D., nursing, allied health, psychiatry); hospital institutional review boards and ethics committee members; families affected by genetic disease; the Deaf Community; high school science teachers; and others. Through partnership with Vermont Center for the Book, four-part book discussion groups were held in 26 locations around the State. The CGEP is now actively organizing town meeting style evening forums in accessible and geographically diverse sites.

Retreats, discussion groups, and community forums focus on the interests or needs of the specific audience using a mixture of lecture, panel discussion, small-group discussion, and case-study formats. Prepared educational materials, slide presentations, selected readings, and case studies are provided in advance. A survey instrument is used pre- and post-retreat to assess changes in knowledge and attitudes; post hoc focus groups and "reunion" events have been used to assess long-term impact on the participants' knowledge, attitudes, and behavior imparted by their CGEP experience. A Web site, newsletter, and listserver are used to maintain contact with participants and facilitate their continued interaction with the project.

To date, significant changes in knowledge and attitude questions have been seen. The target audiences have a high level of interest but divergent viewpoints and varying levels of concern related to new genetic technologies and ELSI issues. Misconceptions and misinformation are abundant in the rural population we serve. Access and "mode of entry" into specific subgroups are a major challenge, but our experience suggests that targeting preexisting well-organized networks or community organizations will be most successful. Barriers to raising awareness and educating the general population about ELSI issues and "the new genetics" can be overcome by sustained and carefully coordinated educational interventions. Broad-based efforts will require more accessible, up-to-date information resources that are relevant to — and at an appropriate educational level for — the specific audiences being targeted.

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Section 9: Research Issues

Using Vignettes to Improve Informed Consent: A Study of Consent for DNA Banking

Jon F. Merz, Pamela Sankar, Emma A. Meagher, Timothy R. Rebbeck

This research focuses on the applied development of an informed consent form and process for donation of blood to a DNA bank solely for research purposes. A thoroughly piloted consent form was developed and run in a mock setting with research subjects in the General Clinical Research Center and patient subjects in the apheresis unit at the Hospital of the University of Pennsylvania. Subjects who agreed to read and respond to the consent form were randomly assigned to receive the consent form with or without a short series of vignettes. The vignettes, presented as a Part 2 of the consent form, are meant to spur conversation and to prompt cognitive processing of the information in the consent form, which we hypothesized would increase understanding and recall. The vignettes also play the role of a minor test of understanding of the consent information, with the research nurse reviewing and discussing any problems identified in the vignettes in the informed consent meeting. After completing the informed consent discussion, we asked individuals to participate in a brief survey about the consent process. Preliminary results (n>180) showed that 19 percent of people solicited stated they would refuse to donate to a DNA bank, 25 percent stated they would donate only in anonymous form, and 56 percent would donate in fully identified form. Multinomial logistic regression showed that subjects who perceived greater risks from donation were less likely to donate and, if willing, more likely to donate in anonymous form. The vignettes had no significant effects on consent form choices, on perceived risks and benefits of the research, or on recall of consent information. Data collection is now closed. Final results will be presented and discussed.

This research was funded by the NIH, DOE, and VA Consortium on Informed Consent Research under RO1-HG-01765.

Informed Consent for Genetic Research in Epidemiological Studies

Lisa S. Lehmann, MD, MSc, Elizabeth Hohmann, MD

Background: Large longitudinal epidemiological studies have banked thousands of patient blood specimens. These specimens were collected many years before the genetic revolution for research purposes, and subjects gave general consent to participate in research. Participants are surveyed on a regular basis, occasionally send specimens for analysis, and have released all of their medical records to the researchers. The desire to do genetic research on these banked and identifiable specimens has raised a series of ethical questions.

Ethical Questions:

I. Do researchers who want to pursue genetic research on banked specimens need additional consent from the research participants?
II. Can researchers do genetic research on specimens of individuals who have died?
III. Do researchers have an obligation to share the results of their research with individual research subjects?

Discussion:

I. Possible responses to the first question include (1) obtaining additional consent from participants, (2) assuming that participants gave their specimens for research purposes and genetic research is therefore included in the general consent that was given, and (3) informing research subjects of the type of research being pursued and giving subjects the opportunity to opt out of genetic research.
II. To answer the second question, we need to understand why genetic research should have specific consent and what this consent is intended to protect. The argument that investigators obtain a specific consent for genetic research is based on the assumption that the results of genetic research have the potential to harm the subject in ways that are different from nongenetic research. Specifically, there is concern about the possibility of insurance and employment discrimination that may result from an awareness of the presence of a genetic alteration. Since research subjects have died, there is no possibility of discrimination to them based on the results of genetic research. There is, however, the possibility that family members could suffer discrimination.
III. The obligations of researchers to share results with participants may depend on (1) the accuracy of the research results, (2) the significance of the information discovered through the research, (3) the ability to provide appropriate counseling about the implications of the research, and (4) the agreement reached between the researcher and the subject before the onset of the research.

Proposed Solutions:

I. There are significant logistical obstacles to re-consenting thousands of research subjects for genetic research.

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Some genetic studies may warrant waivers of specific informed consent. It is possible, however, that some individuals may not want their specimens used for this type of research. We therefore suggest that investigators inform research participants that genetic research is being done and give participants the opportunity to withdraw their participation.

II. No harm will ensue to the deceased individual whose specimen undergoes a genetic analysis. Furthermore, the likelihood of harm to family members of deceased individuals is very small. We therefore argue that it is permissible to use the specimens of deceased individuals in genetic research.

III. Most research studies are not carried out in CLIA-approved laboratory facilities; for this and other reasons, the accuracy of research results is not certain. Researchers may, however, discover information that has significant clinical utility, which, if acted upon, may lead to the prevention of serious illness. Under such circumstances, investigators have an obligation to inform the group as a whole of the research results and allow individuals the opportunity to have tests repeated in a clinical laboratory with appropriate genetic counseling.

Informed Consent in International Genetic Epidemiological Research

Patricia Marshall, PhD, Charles Rotimi, PhD

In recent years concerns about the application of ethical guidelines governing human subjects research in multinational collaborations have increased. Attention has focused on a range of issues, including problems associated with adhering to Western standards for obtaining voluntary informed consent in non-Western countries. This presentation describes a multinational investigation of informed consent practices in genetic epidemiological investigations of hypertension and breast cancer. Study sites include urban and rural settings in Nigeria and the metropolitan area of Chicago in the United States. Quantitative and qualitative methods are used to examine practices for obtaining informed consent to genetic research in these diverse social settings. This study will document ethical challenges to voluntary informed consent and identify mechanisms for ensuring culturally relevant approaches for consent to genetic epidemiological research. The study design includes a case control survey of research participants in the hypertension and breast cancer studies. The survey addresses individuals' motivation to participate in the research and their understanding of the consent discussion. Direct observations of the consent discussion and in-depth interviews will be conducted with a subsample of the survey participants. Focus groups will be conducted with research teams at each site to explore ethical issues surrounding informed consent practices. In this presentation, findings are reported from preliminary interviews with Nigerian investigators. Interviews focused on the challenges faced in meeting ethical requirements for voluntary informed consent. Special consideration will be given to the social and cultural factors that influence consent discussions. Processes of obtaining permission to conduct a study from community and tribal elders will be described, and their implications for expressions of individual autonomy and social agency will be examined. The impact of cultural beliefs about disease etiology and biomedical procedures on informed consent will be explored briefly. Recommendations for enhancing cultural sensitivity in the consent process will be outlined.

A Regional Military Health Care System Response to Expanding Genetic Research: Ethical, Legal, and Social Implications

R.F. Hume, K.A. Azarow, F. Olmstead, J. Myers, B.C. Calhoun, D. McCune, L. Brosch, J. Daniels, B.A. Jones, C. Freund, L.S. Martin

Advances in biotechnology, molecular genetic diagnostic capabilities, and the potential for gene therapy incited an explosion of innovative efforts within existing health care systems. Contemporaneously, the military health care system embraced a massive reengineering process adapting principles of managed care in a rapidly right-sizing environment. These forces required an interdisciplinary learning response in which legal, regulatory, and medical models were integrated with the new knowledge in genetics, information management, and best business practice methods. How do you perform a best business case analysis to provide decision-makers with intelligible choices that can be translated into viable business plans for expanding genetic services? This required Institutional Review Board oversight of multiple clinical investigation demonstration projects such as familial breast cancer susceptibility, prenatal screening and diagnosis, newborn screening, genetic research on existing tissue archives, and the genetics of occupational health within the military health care system.

These advances must be incorporated in the existing legal framework and regulatory procedures for the protection of research subjects from unintended consequences of unrecognized risks. Our collaborative initiative has resulted in the introduction of new policies for genetic research governance, expanded genetic services, and educational outreach. Most important has been the recognition that the genetic paradigm, rather than community health or medical models, optimizes the appropri-

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ate matching of genetic care to our community needs while respecting the multicultural diversity (molecular, social, and religious) of the military community. In all these efforts, we have enjoyed the full support of CIRO, Lead Agent, and Health Affairs.

Data-Sharing and Data-Withholding in Academic Genetics: Evidence from a National Survey of Geneticists in Academic Institutions

David Blumenthal, MD, MPP, Eric G. Campbell, PhD

Context: The importance of sharing in science is widely accepted. However, at times scientists withhold research-related information, data, and materials from their colleagues.

Purpose: To understand the extent, consequences, and trends in data-withholding in genetics.

Design: A mailed survey of a random sample of 3,000 genetics and other life science faculty with a 64 percent response rate. The analytic sample upon which this abstract is based includes only those respondents who identified themselves as a genetics researcher (50.2 percent of all respondents).

Findings: Among geneticists, 84 percent made at least one request of another academic scientist for information, data, or materials concerning published research in the last 3 years. Among those, 75 percent indicated that at least one of their requests had been denied or honored only after a significant delay. As a result of another scientist's withholding behaviors, 23 percent of respondents had a publication significantly delayed, 28 percent were unable to confirm others' published results, 21 percent abandoned a promising line of research, 28 percent stopped collaborating with another academic scientist, 13 percent refused to share with that individual or group, and 18 percent delayed sharing with that person or group. When asked about changes in the overall willingness of academic scientists in their area of research to share information, data, or materials, 35 percent felt other academic scientists are less willing to share now than 10 years ago, while only 14 percent felt other scientists are more willing to share.

Conclusions: On the basis of these preliminary results, it appears that withholding information, data, or materials concerning published research is common in academic science and is related to scientists' ability to publish in a timely manner, their professional collaborations, their data-withholding behaviors, and their ability to replicate the work of others. Further, it appears that a large percentage of academic geneticists believe their colleagues have become less willing to share research results with others.

Consent in the Field of Human Genetics: A Question of Form?

G. Cardinal, M. Deschenes, B.M. Knoppers, K. Glass

The need to obtain consent from a research participant is an internationally well-known fundamental principle of research. In practice, it can be difficult to determine which elements are realistic in order to obtain fully informed and free choices. It is an onerous task for scientists and research ethics committees to identify the various themes that should be addressed in a consent form while respecting legal and ethical standards. This challenge is even greater in the context of human genetic research.

We hypothesized that existing consent forms used in biomedical research do not adequately reflect the concerns specific to research in the field of human genetics. We undertook a study of current consent forms by collecting examples from a variety of research projects. Also, we reviewed relevant normative/policy documents from national, regional, and international perspectives. Taking into account these sources, we identified new elements pertaining to genetics that could be added to the consent form. For each selected theme, we proposed model clauses that respect the current legal and ethical norms, address the varying interests researchers may need, and correspond to the modalities of the research protocol.

We will highlight themes in the model that are specific to the field of human genetic research, such as communication of results, risks, confidentiality, secondary uses of genetic material, and commercialization. New standardized consent forms will be proposed and compared with existing clauses in biomedical research.

Informed Consent for Research on Stored Blood and Tissue Samples

Mary T. White, PhD, Jennifer Gamm

Biomedical research increasingly involves the collection of blood and tissue samples for future research, much of which is likely to include genetic analysis. Because genetic analysis may reveal information about an individual's identity, relationships to others, and predisposition to disease, genetic research on stored samples may pose unique risks for those who serve as sample sources. The aim of this study was to obtain information reflecting whether IRB practices vary in their consent form requirements for such research and some of the factors that may contribute to that variation. A brief survey was mailed to all

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IRB chairs on a mailing list obtained from the OPRR. Questions included whether six different provisions commonly recommended in existing guidelines and position statements are addressed in consent forms for protocols involving the collection, storage, and future use of biological samples. Results indicate that IRB practices do vary, with greater attention to human subjects protection corresponding to the location of the IRB, the number of protocols seen each year, whether the IRB included a member with ethical expertise in genetics, and whether the IRB used both the IRB Guidebook and the NBAC Report on Research Involving Human Biological Materials in its deliberations.

STEPPP — Stored Tissue: Evaluation of Policies, Practices, and Preferences

Susan Metosky, Sara Hull, PhD, Carol Freund, PhD, Elisa Hurley, Holly Gooding, Benjamin Wilfond, MD

Over the last 5 years, various recommendations and ethical guidelines have emerged regarding informed consent for the collection, storage, and secondary use of tissue samples in research. However, it is unclear how these recommendations have been implemented. The purpose of the Stored Tissue Consent Form Evaluation Project is to characterize and examine variations in NIH intramural consent forms as a measure of the range of strategies that investigators and IRBs are using to incorporate these recommendations and guidelines.

We reviewed 826 currently active intramural NIH consent forms that have been approved by 1 of 14 NIH IRBs. We classified these forms according to the primary and secondary purposes of the corresponding research protocol, including drug intervention, physiologic monitoring, natural history, imaging, social science, teaching, screening, tissues collected for immediate therapeutic use for the subject, tissues collected for clinical evaluation, tissues collected for general research, tissues collected for genetics research, and tissues collected for potential/confirmed future use, genetics or otherwise. Drug intervention (n=235) was the most common primary purpose.

We further evaluated the 226 studies whose primary or secondary purpose included tissues collected for genetics research or tissues collected for future use. In 123 of 226 cases, forms were included because the study's primary purpose was tissue collection for genetics research, and in 29 of 226 cases, forms were included because the study's primary purpose was tissue collection for future use. Forty-five forms had tissue collection for genetics research as a secondary purpose and 136 had tissue collection for future use as a secondary purpose. A 71-item evaluation tool including the following domains was developed: layout/formatting of consent forms, disclosing genetic test results, risks, confidentiality, ownership, storage of samples, withdrawal from study, secondary use of samples, and use of boilerplate language to discuss the various content areas. Two independent reviewers are coding the consent forms, reconciling any discrepancies before the forms are entered into SPSS for statistical analysis. Results from this study will guide the development of a survey of public attitudes and preferences concerning the collection, storage, and secondary use of biological materials for genetic research.

Genetics Research in the International Context

Ellen Wright Clayton

The Council of International Organizations of Medical Sciences is currently revising its international guidances for research involving human subjects and for the first time is explicitly addressing the particular challenges posed by genetics research. The group concluded that although this sort of research should not be treated differently from other types of research, genetics nonetheless raises some issues that merit further emphasis, including:

1. Topics that need to be disclosed in the informed consent process, including the storage of human biological materials and creation of cell cultures, and existence of conflicts of interest,
2. Greater attention to the possibility of undue family influence on research participation,
3. Further exploration of the particular ethical challenges of epidemiologic research,
4. Further exploration of disclosure of research results, with the following recommendations.
   - The first step is always to determine whether particular results are sufficiently robust to have clinical utility.
   - The generalizable results should be shared, if possible, consistent with the limitations imposed by the funder(s), with the scientific community. Investigators should advocate for general distribution.
   - Investigators who learn about health risks affecting the community under study should inform the responsible public health officials.
   - With regard to individual results, at the time informed consent is sought, investigators should indicate, based on the expected utility of particular results, whether individual subjects will be given the chance to obtain their personal results. If informed consent was not sought prior to the study, the presumption should be against disclosure. Investigators who wish to overcome this presumption must demonstrate to

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an independent review body that informing individuals of their particular results is likely to avert a serious harm.

Disclosure to family members raises particular problems that may vary depending on cultural practices and norms.

The rationale for these recommendations will be presented.

The Canavan Case: Bargaining for Benefit and the Financial Windfalls of Genetic Discovery

Jon F. Merz

The gene that causes Canavan disease was discovered by researchers at the Miami Children's Hospital (MCH) in 1993, and a patent covering the diagnostic test was issued in 1997. The research was performed with extensive participation of families stricken by the disease, which helped identify other families from around the world, helped solicit blood for the research, and provided financial support for the work. This case study is based on professional involvement of the author and interviews with numerous individuals involved in the research. It examines the research process and the subsequent restrictive licensing program put in place by MCH. The case raises difficult questions about (1) secrecy surrounding commercial interests in medicine, (2) the effects of restrictive licensing on patient access to medical services and on medical practice itself, (3) the ethical requirements for, and scope of, informed consent in genetics research, and (4) the ownership and distribution of commercial products and financial rewards resulting from research. The case highlights the need for patient and other advocacy groups to proactively assert their interests in the intellectual property resulting from research in which they participate. Further, researchers and IRBs must consider these interests in planning research, assessing the potential benefits, and formulating policies for consent. They can best do this by engaging affected groups and participants openly in dialogues about these issues, before the research is done.

Section 10: Genetic Testing — Family Issues

Access and Confidentiality in the Genetic Testing of Children: Separating the Professional from the Personal

Lainie Friedman Ross, MD, PhD

U.S. and U.K. genetic societies have published policy statements that address the ethical issues raised by genetic testing of children. Both statements argued that a child's medical interest should be paramount. In that regard, both statements do not support testing children for late-onset conditions or carrier status. The U.K. statement, however, included an appendix that described survey results of clinician practices. The survey showed that the recommendations would face resistance given the number of clinicians who were already acceding to parental requests.

In this pilot study, I have done in-depth interviews with 12 pediatricians and 10 geneticists to understand their attitudes and beliefs about genetic testing of children I have asked them what recommendations they would offer for policies on genetic testing of children. Cases included (1) early-onset conditions that were and were not treatable, (2) late-onset conditions for which treatment, if it existed, was not available in childhood, (3) carrier testing, and (4) testing for behavioral traits. Although many did not support genetic testing of children for late-onset diseases, carrier status, or behavioral traits, and would not test their own children, many were willing to accede to parental demands. Reasons varied but were often associated with the potential psychosocial benefits that could accrue to the family and the inability to prohibit access to existing technology.

The study also examined the clinicians' attitudes about confidentiality and privacy of genetic data within families. Most believed that it was very important for patients to share genetic information with biological relatives, and most were willing to counsel families with varying degrees of directiveness regarding disclosure, although rarely were clinicians willing to disclose information without their patients' consent. However, when the proband was not a patient but a first-degree relative, most clinicians were willing to disclose this information if their relative would not.

The data support the moral proposition that different practices ought to exist in professional and personal contexts as different moral relationships entail different moral obligations. I conclude with a discussion of what this may imply for clinical practice with respect to access and confidentiality in genetic testing of children.

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Genetic Testing in Adolescents

Lainie Friedman Ross, MD, PhD

Two controversial types of genetic tests in children are tests for late-onset conditions for which no treatments exist in childhood and carrier testing. In this project, I examine whether adolescents should be tested for these conditions and, if so, whose consent should be necessary.

I begin with an examination of the benefits and burdens of testing adolescents for genetic conditions that have no immediate impact on their medical well-being. I conclude that there may be valid reasons why adolescents and their parents would want this information.

I then examine whose consent is necessary. I advocate for the consent of both parents and adolescents. I consider and reject the arguments that support the adolescent's right to consent to these tests without parental involvement. First, I review the growing body of literature that supports the adolescent's right to make health care decisions for himself or herself. I challenge the interpretation of the scant data and argue that, even if adolescents have decision-making capacity (DMC), DMC is necessary but not sufficient for health care autonomy. With respect to genetics, I argue that the prevailing beliefs about the adequacy of adolescent DMC are even weaker given the familial implications of genetic information, the decision of many adults not to be tested, and the inability of most adults to utilize probabilistic information. Second, I consider the argument to expand adolescents' rights in genetics by extrapolating from the specialized consent statutes. These statutes exist in all 50 States and allow some adolescents to make health care decisions regarding contraception, substance abuse treatment, and mental health care without parental involvement. But these statutes were not written because of adolescent DMC; rather, they rely on the pragmatic (although not empirically proven) belief that adolescents will delay care unless they are guaranteed confidentiality.

I also consider and reject the arguments that support parental rights to consent for these tests over the adolescent's dissent. On the other hand, I do not support a prohibition of testing of young children without their assent. I examine the pragmatic and ethical issues that a prohibition would raise, including the possibility that parents may seek testing prenatally, when their rights may be greater.

Living with Genetic Risk: The Well Sibling Revisited

Joanna H. Fanos, PhD

Studies on autosomal recessive disorders have shown that the illness and death of a sibling have far-reaching ramifications on the surviving sibling's life. In cystic fibrosis (CF), our early studies indicated siblings had survival guilt, global anxiety, fears of an early death for themselves, fear of intimacy, excessive concern for others, somatic expressions, including identification illnesses, and sleeping difficulties, including persistent nightmares. Following the identification of the CF gene, our subsequent NIH/ELSI-funded study found sibling resentment, assumptions of carrier status, wishes to be a carrier, and guilt in receiving negative results to be positively correlated. In ataxia-telangiectasia (A-T), we found that, unlike CF, the visibility of A-T caused less resentment of attention given to the affected, less guilt, and less identification or idealization of the affected, but more embarrassment and shame. Unlike CF, the dynamic in A-T-affected families is one of burden rather than trauma. In X-linked severe combined immune deficiency (XSCID), our NIH/NHGRI-funded study showed the period of isolation during the boy's bone marrow transplantation placed great stress on the family, and parental inability to mourn the loss of a son who died created an atmosphere heavy with family secrets. Long-term difficulties for daughters included the desire to repair her mother's loss of her own child, as well as attempts to undo feelings of being flawed by heightened wishes to bear a healthy son.

Thus, the specific phenotypic expression of genetic illness predisposes the way the unaffected sibling will encounter problems. In CF-affected families, there is a need for improving family communication and helping siblings with feelings of resentment and guilt. In A-T-affected families, help is needed in locating caregiving resources so that parents will not burden well children; siblings need help managing feelings of embarrassment and shame. In XSCID-affected families, parents need help balancing needs of well siblings with needs of the affected son, as well as help with mourning so that family secrets will not prevail. Geneticists must be alert to understanding characteristic differences of disorders they encounter to offer the most appropriate support to affected families.

Disclosure of Genetic Testing Results Within Families: The Case of Hemophilia A and Its Research Implications

J.R. Sorenson, T. Jennings-Grant, J. Newman, J. Thrasher

Carrier status disclosure histories were collected by telephone interviews with 98 women from 34 hemophilia A families 6 months after learning their carrier status. 22 women tested positive and 76 tested negative for the mutation in their respective

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families. Data were collected on self-reported disclosure to spouses, parents, spouses' parents, sisters, brothers, and children.

Disclosure of carrier status within Hemophilia A families is selective: 60 percent (51/85) of women with partners reported talking with their respective partners about their carrier status; 56 percent (38/68) of women with children told at least one of their children; and 82 percent (31/38) of women who told at least one of their children told all their children. Women were more likely to report talking with at least one of their sisters (73 percent) or all of their sisters (59 percent) than with at least one (44 percent) or all (30.9 percent) of their brothers. Women were more likely to talk with their mothers (56.1 percent) than with their fathers (20.4 percent). Only 14.3 percent talked with their spouse's mother and 6.2 percent with their spouse's father.

Preliminary analyses suggest disclosure of carrier status within hemophilia A families may be related to several factors. Carriers, more than noncarriers (47.6 percent versus 6.2 percent), reported concern about when to tell their children their carrier status. Similarly, more carriers than noncarriers (38.1 percent versus 14.1 percent) reported concern about what to tell their children. Women planning or unsure about a future pregnancy were more likely to report talking with their respective partners about their carrier status than those not planning a pregnancy (88 percent versus 66 percent).

Disclosure of carrier status to relatives by tested individuals is not universal within kinships. Understanding the barriers/facilitators of such disclosure could lead to improved genetic testing/counseling protocols and provide needed information on the effects of genetic testing on families rather than just individuals. Informed consent challenges to conducting research on genetic testing disclosure within families, along with several study design and measurement issues for conducting such research, will be discussed.

Communication of BRCA1 and BRCA2 Results to At-Risk Relatives

Mary B. Daly, Mark Itzen, James Babb, John Malick, Andrew K. Godwin, Betsy Bove, Josephine Wagner Costalas

Recent advances in molecular genetics have identified a number of genes associated with inherited susceptibility to breast and ovarian cancer. Genetic screening for mutations in BRCA1 and BRCA2 is already being incorporated into clinical practice to enhance prevention and control strategies for individuals at increased risk for developing these cancers. Very little data, however, are currently available on the impact of communicating genetic test results within the family. Results from a survey designed to assess patterns of communication within families shortly after an individual receives test results were analyzed to detect barriers or difficulties encountered when these individuals attempt to communicate their genetic results to their at-risk relatives. We have shown that individuals are disclosing their test results to their relatives but that there is a significant level of difficulty and anxiety in explaining this information. The information also appears to be creating a level of anxiety among family members receiving the information. More difficulty was reported in discussing results with siblings than with adult children. The nature of the test result itself (positive, negative, or indeterminant) was also a factor in determining levels of difficulty and distress in communicating test results. Further studies will be needed to monitor the impact of genetic test information notification and to develop communication skills to help individuals deliver their personal genetic test information to their relatives.

Section 11: Testing — Prenatal

Newborn Screening and Carrier Detection for Cystic Fibrosis: Complementary or Contradictory?

Benjamin Wilfond

Over the last decade, different regions around the globe have developed programs for CF newborn screening and carrier detection for cystic fibrosis (CF). Some regions have adopted both while others have adopted neither. Whether such approaches are complementary or contradictory may depend on the assumed goals and efficacy of such programs. Some advocates of newborn screening (NBS) describe the goals to substantially improve the clinical outcome of children with CF, while others emphasize the potential psychosocial benefits (and occasionally medical, as well) of avoiding delays in diagnosis. For carrier detection, one goal would be the reduction of births of children with CF, while for others the goal is the provision of information to improve reproductive decision-making. There are only limited data about the achievability of these goals, especially the more concrete clinical goals of either improved clinical outcome from NBS or reduction in incidence from carrier detection. The clinical goals may be more contradictory than the psychosocial goals. For example, if NBS dramatically improved the prognosis of children with CF, there may be less impetus to develop a carrier detection program to reduce incidence. Alternatively, if the incidence of CF decreased, the positive predictive value of NBS would diminish, and this makes

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NBS less attractive. While programs with more modest goals, such as diminishing parental anxiety from early diagnosis or offering options for family planning, are more compatible, it is less clear that these goals would justify the cost of such a program in an era of fiscal scarcity. The determination of the goals of programs is in part an empirical question about outcome but has a normative component than can even influence the design of a program.

Can a Eugenic Program Be Morally Right and Politically Correct? Thalassemia Prevention in Cyprus

Ruth Schwartz Cowan

Both countries on the island of Cyprus (the Republic of Cyprus and the Turkish Republic of North Cyprus) have had mandated genetic screening programs for thalassemia for almost 20 years. The populations of both countries as well as the most significant social institutions (including the Cypriot Orthodox Church) not only have signified their assent to these programs but also have supported them financially.

This paper will examine the history of both programs as well as the nature of thalassemia in order to explain not only how social consensus has been achieved but also why so many Cypriots regard the programs as both eugenic and morally correct.

The Disability Critique and Prenatal Testing

Erik Parens, Adrienne Asch

The first purpose of our ELSI-funded project was to give the disability community critique of prenatal genetic testing (PGT) a sympathetic and critical hearing. That critique has two prongs. The first suggests that PGT is morally problematic insofar as it (1) expresses a hurtful message to and about people with disabilities and (2) expresses a problematic conception of and attitude toward parenthood. The second prong suggests that PGT is based on misinformation; insofar as users of tests to screen out fetuses with disabling traits usually know nothing about what life is really like for children with disabilities and their families.

The written materials our project produced show that we achieved that first purpose; they offer a sympathetic and critical review of both prongs of the disability critique.

The second purpose of our project was to attempt to draw lines between tests that it would be reasonable for professionals to offer and those it would not be reasonable to offer. Two arguments made it impossible for us to reach consensus on drawing lines. The first, waged primarily by some mainstream bioethicists, was that line-drawing is paternalistic. The second, waged primarily by some members of the disability community, was that line-drawing would either reinforce dominant conceptions of normality (if the line were between disabling and non-disabling traits) or would divide the disability community (if the line were drawn between severe and not severe disabling traits).

Although we did not achieve our second stated purpose (drawing lines) and did not reach consensus about the substantive question Where should lines be drawn? we did reach consensus about the procedural question How should prenatal genetic tests be offered? The disability community critique convinced our group that if potential parents are going to give truly informed consent to receive prenatal tests, they must be helped to learn much more than they know now about what life really is like for people with disabilities. That critique also convinced our group that medical and genetics professionals must learn much more than they know now about what life really is like for people with disabilities and their families.

Genetic Testing, Disabilities, and Quality of Life

David Wasserman, JD, Robert Wachbroit, PhD, Jerome Bickenbach, PhD

This presentation will report on research under way in an ELSI-funded project on the significance of disability in two health care contexts: the utilization of prenatal genetic testing and the development of quality of life measures for health assessment and allocation. Decision-making in both these contexts may be informed by controversial assumptions about the relevance of disability for quality of life, and of quality of life for the value or priority of lives. In both contexts, genetic technology is having, or will have, a major impact, enabling decision-makers to predict impairments long before their onset. This presentation will highlight three issues on which the research focuses:

1. Prenatal testing and two kinds of determinism:

Critics claim that the demand for prenatal genetic testing rests on two dubious assumptions. One is that genes produce physical and mental characteristics with little environmental mediation; the other is that certain physical and mental characteristics produce great personal and social disadvantages with little environmental mediation. These assumptions are worth distinguishing, but both may be associated with a broader tendency to see biological causes as predominant and environmental factors as inert.

2. Public policy and private decisions:

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Some commentators have argued that we should distinguish the reasons for the public sponsorship of prenatal genetic testing from the reasons for the private use of such testing. Does it make sense to condemn the former but not the latter? Is the expressive significance of the former clearer or more objectionable than that of the latter? How relevant is the history of prenatal genetic testing for the appraisal of public or private decisions?

3. The relevance of expected quality of life for decisions to save and create lives:

Several objections have been raised to allocating scarce health care resources on the basis of expected outcome. Critics argue that even if outcome measures are restricted to the expected benefits and harms for the individuals needing care, a reliance on such measures slights other values critical to fairness and discriminates against people with disabilities. Do these, or analogous, objections apply to a reliance on expected benefits and harms in deciding which lives to create or which pregnancies to continue?

Ethical and Legal Issues in Reproductive Screening for Susceptibility Mutations

John A. Robertson

Progress in sequencing and annotating the human genome will lead to an increasing amount of DNA-based information that couples might wish to use in reproductive decisions. The use of genetic information at carrier, preconception, preimplantation, or prenatal stages of reproductive decision-making is now widely, but not unanimously, accepted if (1) genetic counseling and informed consent have occurred and (2) the genetic information would be helpful in avoiding offspring with serious genetic disease. As knowledge about late-onset and disease susceptibility alleles grows, the question of using that information in clinical reproductive practice will also arise.

Screening of embryos and fetuses for susceptibility mutations, however, is highly controversial, for it raises questions about respect for prenatal life and the propriety of selecting the genes of offspring, as well as issues about the scope of reproductive autonomy. Much more discussion of the ethics of such techniques is needed before clinicians offer screening for susceptibility genes.

As genomic information increases and embryo screening protocols improve, many patients may request preimplantation genetic diagnosis (PGD) to screen out embryos with susceptibility mutations for diabetes, heart disease, cancer, and the like. Some programs already offer PGD for gender selection, and others screen embryos to ensure histocompatible stem cell donors for existing children. With time, requests to screen embryos for intelligence and other nonmedical traits might also occur. Although highly speculative, the prospect of nonmedical uses strongly influences the debate over susceptibility mutation screening.

To identify relevant issues and stimulate ethical discussion, this paper addresses the ethical, legal, and medical issues that susceptibility mutation testing raises at preconception, preimplantation, and prenatal stages of reproductive decision-making. It describes possible disease candidates for susceptibility mutation screening and shows how competing concerns about respecting embryos and fetuses, not objectifying or commodifying offspring, and respecting family desires to have healthy offspring inform the ethical, legal, and policy debate.

Women's Preferences for Miscarriage Versus Down Syndrome-Affected Birth: Implications for Prenatal Testing Guidelines

Miriam Kuppermann, Robert F. Nease, Jr., Lee A. Learman, Elena Gates, Bruce Blumberg, A. Eugene Washington

Background: Current prenatal diagnostic testing guidelines recommend offering chorionic villus sampling or amniocentesis to women whose risk of carrying a fetus affected by Down syndrome is at least as great as the procedure-related miscarriage risk. This guideline implicitly assumes that women find these two outcomes equally burdensome.

Objective: To determine how pregnant women of varying ages, race/ethnicities and socioeconomic backgrounds value procedure-related miscarriage and Down syndrome-affected birth.

Methods: Cross-sectional study of 534 sociodemographically diverse pregnant women. We assessed preferences for procedure-related miscarriage and the birth of an infant affected by Down syndrome using the time tradeoff (TTO) and standard gamble (SG) metrics and calculated differences in these scores (preference score for procedure-related miscarriage minus preference score for Down syndrome birth) for each metric. We also collected detailed information on attitudes toward Down syndrome, abortion, and miscarriage.

Results: On average, we found that procedure-related miscarriage was preferable to Down syndrome-affected birth, as evidenced by the positive differences in preference scores (TTO difference, mean = 0.09; SG difference, mean = 0.11; $p<.001$ for both metrics). These preference differences varied substantially, with only about a quarter of women (24.3 percent and 29.8

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percent for TTO and SG, respectively) providing equivalent scores for these two outcomes. We also found significant associations between the responses to all attitudinal questions and preference difference scores in the expected direction, with women whose attitudes favored testing having stronger preferences for miscarriage over Down syndrome. Among women ages 35 and older, those who underwent prenatal diagnosis had higher preference difference scores than women who did not (TTO, 0.13 versus 0.02, $p=.001$; SG, 0.15 versus 0.01, $p=.005$). Finally, we found no association between age and preference difference score ($p>.10$ for both metrics).

Conclusions: On average, pregnant women find Down syndrome-affected birth more burdensome than procedure-related miscarriage, calling into question the current equal risk threshold for offering prenatal diagnosis. Moreover, individual preferences for these two outcomes vary profoundly but are not associated with maternal age. The assumption underlying the current guideline does not hold, and new guidelines should be developed that better reflect the values of informed patients.

"I'm Not the One They're Sticking the Needle Into": Latino Couples, Gender Rationales, and the Discourse of Reproductive Rights in Amniocentesis Decision-Making

Susan Markens, Carole Browner

Although the past decade has witnessed the widespread normalization of prenatal diagnostic testing, there has been little research on the role men play in this reproductive arena or the technology's possibilities for reinforcing and/or transforming gender roles and power relations. In this paper, we analyze male partners' participation in the amniocentesis decisions of women of Mexican origin living in California. We interviewed 147 women who were at high risk for bearing a child with an anomaly, along with 120 of their male partners. The goals of this research are to interrogate and test two common assumptions. Our primary concern is to determine whether the case of prenatal testing illuminates whether the general rise in new reproductive technologies poses a threat to women's reproductive autonomy and decision-making. Second, we are also interested in whether the decision-making process surrounding prenatal testing reveals gender/power dynamics among couples of Mexican background that can be characterized by male dominance and female subservience. We argue that both assumptions about actual or potential male dominance are largely false. First, we found an array of decision-making styles among these couples of Mexican origin, and for the most part women were active agents in the decision-making process. Second, many of our informants viewed the offer on amniocentesis as a positive opportunity to create or solidify egalitarian relationships with their partners, particularly by sharing equally in their responsibilities as parents. Our findings challenge critics of the new reproductive and genetic technologies who assume that the technologies are inherently male attempts to control women's bodies and the processes of reproduction. Instead, this analysis suggests that we should further evaluate the circumstances under which these technologies are deployed and the means through which they may open up avenues for shared responsibility between women and men for parenting decisions.

Latinas, Amniocentesis, and the Discourse of Choice

C.H. Browner, H. Mabel Preloran

Pregnant Latinas who learn through amniocentesis that their fetuses may be gravely ill or disabled may find themselves faced with a terrible dilemma. No treatments are available for virtually all the conditions detected through fetal testing; abortion is the only intervention medicine can provide. Yet Latino culture teaches that all human life should be cherished, regardless of health, and the religions most Latinas practice prohibit abortion. Many Latinas find amniocentesis additionally problematic because it can cause injury or miscarriage. Yet 60 percent of the 147 "high risk" Latinas in our study agreed to amniocentesis. Our research was designed to investigate the factors that lead Latinas who are told their fetuses are at high risk for a birth defect to either accept or reject amniocentesis.

Those who accepted were similar to those who did not, in age, education, household income, reproductive history, family history of birth defects, religious background, and religiosity. Although foreign-born Latinas were significantly less likely than U.S.-born Latinas to agree to amniocentesis, neither acculturation score nor, for immigrants, length of time in the United States was predictive. Although women who accepted were significantly less likely to be strictly opposed to abortion, most acceptors and refusers alike deemed abortion personally unacceptable. To better understand what leads some Latinas to transgress cultural norms and agree to amniocentesis, we are examining women's narratives. Here we offer one woman's account and analyze how she rationalized her decision to make amniocentesis acceptable to herself, her family, and her community. Our quantitative results and narrative data from the larger sample of 147 will be drawn on to contextualize the case material and draw broader generalizations.

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Cultural and Noncultural Barriers to the Provision of Genetic Services for Mexican-American Women in Five South Texas Cities

M.C. Aguilar, M.L. Urdaneta, S. Suther, S. Cameron, S. González-Bogran, V. Enciso, C.I. Kaye

While 3 percent of all live-born infants in Texas have an identifiable birth defect, only 20 percent receive genetic evaluation (TEXGENE data). There is disproportionate underutilization of services by Medicaid clients, most of whom are Mexican-Americans. This project identified cultural and noncultural barriers to the provision of genetic services. Goals were to identify barriers, to assemble a culturally sensitive information/training manual, and to implement and evaluate effectiveness of this manual in four South Texas cities. Using a qualitative and ethnographic approach, a 50-question interview guide was constructed. Seventy-five interviews were completed, including interviews with 23 clients, 25 health care providers, and 20 folk healers (parteras and curanderas). Barriers to genetic services in South Texas identified included (1) language and communication, (2) cultural differences, (3) poverty, and (4) system issues. Clients and folk healers identified the following causes for genetic disorders: (1) causes by natural forces (eclipses of the sun/moon), (2) causes by emotions (sudden susto or fright/stress), (3) causes by behavior (unacceptable/censored), and (4) causes by outside forces (evil-doing/witchcraft). This study indicates that 49.9 percent of clients have knowledge of and use traditional healers, while 45.5 percent of clients have knowledge about, but do not use, traditional healers. Study analysis suggests the need for educational and preventive outreach programs presented in idiomatic Spanish. The development and implementation of culturally sensitive training manuals for both traditional and nontraditional health care providers are required, although noncultural barriers stemming from political and socioeconomic conditions will remain pervasive and problematic.

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Attitudes Toward Genetic Testing and Reproductive Options in the Achondroplasia Population

Barbara Biesecker, Holly Gooding, Karina Boehm, Don Hadley, Clair Francomano

Achondroplasia is the most common inherited form of dwarfism and is caused by a mutation in the gene encoding fibroblast growth factor receptor 3. Since the discovery of the mutation in 1994, commercial prenatal tests have become available. Couples both with achondroplasia face a 50 percent chance of having a child with achondroplasia, and a 25 percent chance each of having a child of average stature or with lethal homozygous achondroplasia. Scholars claim that affected couples may terminate pregnancies of average-statured fetuses because they desire a child with achondroplasia (Reindal 2000).

This study sought to collect empirical evidence for the awareness, interest, and use of prenatal genetic testing for achondroplasia among affected individuals. A total of 171 affected individuals (117 women) answered a self-report survey instrument. Of these, 81 respondents were married and the average age was 40.6 years. Sixty-eight percent of respondents were aware of the prenatal test. While only 7 percent of respondents had used the test, 61 percent indicated that they planned to use it. Most affected individuals believe it would be important to know whether the fetus has the lethal form of achondroplasia but consider knowing whether the fetus has achondroplasia unimportant. Respondents were evenly split over whether they would choose abortion for the lethal form of achondroplasia, but the vast majority responded that they would not choose to terminate if the fetus was either affected with achondroplasia or of average-stature. Results are not explained by personal beliefs in abortion. These results offer evidence against the assumption that affected individuals are interested in terminating average-statured fetuses and reaffirm the strong commitment of the little people community to support the birth of affected individuals.

Section 12: Philosophical and Conceptual Issues

Technology and the "Safeguarding of Being": Toward a Revised Ethic in Genetic Nursing Care

Ellen Giarelli, EdD, RN, CS

Purpose: The purpose of this paper is to propose a revised ethic of genetic nursing care based on a philosophical analysis of genetic testing (GT) for predisposition to the development of disease.

Background: In a utilitarian manner, advocates for GT for predisposition to disease stack up potential human gains while critics counterbalance these with predictions of new eugenics and social discord. Ethical issues, such as paternalism, privacy and confidentiality, and the risk-to-benefit ratio, have been addressed since the inception of the Human Genome Project. These issues continue to surface and may take on a new significance as genetic technology becomes generally accepted and widely used by consumers and health care providers. The professional nurse's principled intent to do good motivates the search for

ways to integrate this technology with practice. The ability to do good, however, is limited by the state of the continually evolving science, the impossibility of doing all good, and the impossibility of avoiding all harm. A moral philosophy of GT probes the ethical nature of the relationship between technology and human existence, based on values intrinsic to both. A moral philosophy of genetic nursing care evolves as we think about the question How does GT transform experience and clarify the meaning of human existence?

Methods: In this paper a philosophical analysis of GT is conducted from a Heideggerian phenomenological perspective. First, the essence of GT is related to Heidegger's notion of "Being-in-the-world." Then, potential threats to Being are identified as they arise from the ordinary and prescribed use of GT for predisposition to disease. Finally, a revised nursing ethic for genetic care is proposed.

Conclusions: The essence of GT has form, process, and worth. It is a means to an end and a human activity. It brings forth an aspect of humanity that could not be known in any other way. By revealing the genetic nature of a person, the nature of human existence is given a clinical presence. During this geneti-poiesis, humanity is forever transformed. GT is the instrument by which the nature of humanity may be revealed for what it is and for what it might be. Genetic technology reveals our Being-in-the-world as the ordered presentation of the human genome, which is then held in standing reserve as extensive databases of biochemical information. Threats to Being may be found: in the purpose of GT, in the assumption of a net benefit, in the intention to benefit humankind, when correct is considered to be true, and in the uncertainty of what should be a guiding value. Ethical nursing practice has evolved from loyalty to advocacy but must now be revised to the guardianship of Being. The new moral imperative characterizes an existential nursing care that aims to preserve the meaning of our existence as whole beings in the world. The revised ethic will help practitioners as they apply a fundamentally reductionist science.

Reductionism, Determinism, and Discrimination: Tightening the Chains

Evelyne Shuster, PhD

The first ELSI-funded workshop, a decade ago, was devoted to developing a prioritized research agenda for ELSI. The fourth of four items identified was how the Human Genome Project would affect our concepts of disease, normalcy, and humanness. At the heart of this item is reductionism and determinism, and these concepts also govern genetic discrimination. My own contribution to that workshop (and later book, *Gene Mapping: Using Law and Ethics as Guides*, Annas and Elias, eds., Oxford 1992) was a chapter entitled Determinism and Reductionism: A Greater Threat Because of the Human Genome Project? As central as these concepts are to the HGP, it seems fair to conclude that they have been inadequately attended to and that we need to further engage in a critical examination of their meaning.

Health is now genetic. Genes threaten to become the measure of all things. But we humans do not live our lives at the genetic level. French philosopher Georges Canguilhem perceptively noted decades ago that it is a fundamental mistake to think that, by understanding life at the molecular level, we can understand anything about living. In his words, one does not scientifically dictate norms to life. Likewise, his star student, Michel Foucault, warned that the geneticization of diseases would create a new biological underclass and lead to the systematic ostracism of those labeled genetically unfit, the result being a biopolitics of the population. Genetic reductionism and determinism thus align with genetic discrimination and domination and threaten to subvert a beneficent medicine by converting it into a powerful tool for oppression.

This presentation will explore the meaning of life in the context of the genetic imperative to accept reductionistic and deterministic thinking as a necessary price for progress toward a longer life, absolute remedies, and virtual immortality. Only the living can meaningfully inform the living. To resist imputing meaning to life from molecular medicine, we must articulate our goals clearly and develop strategies to meet those goals. These goals cannot (or at least not only) be to eradicate all diseases and live a longer (or even a healthier) life in a youthful body.

Being a Person in the Human Genome Era Is Unreal

Howard M. Ducharme

Prior to the Human Genome Project era, there was very little science, biology, and genetics engaged in the philosophical, ethical, common sense, and religious explanations of the nature of a person, e.g., Plato, Augustine, Aquinas, Descartes, Hobbes, and Hume. Obviously, this is no longer the case. In fact, the pendulum has swung to the opposite extreme — where contemporary science, biology, neurology, and genetics are assumed to be the necessary and/or sufficient determinants that explain what it is to be a person, e.g., E.O. Wilson, Richard Dawkins, Daniel Dennett, and Antonio Damasio. The most outstanding evidence of this pendulum swing is found in the work of contemporary theologians: Their recent holistic (nonreductive materialism) theories reject the traditional soul and replace it with biological, neurological, and genetic substitutes, e.g., *Whatever Happened to the Soul?*, edited by Brown, Murphy, and Malony.

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This paper gives two arguments. First, it argues that there are science-first theories and person-first theories of what it is to be a person. Science-first theories identify and give explanatory priority to genetic facts, neurobiological data, and neurogenesis discoveries. These substances and their activities are assumed to be necessary if not sufficient entities to explain the nature of a person. The common experiences of being a person (e.g., free will, moral capacity, moral worth, self-consciousness, personal identity, aesthetic and religious experience) are subsequently explained by, restricted to, limited by, and/or reduced to genetic and neurobiological functions. The direct and common sense experiences we have of being a person are then explained away as mere folk psychology — old beliefs that have not yet come into the Age of Science.

Person-first theories give priority to basic, immediate experience of being a person. These data are available to all persons. It is open to verification and critical refinement, and in contrast with knowledge of DNA and neurobiology, it is equally available and accessible to the 99 percent of the world's population who are nonscientists and nonacademics. It is direct self-knowledge of what it is to be a person. These data can be refined and corrected, but in person-first theories are assumed true until proven to be false. The nature of genes and neuronal activity are not necessarily restrictive in telling us what it is to be a person. Persons are nonreductive moral agents who exist in the world, whose basic activities may be mediated, modified, and influenced by genetic and neurological factors. But persons are not reducible to genetic activity and/or neurological brain functions. We have first-person knowledge of ourselves as persons (in contrast with inferential explanations of ourselves via abstract theories). This direct knowledge includes being self-conscious, having free will, enduring with a strict personal identity through time in spite of the ever-changing neuronal and genetic identity of our bodies, having an irreducible moral nature and inherent moral value, and being aesthetic and religious beings.

Second, this paper argues that the science-first theories are academically fashionable, but they entail problematic antirealist theories of persons, e.g., that there really is no free will, no self, no irreducible moral agent/moral value, no strict personal identity over time, and no traditional soul (hence, approximately 85 percent of the world's population need to learn that they have a scientifically falsified religious belief that they must give up).

This material is sketched out to engender critical reflection on genetic reductionism, materialism, and nonreductive materialism. Simultaneously, it provides the basis for retention of basic, common sense knowledge of what it is to be a person and moral agent — information that is accessible to all, scientist and nonscientist alike.

The Gene Cathedral: Values and Ethics in Genetics Research

David Lewis, PhD, Manu Rangachari, Rahima Visram, Gail Coulas, MA

Objective: In light of events surrounding therapeutic cloning, we sought to examine the relationship between gene therapy research and biomedical research centers operating at Roman Catholic-sponsored institutions. We therefore sought (1) to test the hypothesis that expert treatments of ethics in gene research, whether conducted by scientists, bioethicists, or theological ethicists, follow a biomedical model of ethics focusing on the individual patient's rights and choice and (2) to test the hypothesis that popular concerns over genetics research are not represented in ethical work conducted by these experts.

Method: We searched the MEDLINE and AHRIS databases to determine whether gene research in Catholic-sponsored institutions is represented there. We then examined the applicable ethical precepts of Catholicism and of relevant secular institutions such as funding agencies. Finally, we searched BIOETHICSLINE, MEDLINE, SocioFILE, and Religious Abstracts databases to determine how genetics research was viewed by various expert groups. We used latent content analysis to identify themes and counts.

Results: We found remarkable congruence in Catholic and secular ethical guidelines relating to gene research. However, we further found that the popular literature tended to focus far more on cloning than on other aspects of gene research and was much more hostile to gene research than any of the other sources. On the other hand, expert sources were quite similar in their levels of approval and specific subject matter.

Conclusions: Genetics researchers may be deemphasizing connections with Catholic-sponsored institutions. Since the religious and secular guidelines are nearly identical, there are few overt impediments to conducting gene therapy research in Catholic-sponsored institutions. However, there is a potential for adverse reactions to such research in the popular press; moreover, expert views are fluid and may not address areas of popular concern.

The Human Genome Project and Women

Mary B. Mahowald

The goal of this project was to examine the impact of advances in genetics on women, to identify areas of inequality related to these advances, and to explore ways by which inequality between men and women related to genetics may be eliminated or reduced. We first identified biological differences, some related to reproductive roles and others to genetic conditions. In the

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latter category are conditions manifested more often or exclusively in either sex, those whose phenotypes are different in males from those in females, those affecting males or females more severely, those determined by the sex of the transmitting parent, and those in which symptoms are exacerbated in pregnancy. We also identified psychosocial differences in the impact of genetics on women. Caregiving roles, cultural influences, and the impact of poverty were among the contexts explored here. Different attitudes of men and women toward the genetic tie to offspring were also investigated.

We then considered how to address the impact of genetics on women in accordance with the principle of justice. We developed a concept of justice that requires identification of differences, determination of whether the differences are associated with inequality, and efforts to change changeable differences associated with inequality and to introduce measures that reduce inequality associated with differences that are not changeable.

Next we focused on three specific conditions — cystic fibrosis, sickle cell anemia, and breast cancer — examining their impact on women through presentations by pertinent experts as well as women affected by the conditions. We also considered policy issues regarding genetic information, particularly as these relate to insurance and employment.

In each of the areas investigated, a public lecture by a relevant expert was offered, and a conference open to the public concluded the grant period.

Articles informed by this research were published in *The Milbank Quarterly*, *Women's Health Issues*, and the *Journal of Women's Health*. Empirical and theoretical components of the project are most fully developed in *Genes, Women, Equality* by Mary Briody Mahowald (Oxford University Press 2000), the writing of which was funded by other sources.

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Section 13: Genetic Enhancement

Anticipating Enhancement: Ethical, Legal, and Social Issues

Eric Juengst, Maxwell Mehlman, Thomas Murray

The goal of our ELSI project has been to delineate the major ethical, legal, and social issues that would accompany the use of genomic information to enhance normal traits in individuals and families and to recommend the precedents that best illuminate those issues for policy-making purposes.

Questions about the use of genomic information for enhancement purposes cut across the development of all three types of new medical tools expected from the Human Genome Project: DNA-based tests, new drugs and biologicals, and new human gene transfer therapies. So far, most ELSI-funded research has focused on the policy implications of only the first category of genomic products: improved molecular diagnostics and risk assessment tests. But the other two kinds of genomic medical tools are no longer hypothetical. By addressing the issues of "nonmedical use" shared by all three sets of tools, our project is designed to bridge the work to date with the challenges that await the Human Genome Project on the other side of its now infamous "therapeutic gap."

Our project has moved through two phases to date: first attempting to learn what we can about the issues to anticipate from a range of biomedical precedents, and then drawing from those analyses a set of basic ethical considerations to inform our social policy. We are now jointly authoring a monograph that attempts to articulate an ethics of enhancement at the levels of personal decision-making, professional ethics, and public policy-making.

Meanwhile, our initial work has already identified three critical challenges in applying our analysis to the development of the effective social policy in this area. First, most interventions that can be used for enhancement are likely to be initially developed and approved for therapeutic use. However, once so approved, the current regulatory structure provides no adequate means of managing the off-label use of such interventions for enhancement purposes by clinicians and their clients. Second, any enhancement interventions performed on pre-implantation embryos are likely to be undertaken in the largely unregulated context of clinical reproductive biology and infertility medicine. While our initial project allows us to outline the considerations relevant to professional ethics in this area, it is still not clear how those standards would be best enforced. Finally, the availability of either pre-implantation or post-implantation genetic enhancement interventions will also depend on our policies regarding access to these interventions outside the boundaries of the United States. We are now undertaking a close-grained analysis of these three problems as they will challenge the management of genetic enhancement technologies in order to develop specific policy recommendations for public policymakers that would allow the issues to be addressed within the framework of considerations we will have already set out.

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The Social and Ethical Dimensions of Genetic Enhancement Technologies

David J. Rothman, Sheila M. Rothman

This paper will explore the social and ethical implications of genetic enhancements. It will first argue that the distinction between cure and enhancement, however commonsensical, is artificial and unable to influence the biomedical research agenda, determine medical practice, or guide health policy. It will demonstrate that in both research and practice, initial efforts to seek a cure are transformed, quickly and almost inevitably, into enhancements. Second, the paper will explore the characteristics and conditions most likely to undergo enhancement, including memory and longevity. It will then analyze the sources of opposition to genetic enhancement, including claims that it is unnatural, that it is frivolous, that it is especially subject to abuse (as per the history of eugenics), and that it is likely to violate principles of distributive justice. The paper will then argue that such objections are not determinative, neither in their intrinsic logic nor in their likelihood of restraining the development or the use of enhancement technologies. To the contrary, the engine driving enhancement runs on many cylinders, and the paper will address each one in turn: the mind set of science (and its impatience with the natural), medicine (and its readiness to serve patients' self-assessed needs), commerce (the role of drug and biotech companies), and culture (the emphasis on peak performance). Finally, the paper will conclude that the critical problem intrinsic to enhancement use involves risk assessment. Each individual will be forced to make his or her calculation of risks and benefits without significant guidance on how to make choices.

Ethical and Social Issues in Human Germline Modification: A Retrospective View From 1990 to 2000

David B. Resnik, PhD, Pamela J. Lanjer, PhD

From 1970 to 1990, many different authors, organizations, and institutions addressed ethical and social issues in human germline modification (HGM). These early debates addressed issues that were, for the most part, still in the realm of science fiction. But the tenor of the dialogue about HGM changed with the first human somatic gene therapy (SGT) trials in the early 1990s. Scientists, clinicians, and scholars began a serious and thoughtful debate about the pros and cons of HGM and under what conditions it might be conducted. Additionally, the American Association for the Advancement of Science organized a work group that published a report on HGM, the Hastings Center sponsored several projects with a direct bearing on germline modification, and the National Bioethics Advisory Commission held hearings and wrote reports on topics closely connected to HGM. Some of the key issues that emerged from these discussions included the short-term and long-term risks of HGM to children and future generations, the rights of parents to use HGM, the difference between therapy, prevention, and enhancement in human genetics, issues of social justice, problems with parental expectations and demands, discrimination and disability, and questions about changing human nature. By the late 1990s, important events in biotechnology, such as mammalian cloning, research on embryonic stem cells, the burgeoning assisted reproduction market, the booming biotech industry, and the near completion of the Human Genome Project (HGP), made HGM appear to be realistic and perhaps technically feasible. As the 21st century begins, several writers have pointed out that some relatively safe forms of HGM, such as egg-cell nuclear transfer (ECNT), have already occurred and that some proposed SGT protocols can inadvertently modify the human germline. The HGM debate itself has also shifted from theoretical issues to more practical concerns about clinical trials, oversight, regulation, government funding, and the role of the private sector. This presentation will give a retrospective overview of the HGM debates in the 1990s and summarize some of the key points of contention and discussion. It will also address some of the important questions and issues that lie ahead.

The Conceptual Challenges of Regulating Genetic Enhancement

Eric T. Juengst

The foundation for the public regulation of human gene therapy in most countries is a simple four-celled matrix of policy-relevant considerations. The matrix is created by crossing the two possible kinds of cellular targets for human gene transfer — somatic or germ-line cells — with the two possible goals one might have in performing it: treatment for disease or human "enhancement" (Walters 1986). Both distinctions are widely debated, but in practice they have been crucial to human gene transfer research by creating the boundaries that protect the one form of human gene transfer that we are scientifically ready to pursue: somatic cell gene transfer interventions aimed at treating disease. Increasingly, however, policymakers are being asked to directly address these boundaries, in anticipation of scientific advances.

Much of that recent literature has been focused on the line between somatic cell and germ-line gene therapy. But while the somatic/germ-line distinction is accused of lacking adequate ethical force, the conceptual line it draws is at least clear. The treatment/enhancement distinction, on the other hand, often seems in danger of evaporating entirely under conceptual critiques

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even before the question of its moral merits is entertained. If "enhancement" is to keep serving as a major policy boundary, we should at least be clear about just what it demarcates. My primary goal in this presentation is to apply the work of our ELSI project to this question. I will argue that, while there are a number of interpretations of "enhancement" available to policymakers, the ones that most accurately identify the public policy interests at stake in human gene transfer research are those that focus on the uses of gene transfer that would serve to exacerbate, rather than reform, the social injustices that flow from our intolerance for human biological variation.

Section 14: Genetic Variation, Race and Culture

When We Discuss Genetics, Our Words Have Different Meanings: Examination of African-American and Caucasian Adults' Understanding of Genetics and Related Implications

E.M. Petty, S.R. Kardia, R. Mahalingham, C.A. Pfeffer, S.L. Saksewski, M.G. Brandt, E.S. Anderson, T.E. Jayaratne

The accelerated pace of genetic discovery has provided novel insights into mechanisms of health and disease. Increased human genome knowledge will impact most Americans' lives within this decade. As part of an ELSI project examining beliefs about individual and group differences, we examined how Americans used genetic terms and understood related genetic concepts. We hypothesized that usage and understanding would vary among individuals. National qualitative telephone interviews of African-American and Caucasian adults included concluding questions specifically assessing genetic knowledge. Many respondents used "genes" and "genetics" when discussing the etiologies of some traits. Most knew that genetic information is heritable and that genes are important to human development. The majority, however, could not accurately explain basic genetic terms/concepts when directly asked What do you mean when you say something is genetic? and Where are genes located? Some became frustrated: I don't know-I feel so stupid right now. Others simply provided examples: ... take Grant Hill ... his daddy ... played football ... son plays basketball, non-scientific descriptions — it's ... where the stars are situated ... people born at different times ... have different attributes, and/or inaccurate answers — genes are in ... cerebrum or the cerebellum? It's one of the two. To some, "genetic" referred to an "innate potential" that could be activated, like a latent virus. Nearly 50 percent thought genes were largely, or only, in the brain/mind. Most did not, however, feel that genes are the primary determinants of intellectual function. Athleticism and sexual preference were noted by some to have significant genetic contributions. Genetic terms are used frequently in America today (e.g., in clinics, classrooms, advertisements, televised sports, synagogues, sitcoms). Our results demonstrate that they mean very different things to different people. Knowledge about genetic concepts also varies among individuals. More accurate use of genetic terms by teachers, health care providers, journalists, policymakers, and others is needed to foster genetic literacy. Public genetic education efforts should be explored. Perpetuation of genetic misconceptions will diminish one's ability to make informed choices about heath care, reproductive options, and public policy as related to genetic research, genetic testing, gene cloning, gene therapy, and genetic discrimination. Results from our study and their implications will be further discussed.

GeneSurvey: African-American College Students Respond to the New Genetics

Fatimah L.C. Jackson

A revolution is occurring in the identification, interpretation, and application of human DNA sequence variation data. However, very little is known about how this research will influence diverse cultural and ethnic groups, interact with current perspectives on ethnic identity, or influence their receptivity to applied genetic and genomic knowledge. We are in the first year of a 3-year research project that examines African-American college students' responses to the new Human Genome Project initiative to study human genetic diversity. The aims and methods of the project are (1) to develop and pre-test a culturally appropriate online questionnaire (using focus groups, a validation study, a scientific advisory board, and a unique fact-based question set format), (2) to recruit to retain 2,200 respondents (1,100 men and 1,100 women) currently enrolled in 1 of 15 colleges and universities and between the ages of 18 and 30 years (using trained local student leaders to facilitate recruitment), and (3) to analyze response data to test 10 project hypotheses. These project hypotheses concern three broad areas of concern: (1) the genetic and genomic research preferences of African-American college students, (2) the anticipated effects of genetic variation data on ethnic identity, and (3) the potential degree to which molecular genetic data will be integrated into existing cultural and ethical paradigms. An online summary of the survey results at the project Web site (www.ourgenesurvey.com) and a discussion forum (listserv) will be activated during the last 16 weeks of the study to allow respondents to interactively discuss

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the survey's conclusions. We have completed the first stages of the development of a culturally appropriate and scientifically valid questionnaire and are placing it online for preliminary testing.

What We Don't Know About the Social and Cultural Implications of Human Genetic Variation and How to Ask

Morris W. Foster

Ethnography can be characterized as a process of first learning what we do not know about a topic and then learning how to ask appropriate questions to find the answers. We are now in the midst of identifying what we do not know about community-specific social and cultural implications of human genetic variation, and we are beginning to develop appropriate ways of finding those answers. Using research experiences with Native-American communities as examples, this presentation will review some of what we have learned so far about what we do not know as well as about how to ask questions to fill those gaps in our knowledge.

Ethical Conduct of Genetic Research in Populations

Bartha M. Knoppers

Contrary to the fields of genetic testing or research involving individuals and families where ethical norms abound, other than population screening, few if any normative frameworks are specific to research involving populations or subpopulations. In some situations, epidemiological guidelines may be applicable, but often the guidelines are oriented to anthropological, demographic, or, more recently, to surveillance studies (e.g., HIV). Considering the current interest in SNP mapping of populations for pharmacogenomic studies on genetic susceptibility (i.e., metabolic pathways, polymorphic variations) that at this early stage have little to do with highly penetrant disease alleles, different normative frameworks need to be developed for population genetics. Current applications of different models need to be critically assessed, and new approaches will be proposed.

Beliefs About the Genetic Origins of Gender, Class, and Race Differences

Toby Epstein Jayaratne, Elizabeth Anderson

Although recent advances in human genetics have inspired hopes for the prevention of many diseases, this research also generates enormous controversy. This controversy stems primarily from concern that acceptance of genetic theories may reinforce discriminatory beliefs about groups perceived as genetically inferior. However, little empirical research has been conducted investigating the public's use of genetic explanations. This study explores two relevant questions:

1. What is the character of the public's beliefs about genetic causes for individual and perceived gender, class, and race differences?
2. Are these beliefs related to resentment toward disadvantaged groups?

Telephone interviews were conducted with 174 African-American and white, female and male respondents residing in southeastern Michigan. A series of questions assessed the respondents' genetic attributions for individual and gender, class, and race differences in intelligence and the willingness to work hard, and their resentment toward women, the poor, and African-Americans.

Results indicate that women were more likely than men to believe individual differences in hard work are genetic; whites were more likely than African-Americans to believe individual differences in intelligence are genetic. Approximately 13 to 45 percent of respondents attribute perceived group differences to genetic causes, the percentage varying by the target group as well as the race and gender of the respondent. Overall, respondents who posited genetic attributions for individual differences were more likely than others to use genetic attributions for perceived gender, class, and race differences. In addition, among white men, genetic explanations for perceived class and race differences are related to greater resentment toward the poor and African-Americans; among African-American men, genetic attributions for perceived gender differences were related to greater resentment toward women. African-American women who used genetic attributions for perceived class differences in intelligence were less likely than other African-American women to indicate resentment toward the poor.

These findings are interpreted and discussed in the context of current sociopolitical agendas and in light of what is known about genetic mechanisms. Most importantly, we suggest that the connection between genetic explanation and resentment is due primarily to the belief that disadvantaged groups receive undeserved opportunities, undeserved because of the perceived inferior and nonmalleable (genetic) character of their intelligence or willingness to work hard.

Lay Understandings of Race and Genetics

Celeste M. Condit, Roxanne L. Parrott, Tina M. Harris

Members of the lay public assembled in 16 focus groups articulated their understanding of the relationships between genes and race. Participants pooled their individual knowledge to generate a collectively articulated account of race and genetics that held that racial characteristics attributable to genes are relatively minor and primarily associated with visible physical traits (e.g., skin color, hair texture, body build). The lay accounts more closely paralleled the accounts of contemporary evolutionary human population genetics rather than older essentialist notions of biological race. These relatively typical members of the public explicitly incorporated into their accounts the concepts of evolutionary adaptation, variable penetrance, dominance, shared human origins, and the impact of migration and mating patterns. They did not explicitly articulate the concepts of genetic drift or founder effects, nor were their accounts precise and detailed. The race by gender groupings (four groups of each) employed different means for accounting for difference and similarity and highlighted different physical features. African-American males emphasized body build and athleticism and highlighted the coexistence of similarity and difference; African-American females emphasized skin and hair and utilized the notion of racial blending; European-American males denied difference and emphasized sameness, while European-American females focused on illness. These results suggest that members of the public are able to use their schooling, media information, and common sense to craft sophisticated accounts of the relationship between race and genetics. These results have implications for educational efforts.

Pharmacogenomics and Minority Populations

Mark A. Rothstein, JD

By predicting which drug will best interact with an individual's genetic profile, pharmacogenomics promises individually matched drugs with minimum side effects and maximum efficacy. Because a significant, but as yet unknown, percentage of genetic variations of pharmacological significance may occur with greater or less frequency in different racial or ethnic groups, pharmacogenomics is likely to raise a variety of ethical and legal issues at every stage of drug development, from basic research through clinical trials to clinical care. Resolution of the concerns requires consideration of how genetic variation research can be conducted in an ethically and culturally sensitive manner, how the concepts of race and ethnicity will shape the development and acceptance of the research, how cultural and other factors will impact the results of the research, and how racial and ethnic groups will be afforded access to research benefits.

The need to conduct pharmacogenomic-based drug trials on particular racial or ethnic groups illustrates how these considerations come into play. In the course of designing a trial, researchers need to ensure that the purpose of the trial is effectively communicated to the participants so the participants can give their informed consent. Language barriers and the complexity of genetic research may make this difficult. The concepts of race and ethnicity can shape how the trial is conducted because the researchers and the participants may define them differently. The trial must be adjusted to respect these differences. Cultural differences may influence the participants' receptiveness to Western medicine or expectations about the proposed treatment and thereby influence the trial results. The ability to recruit certain racial or ethnic groups may depend upon the likelihood that members of the group will gain access to the drug, if approved.

While the ultimate success of pharmacogenomics requires that these important ethical, legal, and social concerns be addressed, little formal consideration has been given to the subject until now. Currently, pharmacogenomics is typically discussed in terms of its potential scientific or business promise. Additionally, current Federal and State laws do not address the unique ethical and legal issues that pharmacogenomics presents, including issues of informed consent, confidentiality and minority recruitment in pharmacogenomic-based drug trials, access to and mandated coverage of pharmacogenomic-prescribed drugs, and appropriate legal and ethical standards of care for physicians and pharmacists in connection with pharmacogenomic-prescribed drugs.

Engaging Diverse Communities in Genetics Policy Dialogues

Toby Citrin, JD, Leonard M. Fleck, PhD, Marian Secundy, PhD

Two successive ELSI-sponsored projects have utilized dialogue methodology to engage communities in learning about genetics and developing recommendations for policies to guide the application of genetics research. The first project, Genome Technology and Reproduction: Values and Public Policy, engaged 7 groups in Michigan, while the second project, Communities of Color and Genetics Policy, engaged 15 communities in Michigan and Alabama, all of which were made up of African-American or Latino members. The dialogue groups in both projects met for consecutive weekly sessions considering issues identified by earlier focus groups whose members were drawn from the same communities. The methodology used in organiz-

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ing and conducting these dialogues was significantly modified in the second project, drawing upon the experience of academic-community partnerships involving multicultural groups.

The combined experience of these two projects suggests a new methodology for eliciting the engagement of diverse communities in considering genetics issues and recommending policies to address them. The characteristics of this methodology include (1) partnerships between academic institutions and community-based organizations, (2) the role of community-based organizations in hosting community dialogues, (3) the role of community-based organization leadership as part of the project team designing dialogue characteristics, selecting and writing educational materials, writing project reports, and disseminating results to policymakers, (4) matching facilitators and observers with the race and ethnicity of dialogue group participants, (5) flexibility in the issues agenda, and (6) flexibility in the manner of dialogue so long as essential elements (e.g., mutual respect, considered judgments, and giving reasons for opinions) are maintained.

The modified methodology in the second project not only was successful in achieving community engagement but also resulted in the desire for further education and the commitment of community members to carry out joint advocacy for genetics policies beyond the formal end of the project. This success suggests that replication of the model can result in a significant increase in community engagement on genetics policies as well as other issues of health policy in which sound policy decisions require value judgments from the public.

Section 15: Posters and Multi-Media Presentations

The Human Genome Management Information System (HGMIS): Making Genome Project Science and Implications Accessible

Anne Adamson, Denise Casey, Betty Mansfield, Sheryl Martin, Marissa Mills, Judy Wyrick, Laura Yust

Since 1989, HGMIS has been funded by the Department of Energy Human Genome Program (HGP) to make information on U.S. Human Genome Project goals, progress, science, applications, and societal implications accessible to many audiences. These audiences include interdisciplinary investigators who may want to get up to speed quickly on genome science and participate in project strategy development or research, those writing proposals, and those already contributing to the project or using its data and resources.

Our work also serves educators, students, medical professionals, bioethicists, lawyers, policymakers, information multipliers such as the media and museum exhibit developers, and the general public who need an understanding of the new genetics and its implications. Such understanding is vital in communicating, teaching, and helping others make related career and personal decisions. One of our overarching goals is to democratize access to genetic science and its societal implications; achieving this goal will increase genetic literacy and help maximize HGP benefits while protecting against potential misuse of personal genetic data. The hope is that more informed personal and public policy decisions can be made concerning applications of genetics and genetic data.

To satisfy the information requirements of various audiences, HGMIS produces print and Web-based information resources and exhibits; makes presentations at legal, medical, student, and minority meetings; and answers e-mail and phone requests from those needing specialized attention. Requesters include meeting organizers, museum content developers, other science agency program managers, media, students, and family members of those with severe genetic conditions. Our primary products include *Human Genome News* (*HGN*), genome primer material, progress reports, factsheets, images, and invited articles in peer-reviewed publications. *HGN*, with a distribution of more than 14,000 subscribers, is written for educators and more technical audiences. It contains a unique compilation of information and resources not found in any single discipline-specific publication. Subscription requests to the print version of *HGN* averaged nearly 200 a month for most months in fiscal year 2000.

The Human Genome Project Information (HGPI) suite of Web sites, initiated in 1994, supports some 200,000 unique user sessions and more than 750,000 text-file transfers each month (there are nearly 6 million graphic+text file hits per month). Over 7,000 external Web pages link to HGPI's more than 3,000 files. HGPI, whose content is driven by users, is a primary resource for most of our audiences, which include news outlets and documentary producers.

Although HGPI Web pages are presented from HGP's perspective, they extend well beyond the project's stated goals to cover the downstream science and societal ramifications of various genome science applications. The convergence of the Internet and the excitement generated by the prospect of elucidating the blueprint of life on this planet offer an unprecedented opportunity to gain a more biology-literate public. Therefore, we are expanding our Web pages to cover postgenomic-era research and applications in an attempt to hold and build upon public imagination captured by the HGP in a way not seen since the moon landing and the early NASA Challenger space flights.

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Development of Genetic Thesaurus Terms for BIOETHICSLINE

Doris M. Goldstein

As research into the ethical, social, and legal aspects of genome mapping and genetic medicine has become more nuanced over the decade, so also have the indexing terms developed to facilitate access to this research through BIOETHICSLINE. When BIOETHICSLINE first went online in 1974, Gene Pool, Genetic Counseling, Genetic Disorders, Genetic Intervention, Genetic Screening, Genetics, and Germ Cells were the basic terms used to index documents discussing genetic issues. Subject access to the concepts found in the documents was guaranteed by using combinations of these terms. A BIOETHICSLINE thesaurus was produced to list the terms and their definitions, and this thesaurus enabled users (originally librarians who had access to the National Library of Medicine's (NLM) databases, and then health professionals and the general public using NLM's Grateful Med interface) to search independently. In 1989 the term Genome Mapping was introduced followed by Gene Therapy in 1980. By 1995, the terms Genetic Predisposition and Genetic Research were needed. Genetic Enhancement, which was added in 1996, was followed by Genetic Information, Genetic Materials, and Genetic Services in 1997, and by Genetic Determinism and Genetic Identity in 1998. New terms in 1999 prompted by developments in ELSI research address issues related to concepts (such as Commodification) and services (such as Registries). As NLM consolidates all its existing databases into one, maintaining differentiation in searching through the use of BIOETHICSLINE thesaurus terms will be essential for continued access to ELSI research.

GeneTests and GeneClinics: Two Online Genetic Information Databases That Help Integrate Genetic Information Into Clinical Settings

Roberta A. Pagon, MD, Peter Tarczy-Hornoch, MD, Maxine L. Covington, Patricia K. Baskin, MS, Joe Edwards, PhD, Bradley Popovich, MS, PhD, Thomas Bird, MD, Cynthia Dolan, MS, Nancy Hanson, MS

GeneTests (www.genetests.org), established in 1992, and GeneClinics (www.geneclinics.org), established in 1997, provide current information on genetic services, including the availability and use of genetic testing and genetic counseling. GeneTests contains a directory of medical genetics laboratories, a directory of genetic and prenatal diagnosis clinics, educational materials to place genetic counseling and testing in context of patient care, teaching tools for genetics professionals, and summary reports of genetic test availability. GeneTests lists over 440 laboratories offering testing for more than 770 diseases and more than 900 U.S. genetics clinics. A PowerPoint module of over 70 slides is available to genetics professionals for instructing other health care professionals.

GeneClinics provides expert-authored, peer-reviewed disease-specific information on the use of genetic testing in patient diagnosis, management, and counseling. Each entry is highly structured for storage in a database. For each disorder, GeneClinics links to patient-specific resources, policy statements and guidelines for testing, citations in the biomedical literature, and genomic databases. About 100 entries are currently posted, with one new entry being added each week. Entries will be updated annually.

As of October 2000, GeneTests was receiving about 600 to 700 inquiries per day and GeneClinics over 1,800 per day. Both GeneTests and GeneClinics are targeted to health care professionals but are easily accessible to the public. Both resources are increasingly used in undergraduate medical education and have been identified as significant resources for primary care physicians in the Genetics in Primary Care project.

Using the Database HUMGEN To Discover Policies in Human Genetics

Denise M. Avard, Bartha M. Knoppers

Issues in human genetics reach beyond one sector, one discipline, one country, and one specific area of activity. Human genetics is a highly interdisciplinary and collaborative field, involving geneticists, molecular biologists, epidemiologists, clinicians, sociologists, jurists, psychologists, and ethicists working together. Given the rapid expansion in the field of human genetics, policymakers, medical practitioners, and the public are struggling to keep informed and to come to grips with a host of complex ethical, legal, and social questions. To this end, guidelines and policy documents are being continuously developed worldwide on legal, social and ethical aspects of human genetics.

We have developed a compendium of policy documents limited to professional guidelines, ethical codes, recommendations, and legislation dealing with topics such as human rights (consent, professional ethics, children and incompetent adults, privacy and confidentiality, discrimination, employment, and insurance), biomedical issues (DNA banking, gene therapy, cloning, genetic screening, genetic testing, genetic research, and population genetics), and bio-commerce (intellectual property, patenting) geared for researchers, policymakers, and the public. All these documents are stored in a data base, HUMGEN, developed in the Law Faculty of the University of Montreal.

To facilitate public access to policy documentation, the HUMGEN policy database is freely accessible via the HUMGEN Web site (http://www.humgen.umontreal.ca). Several strategies are used to locate documents, via databases searches, literature reviews, and a list of contacts with major people and institutions in the field. Information is grouped according to international, regional, and national jurisdictions, and over 30 countries are represented in the database. Wherever possible, direct HTML links are provided to the online text of the document.

Because of the Internet, HUMGEN provides an ideal mechanism for rapidly and inexpensively disseminating policy statements to a very wide and varied audience. This easy access to human genetic policies, from all over the world, will give valuable information to researchers, policymakers, and industrialists alike about the political landscape and can serve to assist in formulating ethical guidelines as well as promote international harmonization of the policies.

Layered Access to ELSI Literature Through the National Information Resource on Ethics and Human Genetics

Doris M. Goldstein

As Internet surfing becomes commonplace and Web sources multiply exponentially, the need to structure Web retrieval becomes essential. Basic concepts and content are lost to Web browser algorithms based on the number of links to a page or the repetition of specific terms. Narrative overviews of topics presenting multiple viewpoints are rare as organization and company home pages trumpet their versions of issues but produce their pages with the aura of neutrality. In a global information environment where most peer-reviewed journals are fee-based, National Information Resource on Ethics and Human Genetics is designed to give searchers the good information they want at the level of detail that they need. There are three components: online overviews, database searching, and full-text documents. The online overviews consist of annotated bibliographies (Scope Notes) on genome mapping, gene therapy, genetic screening and counseling, and eugenics, each spanning many years of the literature. These are prefaced by one-page E-Notes, synopses of the current state of each topic. The Scope Notes and E-Notes orient searchers to the literature on their topics, which then can be further explored by searching BIOETHICSLINE and the Ethics and Genetics Database.

Digital Image Archive on the American Eugenics Movement

David Micklos, Susan Lauter, Jan Witkowski

The Eugenics Archive, an NIH ELSI project, provides students, teachers, scholars, and the interested public with an extraordinary window into a hidden chapter of history. We hope that the opportunity to revisit this period will stimulate people to think critically about our current involvement in human genetics. By providing access to the eugenicists' own words and data, we hope to challenge visitors to assume the role of historian/researcher. By focusing primarily on visual documents, we hope to engage young people and others who would not normally access a scholarly collection.

The site contains more than 1,500 images drawn from six major scholarly archives, for most of which this project marks their first large-scale release of items via the Internet by a second party. We developed guidelines for online publication, educational fair use of documents, and privacy protections that can guide other projects dealing with the release of sensitive documents via the Internet. These policies were developed by consensus during 6 days of workshop sessions by a 15-member advisory panel representing the viewpoints of human genetics, history of science, cultural anthropology, public education, minorities, and the disabled community.

The Eugenics Archive makes no attempt to lead users to a correct interpretation of the materials. However, the site assists users in understanding the historical, social, political, and ethical context in which the American eugenics movement developed, flourished, and finally collapsed. Context is built into the Archive on two levels. First, users are encouraged to enter the site through a series of virtual exhibits, which introduce the key events, persons, and social conditions that contributed to the development of eugenics. Second, all images are sorted into more than 20 topic areas. Browsing by topic or searching by keyword returns a set of related images with extended captions. The topic captions are designed to help the user understand relationships among images and the relationship of the image to the eugenics movement and society. Both levels were developed in collaboration with several leading historians of eugenics. At each level, users are reminded that the vast majority of scientific facts presented by eugenicists were fundamentally flawed and have been discredited by modern research standards.

Opinions and Attitudes Toward Possible Future Genetic Testing for Prostate Cancer: A Qualitative Approach

Nedal Arar, Ian Thompson, Rosemarie Plaetke

Recent research links hereditary prostate cancer (PC) to a region on chromosomes 1 and X. Gene testing (GT) can be performed to determine mutation carriers and noncarriers within PC families. A qualitative study was conducted to investigate PC patients and relatives on (1) opinions concerning possible GT for PC, (2) awareness of ethical issues surrounding GT, and (3) attitudes towards performance of GT for PC. Participants were recruited from 25 families enrolled in a PC family study conducted at the University of Texas Health Science Center at San Antonio. Each family averaged 4.0 PC cases (range: 3-9). Applying ethnographic techniques of open-ended questions, we performed 21 semistructured interviews with 12 patients and 9 relatives. Interviews were tape recorded and transcribed.

Subjects were on average 63 years old. Sixty-six percent were European-American and 33 percent African-American; 81 percent had at least a college degree and 64 percent were retired; 9 percent had heard about GT for hereditary cancer and almost 86 percent did not know the definition of GT. However, after being informed about GT, participants indicated that GT is an important advancement, and relatives were willing to have GT for PC done if available.

Patients indicated that they would share test results with family members. Sixty-six percent of subjects agreed that GT should be offered for everyone: (1) those who wished to learn about their risk of developing PC and (2) infants. Twenty-nine percent proposed being tested before marriage while 38 percent believed testing should occur before having children. Nine percent suggested testing be performed on individuals with a positive family history of PC. The most common reason for undergoing GT was for preventive measures, while issues related to expenses and confidentiality of test results were mentioned as reasons for declining GT.

Participants developed a strong positive attitude towards possible GT for PC. This attitude did not change even after discussing ethical dilemmas surrounding GT. Participants considered GT to be more beneficial than harmful, indicating that positive results should be confidential and insurance companies should cover the expenses.

This research was funded by NIH HG00191/ ELSI and the San Antonio Area Foundation (SAAF). The ascertainment of PC families was funded by grants from SAAF and NCI (CA 70183).

Partnership for Genetic Services Pilot Program: Provider-in-Training Education in Genetics

Mary Davidson, Vicky Whittemore, Bowie Little, Nachama Wilker, Ann Smith, Nisha Isaac, Cindy Holmes

The goal of the Partnership for Genetic Services Pilot Program is to provide the information, resources, and tools that health care providers need to deliver quality genetic services. The providers-in-training component of this program used consumer educators (individuals with genetic conditions and/or their families) in the education process to highlight the ethical, legal, and social implications of genetics.

Partnership staff worked directly with faculty sponsors in six medical schools and one genetic counseling program to incorporate consumer educators in the educational process during the 1998-1999 and 1999-2000 academic years. They identified and trained consumer educators and coordinated their presentations. Genetics networks in the Great Lakes and Mid-Atlantic regions collaborated in the training sessions and presentations. The goal of each presentation was to foster a better understanding of the role of the health care provider in the delivery of family-centered, culturally competent, and consumer-informed genetics services. Evaluations completed by consumers, faculty, and students documented the effectiveness of the consumer presentations.

Approximately 1,020 medical students at Indiana University, Southern Illinois University, University of Chicago, University of Wisconsin, and Wright State University, and genetic counseling students at Howard University attended consumer educator presentations. The majority of the students and faculty indicated that the consumer presentations were very useful and that the consumer educators were very prepared for their presentations. Personal and real-life experiences shared by the consumers about the impact of a genetic condition on the family were the most useful part of the presentations. Information on the genetic condition, resources, and lay advocacy organizations were also very useful. Detailed summaries of the evaluations will be described.

The use of trained consumer educators in provider-in-training education is a very effective way to communicate the impact of a genetic condition on the individual and his or her family. The providers-in-training gain an important perspective on the ethical, legal, and social implications of a genetic diagnosis on the lives of those affected.

Funded by the National Human Genome Research Institute's Ethical, Legal, and Social Implications Program, and the Genetics Services Branch of the Maternal and Child Health Bureau (Health Resources and Services Administration).

Educational and Advocacy Behaviors of Genetic Counselors During an Ethical Situation

Marcia Sue DeWolf Bosek, DNSc, RN

Rapidly increasing genetic testing abilities and genetic knowledge are creating new ethical situations for genetic counselors. However, little is known about the type and frequency of ethical situations experienced by genetic counselors during their clinical practice or how genetic counselors resolve ethical situations in the clinical setting. Therefore, the purposes of this study were to (a) identify the types of ethical situations experienced by genetic counselors and (b) describe and analyze the process used by genetic counselors when they are resolving an ethical situation.

A grounded theory method was implemented. Twelve Caucasian genetic counselors (8 registered nurses, 1 master's prepared counselor, and 3 physicians; 2 males and 10 females) were self-selected from a population of genetic counselors employed in a Midwest urban genetics department. The subjects had a mean age of 43.6 years and on average had been employed in genetic counseling for over 9 years. Educationally, four subjects held baccalaureate degrees (three were currently enrolled in master's programs), five were master's prepared, and three were M.D. and/or Ph.D. educated; 92 percent of the subjects had participated in an ethics in-service program, but only one third had taken a college ethics course.

Each subject participated in a semi-structured, audio-taped interview. The genetic counselors described experiencing a variety of ethical situations, including testing minors, paternity questions, how to disclose research findings to individual family members, prenatal testing (including abortion issues), autonomy, maintaining confidentiality, financing genetic testing, Huntington disease protocols, diagnosing without cure or treatment, and aggressiveness of treatment for neonates with genetic defects. Fifty-eight percent of the subjects indicated they made ethical decisions on a daily basis, while the remaining subjects believed they made ethical decisions on at least a weekly basis.

A conceptual model will be presented that describes the genetic counselors' perception of the ethical situation as well as the variables and philosophies influencing the resolution of the ethical situation. The Basic Social Structural Process describing the environment in which ethical decision-making by genetic counselors occurs is patient education. Patient autonomy is the Basic Social Psychological Process guiding the resolution of ethical situations.

Emerging Ethical Issues in Pharmacogenomics

Carol L. Freund, Benjamin S. Wilfond

Pharmacogenomics aims to improve the safety and efficacy of drugs. At the intersection of drug development and genetic testing, pharmacogenomics raises the ethical issues associated with each of these areas. This analysis will consider issues related to drug development, clinical trials, test development, and clinical applications.

Drug Development: The correlation of drug response and polymorphic genetic markers will require collecting genetic information and medical records from numerous people. The complexity of acquiring and interpreting this epidemiological data is likely to be a rate-limiting step for this technology. Pharmacogenomic investigation brings a novelty of scale to considerations of human subjects protection, including the use of human biological material, privacy, confidentiality, and informed consent.

Clinical Trials: Pharmacogenomic data may impact the conduct and interpretation of clinical trials. Genetic markers associated with increased risk of an adverse reaction may result in the exclusion of high-risk subjects. Selection of genotypes could influence drug labeling and create orphan genetic subgroups. Expedited clinical trials may require significant post-market research to evaluate untested subgroups. Such research will require attention to human subjects protections.

Test Development: Marker selection and test format raise important issues. Genetic markers associated with drug response may also convey additional medical information, as either predictive or prognostic factors, or may convey information about behavioral traits. Some markers may vary in prevalence in social groups, creating further potential for group stigmatization. These issues may be exacerbated if, rather than testing only for a limited number of markers, tests are done in a format that allows for a very large number of markers. Further, such multi-gene testing will raise further issues if this is done in newborns or children.

Clinical Application: The integration of pharmacogenomic information into clinical practice will be complex for both providers and patients. Guidance will be needed for the interpretation of relative and absolute risk associated with pharmacogenomic markers and the implications of such tests for clinical decisions. It is possible that drug-labeling will recommend that genetic tests are bundled with drugs. However, some patients may have genotypes not identified during the trials or some may decline testing.

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The Sense and Reference of Surveillance: How Much More Than Watching and Waiting?

Ellen Giarelli, EdD, RN, CS

Purpose: To analyze the concept of individual surveillance, with special attention to potential utility in individualized genetic cancer risk management.

Sources: Literature was reviewed from a range of historical and contemporary primary and secondary sources, including the fields of military intelligence, criminal justice, physical and biological sciences, technology, epidemiology, public health, and genetic counseling.

Method: Epistemological propositions from Gottlob Frege's *Sense and Reference* informed the concept of "individual surveillance." A Heideggerian philosophical perspective provided the frame for interpretation, application to practice, and the construction of a research paradigm.

Findings: Individual surveillance is a complex concept that has cognitive content and is situated in social context. It is used across social institutions, but it may have attribute predilection, or fore-structure, such that criterial attributes are present in advance of participation. The fore-structure is created by personal experience and the context of social relations. Variability in meaning may confound the potential benefits of its use in genetics cancer care. The historical association between surveillance and oppression may infuse the contemporary link between surveillance and genetic cancer risk management. A phenomenological research paradigm can guide the development of research questions that aim to discover the nature of the experience of surveillance to participants and secure potential benefits. The analysis is part of a larger Grounded Theory investigation of individual surveillance and illustrates a formal process of "bracketing," when preconceived ideas are identified and set aside. There is a need to study the meaning of individual surveillance as it exists as a component of lifelong follow-up care for hereditary cancer risk management.

An Exploratory, Qualitative Study of Prenatal Diagnosis Decision-Making

R. Kenen, A.C.M. Smith, C. Watkins, C. Zuber-Pittore

Aims: The aims of the study were to investigate (1) male partners' decision-making regarding the use of prenatal diagnosis, (2) factors influencing the use of genetic counseling and prenatal genetic testing for two groups: pregnant women ages 35 years of age and older (AMA) at the time of delivery and pregnant women with an abnormal maternal serum triple screen.

Methods: We conducted an IRB-reviewed exploratory, qualitative study at four genetic counseling clinics in the Eastern United States from 1995 to 1997. The convenience sample consisted of 25 semi-structured interviews and 50 observations of genetic counseling sessions with middle and working class women/couples. The Interview Guide included questions about the value of knowledge, risk, termination of pregnancy, genetic counseling and prenatal technologies, how decisions were made, and how disagreements were resolved.

Results: We identified three decision-making styles used by the males and their partners: (1) domain, (2) joint-delegated, and (3) saliency. The male partners also seemed to view prenatal diagnosis as either an information decision or an action decision. Men appeared to take a more active role in decision-making when the decision was viewed as an action decision. Worry was the most important variable influencing decision-making about prenatal diagnosis and was greater in the MSAFP3 group than in the AMA group. The women in the AMA group appeared to assign the risk of having a child with Down syndrome to their age category rather than to them individually. The risk perception for women with an abnormal MSAFP3, however, appeared to have shifted from a general population risk for pregnant women to an individual, personal risk. There was a general lack of understanding and also more misinformation about the MSAFP3 screen compared with amniocentesis. Women in both groups were torn between fear of an invasive test and worry about the health of the fetus.

Discussion: Further study is warranted into both the prenatal diagnosis/worry conundrum as an iatrogenic consequence of technological pregnancies and the relationship between the information and action perspectives of men's decision-making and their decision-making styles.

Knowledge, Attitudes, and Interest in Breast-Ovarian Cancer Gene Testing: A Survey of a Large African-American Kindred with a BRCA1 Mutation

Anita Yeomans Kinney, PhD, APRN, Robert T. Croyle, PhD, Christine A. Bailey, BSN, RN, Mary Kay Pelias, PhD, JD, Susan L. Neuhausen, PhD

Background: This study assessed counseling and testing needs from the perspective of adult members of a large African-American kindred (K2099) with a BRCA1 mutation.

Methods: Interviews were conducted with 95 male and female adult kindred members to elicit information on

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sociodemographics, attitudes toward health care providers, breast cancer screening behaviors, informational needs, and religious/spiritual beliefs and to evaluate psychological distress, beliefs, knowledge, and attitudes related to genetic testing.

Results: Knowledge about breast and ovarian cancer genetics was limited. Adherence to screening recommendations was low among females with no breast or ovarian cancer history. The majority (67 percent) wished to discuss cancer risk factors with a care provider. Most participants (82 percent) indicated they would have a genetic test if it were available. Significant predictors of intent to undergo genetic testing were younger age (OR=0.6; 95 percent CI=0.4-0.9), perceived risk of being a gene carrier >50 percent (OR=17.5; 95 percent CI=1.6-186.1), and presence of depressive symptoms (OR=5.3; 95 percent CI=1.1-26.3). Participants tended to minimize the risks and limitations of testing. Commonly cited barriers to testing included cost and availability.

Conclusion: There is a high interest level in genetic testing despite limited knowledge about cancer genetics among these high-risk African-Americans. Our study provides information for designing a culturally sensitive genetic education and counseling intervention for this and similar families. An ongoing prospective observational study of K2099 will address a critical gap in the literature regarding cancer susceptibility testing by examining utilization of genetic counseling and testing and behavioral and psychosocial responses to the receipt of genetic information among African-Americans.

Preferences of Women Facing a Prenatal Diagnostic Choice: Long-Term Outcomes Matter Most

Miriam Kuppermann, David Feeny, Elena Gates, Samuel F. Posner, Bruce Blumberg, A. Eugene Washington

Background: Women ages 35 or older who wish to undergo prenatal diagnosis for chromosomal disorders are typically offered a choice between chorionic villus sampling and amniocentesis. These two tests are performed at different times during pregnancy and impose different miscarriage risks. In deciding which test to use, therefore, women need to consider both short-term consequences (e.g., timing of news of either an unaffected or an affected fetus, or timing of a pregnancy loss, should it occur) and long-term consequences (e.g., whether a pregnancy loss is followed by a future birth).

Objective: We examined how women receiving prenatal diagnostic services value the outcomes of testing, with special emphasis on potential differences in the importance of short- and long-term outcomes.

Methods: We conducted a cross-sectional study of 72 women obtaining genetic counseling at the University of California at San Francisco or Kaiser San Francisco. We elicited preferences for various outcomes of prenatal diagnosis, which outcomes differed in both their short- and long-term consequences. Preferences were measured as utilities using the standard gamble metric. We also assessed demographics and attitudes toward Down syndrome and other disabilities, abortion, spontaneous abortion, and future births.

Results: Utilities for first and second trimester pregnancy losses with similar long-term sequelae did not differ ($p<.05$ for all comparisons, paired t-test). Utilities for losses followed by future birth, however, were significantly higher than utilities for losses without future birth (range, 0.91 to 0.93 versus 0.84 to 0.86; $p<.05$ for all comparisons, paired t-test). In addition, we observed substantial variation in utilities across women; this variation was unrelated to age.

Conclusions: Long-term outcomes matter most to these women. In presenting prenatal diagnostic options to their patients, clinicians should include discussion of outcomes such as the likelihood of a future birth in the event of a pregnancy loss. Furthermore, the substantial variation in utilities we observed suggests that future prenatal testing policies account for the preferences of the individual woman.

Genetics Education for Health Professionals: What Works, What Doesn't Work, and Why

E. Virginia Lapham, PhD, Chahira Kozma, MD, Joan O. Weiss, MSW, Judith L. Benkendorf, MS, CGC, Mary Ann Wilson, BA

A three-pronged approach to genetics education of health professionals was carried out for 3 years (1997-2000) as part of the Human Genome Education Model (HuGEM) Project of Georgetown University and the Genetic Alliance. Preliminary analysis of effectiveness shows strengths and weaknesses of each approach. The seven collaborating associations included AOTA (occupational therapists), ADA (dietitians), APTA (physical therapists), APA (psychologists), ASHA (speech-language-hearing therapists), and CSWE and NASW (social workers). Approaches included orientation, intensive 5-day courses, and training workshops.

Orientation sessions were held with 130 national staff and board members of the associations to acquaint them with HuGEM, the Human Genome Project, and ELSI and implications of genetics for their professions. This was crucial to the success of the intensive courses and workshops. The professional culture, commitment, interaction with representatives, and other endorsements influenced the timing and extent to which each organization participated in the project and made progress toward integrating genetics into practice and policies.

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Follow-up activities of 60 health professional leaders selected by the associations to participate in 5-day intensive genetics courses included initiating continuing education courses in genetics, incorporating genetics in curricula, writing articles for newsletters and journals, developing a correspondence course in genetics, and developing practice guidelines and influencing policies of organizations. The extent of follow-up depended upon the initial method of selection for the course, organization expectations and opportunities, and technical assistance.

Training workshops of 2-8 hours were held at 36 national and regional conferences of the associations for over 1,200 professionals. Female attendees considered themselves more knowledgeable about genetics than males. Overall, 40 percent considered themselves to know a lot or a fair amount prior to the workshops, with attendees in their forties and fifties more knowledgeable than other age groups. There was a significant increase in how important the attendees considered genetic issues to be to society and to their professions after the training. About one-third signed up to do training of colleagues, students, or clients in their communities. More educators than practitioners followed through on training. Practitioners did not follow through because supervisors did not allow time, their colleagues were not interested, and they lacked confidence or forgot, and because of personal crises.

Conclusion: The orientation sessions for national staff and boards and the intensive courses for leaders selected by the associations were more effective than the training workshops. Follow-up communication including electronic, technical assistance including partnering, and materials and resources were important to the success of all three approaches.

A Statewide Genetics Continuing Education Program for Public Health Nurses

Dale Halsey Lea, RN, MPH, Ellie Mulcahy, RNC, Valerie Ricker, MSN, PNP, WHNP, Jane Congelton, RN, MS, CGC, Rhonda P. Spiro, MD

The goal of this 3-year genetics continuing education project is to help ensure that Maine public health nurses have the knowledge, skills, and resources to provide high-quality services for patients, families, and communities affected by genetic-related conditions. Maine is a rural State that currently employs 136 public and community health nurses to deliver community-based health services. Our specific purpose is to offer public health nurses genetics continuing education in current clinical applications of the principles of human genetics. New genetic discoveries are making genetics a lifespan issue with implications for individuals from prior to conception through death. Public health nurses have a long history of caring for the genetic health needs of families and communities. They must have current knowledge about genetic conditions, interventions, and available resources so that they can continue to support patients and families to incorporate new genetic information into their lives in a meaningful way. To address this need, the Southern Maine Genetics Service, with support from the Maine State Genetics Program, conducted a survey of public and community health nurses to identify the current level of genetics knowledge, needs, and preferences for receiving ongoing genetics education. A similar survey was conducted among Arizona public health nurses for comparison. Barriers to receiving genetics continuing education were identified. Results of the surveys were used to develop a four-pronged approach in ensuring that Maine public and community health nurses have continuous and easy access to accurate and relevant genetics information to use in caring for individuals' genetic health. Our educational approach includes (1) development of a Genetics Resource Guide for each public and community health nurse and office that can be easily updated, (2) an annual statewide genetics educational conference repeated for both northern and southern parts of Maine, (3) creation of six case-based continuing genetics educational modules, and (4) establishment of a telemedicine network for continuing education offerings. Evaluation of the project will be ongoing and will provide documentation of the effectiveness and efficacy of project activities, including follow-up evaluation of public health nurses' response to and use of the Genetics Resource Guide and evaluation of the annual genetics conferences. In this presentation to ELSI, the first 2 years of the project will be described. This will include results of the surveys, development and components of the Genetics Resource Guide, dissemination, and evaluation plans. The authors hope that this genetics continuing educational approach will serve as a model for other State public health nursing agencies.

Disclosure of Familial Genetic Information: Perceptions of the Duty to Inform

Lisa S. Lehmann, MD, MSc, Jane C. Weeks, MD, MSc, Neil Klar, PhD, Lois Biener, PhD, Judy E. Garber, MD, MPH

Background: The familial implications of genetic information can lead to a conflict between a physician's duty to maintain patient confidentiality and a physician's ethical duty to inform at-risk relatives about genetic disease susceptibilities. As genes are discovered that can identify individuals at significant risk for adverse personal outcome, this conflict has become the subject of discussion and debate.

Methods: A one-time telephone survey of a population-based sample of 200 Jewish women assessed knowledge and

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attitudes about genetic testing. Attitudes toward sharing genetic test results with family members were evaluated with hypothetical scenarios describing an easily preventable disease, a disease in which the only prevention option is prophylactic mastectomies (breast cancer), and a nonpreventable disease.

Results: Nearly all respondents believed a patient should inform at-risk family when the disease is preventable (100 percent and 97 percent in the relevant scenarios) compared with only 85 percent who felt a duty to inform at-risk family about a nonpreventable disease ($p<.001$). The proportion of respondents who believed physicians should seek out and inform at-risk family against a patient's wishes was much lower. Only 18 percent of respondents to the easily preventable disease scenario, 22 percent of respondents to the breast cancer scenario, and 16 percent of respondents to the nonpreventable disease scenarios believed physicians should seek out and inform at-risk family against a patient's wishes.

Conclusions: Most women surveyed believe genetic information should be shared within families. Most women did not, however, believe physicians should seek out and inform at-risk family against a patient's wishes, even when this knowledge could result in disease prevention. Public opinion in this emerging area should be considered in the debate over the confidentiality of genetic information.

Use of Standardized Patients in Undergraduate Medical Genetics Education

Margaret M. McGovern, Randi Zinberg, Rosamond Rhodes, Mark Swartz, Devra Cohen

This project is a collaborative effort of faculty from the Mount Sinai School of Medicine in the Departments of Human Genetics, Medicine, Medical Education, Community and Preventive Medicine, and The Morchand Center for Clinical Competence. The project is designed to develop and evaluate the use of standardized patients (SP) for the education of medical students about the appropriate use and issues associated with genetic testing and evaluation. The project draws on the expertise of the participants in curriculum development, medical education, and medical genetics education for the development of SP cases relevant to genetics, and the accompanying curriculum, which provides students with skills to participate in the cases. In year 2 of the project, tools will be developed to evaluate student performance. The overall objectives of the research are to (1) develop SP cases that represent common encounters related to genetic evaluation and testing, (2) deliver a companion curriculum to achieve specified educational objectives, (3) integrate the new curriculum and cases into the undergraduate education of medical students at the Mount Sinai School of Medicine, (4) evaluate student performance in the SP encounter, and (5) evaluate student attainment of the educational objectives defined. The project is informed by two committees, an advisory committee of educators and a consortium committee with representatives from the medical schools in the New York area. In the first year of this project, the curricular components related to the enhanced ethics education have been developed, and the first two SP cases have been outlined according to a developmental blueprint utilized by The Morchand Center. Information about the case development process, the educational objectives, and the rationale for the SP encounter selection will be presented.

Coping Style Correlates of Participation in Genetic Testing for Inherited Breast and Ovarian Cancer Risk

Suzanne M. Miller, Jennifer L. Driscoll, Victoria A. Green, Michelle Rodoletz, Mary B. Daly, Michael A. Diefenbach, Joanne S. Buzaglo, Andrew K. Godwin, James S. Babb

This study focused on the psychosocial factors involved in decision-making for BRCA genetic testing among women ($n=281$) undergoing genetic counseling because of a putative hereditary family history of breast and/or ovarian cancer. We explored whether high monitors (who scan for and amplify disease-related threats) differed in their cognitive and affective reactions to testing in comparison with low monitors (who typically distract from threat-related cues). Participants attended an initial group breast and ovarian cancer educational session, followed by an individualized cancer-risk-counseling session. Psychosocial assessments were completed before the group educational session, before individualized risk counseling, and 1 week following the decision to donate blood for genetic testing. Outcomes of interest included satisfaction with the decision to donate blood and disease-related intrusive and avoidant ideation, as measured by the Revised Impact of Events Scale (RIES). The majority of the sample (60 percent) were unaffected relatives and well-educated, with 46 percent having at least a college education and 21 percent having graduate degrees. The average age was 47 years ($SD=12$). The results indicated that there were no significant differences in the decision to donate blood based on attentional style, with 95 percent of women providing a blood sample. However, there were significant differences between high and low monitors on measures of psychosocial adjustment 1 week post-blood draw. Specifically, high monitors were more satisfied with their decision to donate blood than low monitors ($p<.05$). Nonetheless, high monitors displayed greater disease-specific intrusive ($p<.05$) and avoidant ($p<.01$) ideation than did low monitors. These results suggest that dispositional attentional style is an important factor when designing and tailoring programs for genetic testing outreach and education. Ongoing research seeks to extend these findings to a sample of low income, minority women.

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Living with Marfan Syndrome: Perceptions of the Condition and Its Management

Kathryn Peters, Fanhui Kong, Robert Horne, Clair Francomano, Barbara B. Biesecker

We present data from a cross-sectional survey of 174 adults with Marfan syndrome regarding cognitive representations of the condition, use of prescribed cardiovascular medications, and adherence to physical activity guidelines. Approximately 71 percent of the participants reported regularly taking a beta or Ca++ channel blocking agent, and over 80 percent of these medication-takers reported that they adhere to their prescribed medication regimen. These medication-takers are psychologically receptive to the use of medication for prophylactic treatment of their cardiovascular problems; however, they do not view their medication as essential for health. Type of medication, duration of the medication regimen, and perceived risk of aortic root dissection were each significantly associated with an increased perception of the necessity of taking beta or Ca++ channel blockers. Additionally, the presence of cardiovascular symptoms and fatigue predicted use of medication. Over 80 percent of the respondents reported that they choose their physical activities with their diagnosis in mind. Modifying one's exercise activities was significantly correlated with an increased perception of Marfan syndrome as having negative consequences on the respondents' lives. Over 90 percent of the cohort experiences pain and/or fatigue on a regular basis. Also, over 95 percent view their condition as a serious or lethal disorder. The group reported ambivalence regarding the amount of control it has over the condition. These findings suggest that although affected adults adhere well to cardiovascular medication regimens and exercise modifications, they feel somewhat fatalistic about the health risks associated with Marfan syndrome. Genetic counseling should address beliefs about medication use and illness perception since they may have significant consequences for preventive health behaviors for individuals with Marfan syndrome.

Oncology Nurses' Attitudes Regarding Ethical Issues in Cancer Genetics

S.K. Peterson, P.T. Rieger, S.K. Marani

Cancer genetic counseling and testing are becoming increasingly available in clinical practice. It is important to understand oncology health care providers' awareness of and attitudes toward ethical issues in cancer genetics and their subsequent impact on the delivery of such services. We report findings from a study of U.S. oncology nurses regarding their attitudes toward ethical issues in cancer genetic testing. Study questionnaires were mailed to a random sample of 1,200 members of the Oncology Nursing Society (ONS) and 75 members of the ONS Cancer Genetics Special Interest Group (CGSIG). Of the 51 percent who responded, most were white and female and worked in hospital or outpatient settings. One-half were staff nurses and 8 percent stated a specialization in cancer genetics and/or affiliation with the ONS-CGSIG. Respondents indicated their level of agreement with a series of statements that reflected ethical issues in cancer genetics. Nearly all agreed that the risks of testing should be disclosed and that informed consent should be required prior to performing genetic testing. The majority also agreed that physicians should not contact at-risk relatives of patients with hereditary cancers without the patients' permission. Twenty-five percent agreed that it was appropriate to test minors for adult-onset diseases. Respondents who had completed higher levels of nursing education or continuing education in genetics and stated a specialization in cancer genetics were more likely to agree that persons have a right to decline receiving the results of genetic testing. Nurses who worked in managed care or private practice settings were more likely to agree that health care insurers should have access to genetic information. Our findings indicate that oncology nurses' attitudes toward ethical issues in cancer genetics may vary by educational level, position, practice setting, and experience. New technologies and the broadening application of genetic tests in oncology practice are likely to raise ethical issues that will impact oncology nursing practice. For this reason, it is important to make oncology nurses aware of current and emerging ethical issues in cancer genetics and to address these issues in primary nursing education as well as continuing education efforts regarding cancer genetics.

Outcomes of a Genetics Educational Program for Nursing Faculty

Cynthia A. Prows, MSN, RN, Carol Hetteberg, MSN, RN, Nancy Johnson, PhD, CGC, Kathy K. Latta, MSN, RNC, Anne Lovell, MSN, RN, Howard M. Saal, MD, Nancy Steinberg Warren, MS, CGC

The Genetics Program for Nursing Faculty (GPNF) is a multifaceted program funded by the Ethical, Legal, and Social Implications (ELSI) Research Program of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH). The specific aims of the program are to increase faculty knowledge about genetics and to increase the amount of genetics content in entry level nursing education program curricula. The components of the GPNF include an annual Genetics Summer Institute (GSI) and a variety of ongoing participant support strategies. From 1997 to 2000, four on-site GSIs provided genetics education and curriculum resources for a total of 126 nursing faculty participants representing 99 different nursing schools in 32 different States and the District of Columbia and Puerto Rico, Japan, and Singapore.

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Measured outcomes included change in knowledge and change in genetics content in nursing curricula. Analysis of 1997-1999 knowledge pre-test and post-test data revealed significant improvement ($p<.01$) in post-test scores in each of the 3 years. Analyses of pre- and post-curriculum survey data have been completed for 1997 and 1998 GSI participants. Comparison of pre- and post-data using paired t-test analyses revealed statistically significant ($p<.001$) improvement in the mean total number of genetic topics and conditions (18.6 at time 1 and 23.6 at time 2, with a t score of 4.4) taught in nursing curricula. Likewise, there was statistically significant ($p<.001$) improvement in the mean total time spent on genetics topics and conditions (13.3 hours at time 1 and 22.5 hours at time 2, with a t score of 3.7).

Ethical Issues in Human Population Genetics: An Analysis of the Human Genome Diversity Project

David L. Ross, Richard R. Sharp

The Human Genome Diversity Project (HGDP) is an international effort to collect DNA samples from human populations around the world. The goal of the project is to facilitate studies in population genetics, which ultimately could lead to a better understanding of human evolution, population-specific genetic markers, and inherited resistance and susceptibility to disease. Throughout the project's development, many ethical issues have been raised about the collection of biological materials from indigenous populations. Critics of the HGDP argue that proper informed-consent standards will not be followed, that participating communities will be harmed by the research, and that the project will not result in any benefits for those who agree to participate in the research. In response to these challenges, proponents of the project developed a Model Ethical Protocol for the collection of biological materials. This Model Ethical Protocol provides guidance to researchers, Institutional Review Boards, and funding agencies involved in collecting DNA samples for research in human population genetics. In this presentation, we (1) review the development of the HGDP, (2) analyze several ethical criticisms raised against the HGDP, (3) evaluate the HGDP Model Ethical Protocol, and (4) suggest how the collection of biological materials for research in human population genetics can be improved.

Culture and Family Interpretations of Genetic Disorder

Debra Skinner, Myra Roche, Don Bailey

This poster presentation, through graphic displays and text, will outline the design and theoretical framework of a recently funded project entitled Culture and Family Interpretations of Genetic Disorder. This ethnographic and longitudinal project employs observations of genetic counseling and evaluation sessions and a series of follow-up interviews to assess how parents from different cultural backgrounds who have a child or who are at risk for having a child with a genetic disorder seek out, understand, and use knowledge to interpret genetic disorders and their experiences and to make decisions about reproduction, health, and services. Participants are 100 families (40 African-American, 40 European-American, 10 Latino, and 10 Native-American) that attend a medical genetics clinic for counseling and/or evaluation and 30 families that are referred, but do not attend. Observations and interviews are designed to provide a comprehensive view of (1) families' understanding of the information disseminated in the clinical genetic visit (or, for nonattendees, reasons for not attending), (2) the nature of the communication in the clinical genetic session, (3) other sources of information and beliefs from which parents (or potential parents) draw to make sense of disability and genetic disorder, (4) the process of how they piece together knowledge and beliefs, (5) their experiences associated with the disorder, (6) how they make decisions for themselves and their (future) child based on their interpretations and experience, and (7) how these factors vary by culture or ethnicity as well as by family and child characteristics. Analyses will examine the social and cognitive practices through which families make sense of genetic knowledge in their social worlds, and findings will be used to assess the ways in which clinical genetic professionals frame and disseminate biomedical and genetic knowledge to culturally diverse clients.

Early Warning: Cases and Ethical Guidance for Presymptomatic Testing in Genetic Diseases

David H. Smith, Kimberly A. Quaid, Roger B. Dworkin, Gregory P. Gramelspacher, Judith A. Granbois, Gail H. Vance

Advances in genetic testing are making possible identification of a growing number of diseases linked to genetic defects. For people at risk for a genetic disorder — and for those who counsel them — genetic testing brings with it a host of difficult ethical concerns.

Should a fetus be tested for Huntington disease?
Should a woman try to find out whether she is at high risk for breast cancer?
Should counselors reveal negative test results to clients who decide after testing that they would rather not know?

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As we increase our ability to predict the likely onset later in life of a genetic disease, what shall we do with this knowledge?

Our book presents 29 case studies that identify the most important ethical issues that are likely to emerge from new technologies of genetic testing. We develop a series of guidelines based on the case studies. We argue that guideline formation is dependent on case analysis and that any statement of general guidelines must follow from concrete, practical discussion of specific situations. For these reasons, the guidelines presented are rooted in the case discussions and follow from the resolution of the cases. By providing the clinical origins and rationale behind each of its recommendations, the book aims to provide guidance for thinking through the ethical issues as well as a starting point for development of additional guidelines.

At one point in the Introduction, we wrote that the issues with which we are wrestling in this book are new, however much they many embody perennial human problems and concerns. The stakes for individuals at risk for genetic disease are high. Although our technical knowledge is expanding rapidly, our fund of moral wisdom and our level of social consensus are low. In these circumstances, it seems most appropriate to adopt a stance of moral modesty and a style in which the dominant tone is inquiry and deliberation, rather than the enunciation of a crisp verdict. We think the place to begin is with the conflicts and uncertainties that confront counselors and consultants; any guidelines that may be drawn should be clearly and explicitly connected to those conflicts and to uncertainties that may be unresolvable.

Lay Advocacy Organizations as Collaborative Partners in Genetic Research

S.F. Terry, V.H. Whittemore, P.F. Terry, J. Cody, M.E. Davidson

On the basis of decades of partnering with lay advocacy organizations involved in research, the Genetic Alliance defines research as a shared enterprise — collaboration between participants and researchers. The Alliance encourages and enables research that integrates human participant protections as a basic element of the research.

The Genetic Alliance provides education and mentoring to lay advocacy groups and researchers. An important Alliance publication, the Informed Consent brochure, has gradually become the prototype for research as well as clinical protocols. Informed consent procedures can serve as a useful starting point for developing consensus among research and consumer groups. Several Alliance programs increase awareness of researchers about the emotional, financial, and physical needs of families participating in research studies.

Although the cultures and strategy of scientific investigators and lay advocacy groups often differ, resulting in dynamic and creative tensions between shared and divergent interests, shared areas of interest can be articulated and research can be more ethical as a result. Inadequate genetic and medical privacy protections, on State and Federal levels, contribute to consumer reluctance to participate in research studies. Consumer concerns about genetic privacy, security of research records, and genetic discrimination are further deterrents.

Condition-specific groups represent the interests of research participantsCboth those directly involved in the study and those who stand to be impacted by study results. Issues of comprehensive informed consent process, confidentiality, test result reporting, and ethical treatment of participants are inherent areas of expertise for lay advocacy groups. By living in a virtual "community," which promotes the sharing of stories, it is easier to conduct research that is culturally competent, integrating the needs of the participants. This is essential information that is usually inaccessible to clinicians or researchers who employ more traditional participant and tissue solicitation methods. A number of collaborative models will be described.

The Partnership for Genetic Services Pilot Program: Consumer Indicators of Quality Genetic Services

Vicky Whittemore, Nisha Isaac, Nachama Wilker, Bowie Little, Cindy Holmes, Ann Smith, Mary Davidson

The provision of health care is undergoing revolutionary change and will continue to change as the results of the mapping of the human genome are translated into clinical care. Individuals with genetic conditions should receive quality, family-centered, and culturally competent health care, including genetic services. The Consumer Indicators of Quality Genetic Services and the Fundamental Elements of Genetics Services articulate the needs of genetic consumers (individuals with genetic conditions and their families).

The Consumer Indicators of Quality Genetic Services and the Fundamental Elements gathered input through four focus groups held in conjunction with the Consumer Committees of the Great Lakes Regional Genetics Group, the Pacific Northwest Regional Genetics Group, and the Mid-Atlantic Regional Human Genetics Network. The focus groups included 73 family members and individuals with, or at risk for, a range of genetic conditions. Data from the focus groups generated a list of the critical components of genetic services. Surveys were developed and mailed to more than 3,000 consumers and professionals and posted on the Genetic Alliance Web site.

Twelve Consumer Indicators of Quality Genetic Services were prioritized from the responses of 144 consumers. These Consumer Indicators focus on the needs of consumers for referrals to specialists and access to health care and resource

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information and call for consumers and providers to collaborate on the management of their health. The Consumer Indicators also point out the importance of the psychosocial impact of a genetic condition on the individual and his or her family at each stage of life.

The hallmarks of the Fundamental Elements are family-centered care throughout the life of an individual with a genetic condition and the importance of both peer support and a consumer-provider team for quality health care. The applications of the Consumer Indicators to health care provider education through the Partnership for Genetic Services Pilot Program will be illustrated.

Funded by the National Human Genome Research Institute's Ethical, Legal, and Social Implications Program, the Genetics Services Branch of the Maternal Child Health Bureau (Title V, Social Security Act, Health Resources and Services Administration), and SmithKline Beecham's Community Partnership Program.

Nursing CD-ROM on Ethical Issues of Genetic Testing

Janet K. Williams, M. Patricia Donahue, Debra Schutte, Kathleen Calzone, Juanita Strait, Susan Lenoch, Susan McCann

A thorough understanding of ethical principles underlying genetic testing decisions by individuals and families is essential for professional nurses. Yet few educational materials on genetic and ethics topics are available to student and practicing nurses, and none provide the opportunity for self-directed learning in an interactive, computer-based format. The purpose of this project is to develop an interactive multimedia educational product in a CD-ROM format to provide education to nurses on ethical and genetic principles and their application to genetic testing in the practice setting. Breast cancer and Alzheimer disease case studies will be presented to allow application of ethics and genetics principles by nurse learners. The 3-year project is currently nearing completion of the first year in which content, format of information presentation, and evaluation components are being developed. Experts in oncology, gerontology, and genetic nursing will evaluate the product before field testing with students and practicing professional nurses. Elements of the content and product development will be presented.

Ethical Issues Related to Relatives' Enrollment in Genetic Family Studies

N. Arar, R. Plaetke, H. Hazuda, R. Duggirala, V. Sartorio, B. Kasinath, M. Stern, H. Abboud

The success of any genetic family study (GFS) depends on the recruitment of an adequate number of unaffected family members. Little is known about subjects' opinions concerning participation in GFS and their attitudes toward involving their relatives.

We applied ethnographic techniques of open-ended questions to explore patients and relatives' perspectives on the following ethical questions: (1) should probands or investigators contact relatives for GFS? (2) Should probands reveal information about relatives without first obtaining their written consent and (3) How far should researchers go in ensuring that subjects understand the risks and benefits associated with participation in GFS?

Subjects were recruited from the Family Investigation of Nephropathy and Diabetes study at the University of Texas Health Science Center at San Antonio. Semistructured interviews were conducted with 15 subjects, mainly diabetic nephropathy probands with a family history of diabetes (data collection still ongoing). Interviews were recorded, transcribed, and content-analyzed. For subjects, the mean age was 53 years (range, 42-62). Eighty-seven percent were Mexican-American, not formally educated beyond high school and unemployed due to disability. All subjects indicated that they would like to participate in GFS to learn more about diabetes and to help future generations. Seventy-three percent were willing to invite relatives to the study but favored that they contact relatives, not investigators. Of these, 87 percent preferred contacting close relatives (i.e., children and sibs) and not members of their extended family. Forty percent believed that their relatives would participate while 34 percent did not, suggesting alternative strategies may be needed to enroll adequate number of relatives. All participants found it acceptable to provide some information about relatives prior to obtaining their consent. Seventy-three percent agreed that health and demographic information (except for income) were acceptable to provide, whereas releasing names and addresses was less appropriate.

None of the subjects were fully aware of the risks associated with participation in GFS. While discussing potential risks (i.e., confidentiality of genetic information), they did not consider these issues to influence their participation or potentially have an impact on their lives.

We suggest that while conducting GFS, researchers address these ethical issues to better identify ways of conducting genetic research and protecting subjects.

The ascertainment of families has been funded by NIDDK, NIH.

Genetic Research on Smoking: Foreseeable Applications and Policy Issues

Lorraine Caron, PhD, Thomas A. Raffin, MD, Jennifer R. Fishman, MA, Barbara A. Koenig, PhD

Background/research question: Genetic research on susceptibility to nicotine addiction is emerging, with the hope of generating innovative interventions. How will this genetic paradigm influence policies and practices to reduce tobacco use?

Methods/progress to date: To address this question, we reviewed the literature and developed qualitative interview guides. To date, we conducted and analyzed interviews with scientists studying genetic and environmental determinants of smoking as well as scholars and policy experts working on tobacco control strategies (n=13) to assess (1) genetic research current status, (2) its foreseeable applications, and (3) the potential impact of genetic explanations.

Results: Experts' views about the genetic contribution to smoking converged toward the following scenario: It will be 10 years or longer before genetics-based interventions are developed; many genes with weak effects will be identified instead of a gene with strong effects; the genes will most likely play a role in addiction and in the inability to quit, rather than in the initiation of smoking.

A majority of experts believe that preventive genetic testing, i.e., testing an identifiable population such as adolescents and then targeting interventions to individuals most susceptible, is highly improbable. In contrast, tailoring cessation treatments through differential genetic testing of smokers, i.e., testing for common genetic differences correlated to the inability to quit, and adapted pharmacotherapy (pharmacogenetics) are seen as the most likely and useful applications.

Most experts recognize the power of genetic explanations to influence public policy. One positive impact is that genetic understanding might further medicalize smoking, leading to increased health insurance coverage for cessation treatments. The downside is that providing a biological explanation for why people smoke might be viewed as fatalism, making people more resistant to preventive messages. One immediate concern is that research findings could be used to single out and discriminate against socially or racially identified groups already labeled as more likely to smoke.

Significance/future directions: Future inquiry should focus on the perspectives of pharmaceutical and health insurance companies, which will be key players in developing genetic research applications. Moreover, in normative analysis aiming to maximize benefits over harms, it is imperative to pay close attention to the sociocultural context that shapes research into the genetic epidemiology of smoking.

This project is supported by a grant from the State of California Tobacco-Related Disease Research Program.

Insurance Experience in a BRCA1/2 Testing Protocol

A. Chittenden, K. Smith, L. DiGianni, C. Lund, K. Schneider, K. Kalkbrenner, J. Garber

Information on insurance coverage of BRCA1/2 testing was collected for participants in an ELSI-funded multicenter randomized trial of two different counseling interventions. In our program, test cost is subsidized for women who self-pay rather than use insurance. The fees are approximately $200 for single site (SS) analysis, $250 for multisite panel (MP), and $1,000 for patients having full sequencing (FS) of both genes.

As of 9/1/2000, 198 women had been tested through our program; six women are excluded from the current analysis because their center covered the costs in full. Of the remaining 192 women, all had some type of health insurance at enrollment, and 1/26 (4 percent) with SS analysis, 22/70 (31 percent) with MP, and 55/96 (57 percent) women with FS chose to go through their insurance company. Of the 77 women who pursued insurance coverage, 62/77 (81 percent) received insurance coverage for at least 70 percent of the usual and customary cost of the test and (45/77) 58 percent were covered at 100 percent.

Participants were more likely to pursue insurance coverage as the test cost increased (chi2 analysis, $p<.001$). Interestingly, 25/26 (96 percent) women from families with known mutations chose not to pursue insurance coverage for the cost of the test. Cancer survivors were more likely to pursue insurance coverage than unaffected women. This was true even excluding women testing for a known familial mutation (chi-square analysis, $p<.001$ including known mutation families, $p<.01$ excluding known mutation families). However, for MP analysis, cancer survivors were no more likely to pursue insurance coverage than unaffected women when we excluded participants testing for familial mutations. While there was no difference between self-pay and insurance participants regarding their perceived mutation risk, self-pay participants had significantly higher baseline insurance knowledge scores than women who sought insurance coverage (chi-square analysis, $p<.001$).

Areas for further study include balance billing and trends in insurance usage. We will collect actual participant test costs as usual, and customary coverage can be misleading. We will also look at the cohort of approximately 100 women remaining to be accrued to evaluate changes in the pursuit of insurance coverage due to recent Massachusetts genetic discrimination legislation.

Eliciting Community Voices in Shaping Genetics Policy

Tene Hamilton, Yvonne Lewis, Dan Soza

This poster session is based on the ELSI-sponsored project, Communities of Color and Genetics Policy. The project engaged 15 African-American and Latino communities in Michigan and Alabama in learning about genetics, identifying issues of concern and utilizing the process of dialogue in developing recommendations for policies to address these issues.

The poster session will describe the partnerships that were developed to carry out the project, connecting three universities (University of Michigan, Michigan State University, and Tuskegee University) and 15 community-based organizations (CBOs). The role of the CBOs proved crucial to the project's success in eliciting grassroots community engagement by utilizing their knowledge of community culture and characteristics in structuring the dialogues and relying on the trust of their respective communities in securing participation in the dialogues and frank, open discussion of the issues.

The poster session will describe the characteristics of the communities and the CBOs that secured their engagement, will identify the issues of concern drawn from the focus groups comprising community members, and will present the voices of community members elicited through the dialogue process. Photographs and audiotape samples will be presented to convey the flavor of the dialogues and to describe the CBO leadership who were an integral component of the project.

Information Needs of Women at Risk for Hereditary Breast Cancer

W.F. Cohn, G. Fraser, S.M. Jones, M.E. Ropka, S. Miesfeldt

Problem: Breast cancer survivors facing the possibility that their cancer may be associated with an inherited syndrome may have unique information needs.

Purpose: To identify and understand information needs related to concern for hereditary breast cancer (HBC).

Methods: A multimethod approach, combining qualitative data from in-person interviews and quantitative data from mailed surveys. A total of 314 women with a history of breast cancer, recruited from 34 Virginia hospitals, completed a family history questionnaire to identify those at presumed risk for HBC. Of the 138 respondents at risk for HBC, 20 from diverse backgrounds were selected for an in-depth interview. Interviews were conducted using an interview guide about HBC issues developed by literature review and clinical expertise. The qualitative data were content-analyzed, yielding information themes. All 138 at-risk women were mailed a survey to assess knowledge, beliefs, and attitudes regarding HBC. Survey topics included (1) health history and experience, (2) knowledge about HBC, (3) beliefs about breast cancer causes, and (4) information resources about HBC.

Results: Content analysis of the interviews identified the following major themes related to breast cancer information: needs, timing/delivery, sources, and comprehension. Questionnaire results revealed that only 53 percent of women reported looking for information regarding HBC after their diagnosis. Women who sought information were interested in (1) how their diagnosis affected their children's risk (41 percent), (2) how their diagnosis affected other family members' risks (23 percent), (3) DNA testing information (13 percent), (4) treatment differences for inherited breast cancer (13 percent), and (5) genetic counseling (10 percent). Most women (78 percent) reported that they were able to find the information they sought.

Conclusions: Many at-risk breast cancer survivors may not pursue information about HBC. Among respondents, there was a greater interest in information concerning their children's breast cancer risk than in information about genetic counseling or testing.

The Human Genome Project and Mental Retardation: An Educational Program

Sharon Davis, PhD

In February 1996, the DOE ELSI Program granted funds to The Arc of the United States to conduct an educational program with two major purposes. The first was to make our members aware of this scientific undertaking, the Human Genome Project. The second was to examine the critical issues related to new genetic discoveries affecting people with mental retardation and their families.

The Arc is a national organization on mental retardation whose 140,000 members are united by a concern for the welfare of people with mental retardation and their families. When we began our education program, we surveyed The Arc's Board of Directors and learned that only 2 of 24 had heard of the Human Genome Project. Represented on the board were individuals who had a family member with Down syndrome, fragile X syndrome, Angelman's syndrome, Cri du Chat, and perhaps other genetic disorders. Not only had they not heard of the Human Genome Project, they also were generally unaware of the ethical, legal, and social issues affecting families with genetic disorders.

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To remedy this general lack of knowledge among The Arc's leaders and members, we developed educational materials that were furnished to our chapters and members on topics such as these:

- Overview of the Human Genome Project
- Genetic causes of mental retardation
- Genetic discrimination
- Genetic testing, screening, and counseling
- Genetic privacy
- Gene therapy

We also produced factsheets on such specific disorders as PKU, Prader-Willi syndrome, and fragile X syndrome. Interested persons can access materials through our Web site (http://thearc.org/).

Workshop presentations at chapter conferences were another part of our educational efforts. We introduced the Human Genome Project using clips from the video developed by the NIH National Human Genome Research Institute. We reviewed the ELSI Program and specific issues we addressed in three major areas:

- Discrimination based on a person's genes
- Decisions involving genetic testing, screening, and counseling
- Ethical issues involving genetic therapies that may eventually cure mental retardation (and issues related to deciding to participate in genetic research)

Workshop participants discussed issues and presented their views in small groups as they discussed case scenarios designed to elicit a range of perspectives. We conducted pre- and post-testing to gather our members' views on various issues. The Arc's goal was to begin to create an adequate level of awareness among its members on the ethical, legal, and social issues so that family voices can be heard before harmful policy is created.

The poster session will display materials developed through the project and summarize views of members of The Arc on selected ELSI issues.

Development of a Genetic Education Curriculum for Practicing Health, Education, and Human Service Professionals

Valerie DeCoux, PhD, RN

A lengthy survey of 306 Mississippi professionals revealed a lack of knowledge regarding the basics of human genetics as well as recent advances in the field. With the exception of one item regarding confidentiality safeguards, all other questions on genetic ethical, legal, and social issues elicited a not-applicable response by respondents. The results of the survey, as well as comments by participants in initial workshops, led the Institute for Disability Studies to identify a need for basic training in human genetics, more advanced clinical training, and a focus on ethical, legal, and social issues. In response to these educational needs, a consortium was formed among the Institute for Disability Studies, a university-affiliated program at the University of Southern Mississippi, the Mississippi State Department of Health, and the University of Mississippi Medical Center to develop the GenESES project, which was funded by the National Institutes of Health. The purpose of the project was to develop, implement, and evaluate a model comprehensive genetic education program for health, education, and human services professionals that emphasized clinical relevance for the practicing professional. It was critical that the delivery be carried out with content and in a format relevant to the daily practice and communities of professionals in rural and poor regions of the Southeast. The GenESES curriculum was utilized, evaluated, and modified through workshops for practitioners in the field and lectures for undergraduate and graduate students at the university.

This presentation will share the results of the survey of professionals and a display copy of the curriculum. Specific methodologies used in presenting the curriculum to approximately 1,359 trainees representing more than 10 disciplines will be delineated. Successful strategies for meeting genetic education needs across a multitude of diverse disciplines will be shared by project staff.

How Accurate Were Participants in Two Cancer Genetic Testing Programs (P53 and BRCA1) in Anticipating Their Emotional Reactions to Disclosure of Their Test Results?

Michel Dorval, Andrea Farkas Patenaude, Katherine A. Schneider, Stephanie A. Kieffer, Lisa DiGianni, Kathy J. Kalkbrenner, Jonas I. Bromberg, Laura A. Basili, Kathleen Calzone, Jill Stopfer, Barbara L. Weber, Judy E. Garber

Background: Structured encouragement to consider the potential emotional impact of disclosure of a positive and a negative result is a commonly used strategy in genetic counseling. However, the extent to which this approach helps individuals

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considering genetic testing to conceptualize the experience is not known. Individuals who accurately anticipate how they will react emotionally to the forthcoming disclosure of their test results may be less likely to experience psychological difficulties following genetic testing.

Purpose: We examined the ability of individuals undergoing genetic testing for cancer susceptibility in two structured research protocols at the Dana-Farber Cancer Institute and University of Pennsylvania to accurately anticipate emotional reactions to learning their test results. We also explored whether accuracy of emotional anticipation was associated with post-disclosure psychological adjustment.

Methods: Data from 65 individuals were analyzed: 24 tested for p53 mutations in Li-Fraumeni syndrome families (unaffected) and 41 for BRCA1 mutations for hereditary breast-ovarian cancer susceptibility (34 unaffected, 7 affected). Subjects were from families where a germline mutation had been previously identified. At pre-test, subjects rated the extent to which they anticipated feeling each of six emotional states (relief, happiness, sadness, guilt, anger, worry) following disclosure that they (1) did or (2) did not carry the familial mutation. After receiving their test result, they rated their feelings on the same scale of emotions. Extent of accuracy and association with psychological distress at 6 months, assessed with standardized measures, were evaluated.

Results: Overall, mean levels of emotional reactions after receiving test results were not different from those anticipated before result disclosure. However, affected BRCA1 carriers experienced higher levels of anger and worry than they had anticipated. Underestimation of subsequent distress emotions related to test result was associated with a significant increase in general psychological distress at 6 months.

Conclusion: Unaffected individuals in cancer predisposition testing programs are generally accurate in anticipating emotional reactions to test results. However, cancer patients may underestimate their distress following disclosure of positive results. Until information from larger cohorts is available to clarify these issues, it may be important to ensure that individuals with cancer contemplating genetic testing receive careful preparation and support.

How Men View Genetic Testing for Prostate Cancer Risk: Findings from Focus Groups

D.J. Doukas, M. Fetters, J.C. Coyne, L.B. McCullough

Objective: To determine the values, beliefs, and attitudes that influence a man's intention to undergo or defer genetic testing for prostate cancer risk.

Design: Qualitative. Focus group interviews in 12 focus groups were conducted to identify key values and beliefs about genetic testing for prostate cancer risk in anticipation of its future availability.

Setting: Medium-sized, Midwest, U.S. city.

Participants: Community sample of 90 laymen of diverse educational, ethnic, and age backgrounds.

Analysis: Descriptive statistics and immersion; crystallization to identify themes and subthemes.

Results: The major areas of concern were distilled into the following themes: beliefs about consequences, expectations, benefits for patients, beliefs about barriers, and susceptibility concerns.

Conclusion: Identifying these men's values will help health professionals anticipate the informational and ethical needs of patients in the informed consent process. Men will need to understand how such testing may affect their planning regarding future prostate health and how medical information is used outside the physician-patient relationship.

From Doukas DJ, Fetters M, Coyne JC, McCullough LB. How Men View Genetic Testing for Prostate Cancer Risk: Findings from Focus Groups. Medical Genetics 2000; 58: 169-176.

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Genetic Testing of Children and Adolescents: Voices from Affected Families

Joanna H. Fanos, PhD, Jennifer M. Puck, MD

Professional societies and others have suggested a careful weighing of potential benefits and harms of genetic testing (carrier or predisposition) of children and adolescents. There is general agreement that presymptomatic testing for adult-onset disorders is recommended if positive intervention is available, with considerably more controversy surrounding carrier testing for other types of conditions. However, attempts to date to establish general guidelines have not taken into account that disorders vary considerably along multiple dimensions that may influence attitudes, behavior, and outcomes in relation to carrier testing. In our NIH/NHGRI study of siblings of males with X-linked severe combined immune deficiency (XSCID), only 10 percent of our respondents favored waiting to test girls for carrier status until they reached the age of maturity. The particular features of this disorder — X-linked inheritance, severe infantile presentation, and recent development of effective treatment — shaped family attitudes; their level of knowledge was high and their attitudes more optimistic than those of families experiencing

genetic disorders with progressive, relentless deterioration. The cohort of XSCID-affected siblings showed strong enthusiasm for carrier testing and the pursuit of medical knowledge. Rather than adopting generalized standards for all carrier testing, we urge an approach that takes into account specific characteristics of disorders — autosomal recessive versus X-linked, treatable versus untreatable, and progressive. Geneticists and counselors should entertain adolescents' requests for carrier testing when identification of carrier state may confer beneficial early diagnosis and treatment of a carrier's affected offspring. For our subjects who had directly participated as children in early linkage studies, it is important to note that their concern was not for which particular developmental stage is best for genetic testing, but rather for the crucial role that communication about the procedure should play. Siblings need help understanding personal risk as well as medical procedures. More attention to helping families communicate as they struggle to make sense of the medical complexities with which they are confronted would be invaluable.

Population Genetic Screening Programs: Recommendations of the European Society of Human Genetics

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Genetic screening is increasingly possible for a large number of disorders. The question of whether this approach should be offered at the population level is a challenge to health care providers, the medical community, and policymakers. Genetic screening can be of benefit but can also do harm. The availability of genetic tests at low cost may lead to the systematic offer of screening tests without the appropriate medical environment for providing information prior to testing and counseling afterwards. There is therefore a need to introduce effective and acceptable safeguards, standards, and procedures relating to implementation and organization of genetic screening programs.

To discuss these issues and produce recommendations from the professional point of view, the Public and Professional Policy Committee (PPPC) of the European Society of Human Genetics (ESHG) organized a workshop on November 19-20, 1999, in Amsterdam to which 51 experts from 15 European countries were invited. They received before the meeting a working paper developed by the PPPC which was revised after the meeting to take into account the points of views expressed by the participants. This document (available on the Web site of the ESHG) focuses on the issues surrounding potential screening programs that require further discussion before their introduction. This document aims to increase, among the health care professions and health policymakers, an awareness of the potential screening programs as an issue of increasing concern to public health.

Following the workshop, the PPPC issued statements and recommendations that are expected to reflect the views of the scientific community. They were endorsed at the Amsterdam ESHG annual meeting in May 2000. They focus on (1) the definition of genetic screening, (2) the potential benefit and harm of genetic screening, (3) criteria for introducing genetic screening programs, (4) the principles for defining screening programs, and (5) the organization of genetic screening programs.

Positive Exposure

Rick Guidotti, Diane McLean, PhD, MPH

The project is a unique collaboration between photography and genetics that challenges the stigma associated with difference. Utilizing photography and video interviews, POSITIVE EXPOSURE investigates the social and psychological experience of having albinism. The project explores the unique, positive and negative perceptions, myths, and stereotypes of albinism within diverse cultures as well as those that cross cultural boundaries.

At the heart of the project, albinism is a metaphorical lens through which one can explore social attitudes toward all difference. The results resonate for everyone who has experienced the negative, social repercussions of an unconventional physical appearance or has been forced to hide all or part of his or her identity for fear of public ridicule and judgment.

Insults, fear, and discrimination color the experience of many people with genetic conditions. The majority of stereotypes for people with genetic conditions are negative, with little opportunity to celebrate difference, illustrate strengths, and share experiences.

The photography and videography process of POSITIVE EXPOSURE offers a fresh exploration of the joy that comes with self-acceptance, providing a look at how an individual's self-perception can be altered by having his or her own image reflected through a positive and life-affirming lens.

POSITIVE EXPOSURE will serve as a model for future projects and a metaphor for all conditions that vary from what is considered "normal," promoting the concept of universal and intrinsic humanity.

POSITIVE EXPOSURE has implemented the photography and interview process extensively throughout the United States

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and Canada as well as French Polynesia, the Cook Islands, Fiji, Samoa, New Zealand, Australia, Panama, and England. The team is currently planning to travel to Puerto Rico, Africa, Asia, and India.

POSITIVE EXPOSURE is collaborating with the Genetic Alliance and National Organization for Albinism and Hypopigmentation (NOAH). The project was featured in *LIFE* magazine and *WE MEDIA* magazine and on Fox News WebMD.

Family Stories, Communication Patterns, and the Use of Heuristics in Familial Breast/Ovarian Cancer Families: An Exploratory, Qualitative Study

R. Kenen, R. Eeles, A. Ardern-Jones, Mitchell

Aims: This study explored how clients seeking breast/ovarian cancer genetics risk counseling are influenced by (1) family stories, (2) family communication patterns, and (3) the use of heuristics (inferential shortcuts used to make sense of complicated information) in interpreting and assigning meaning to the genetic information they receive. This research was undertaken to better understand and improve breast/ovarian cancer genetics risk counseling.

Methods: We conducted an IRB-reviewed exploratory, qualitative study at a major clinical and research cancer center in the United Kingdom from January to June 2000. Twenty-one semi-structured, in-depth interviews were conducted using a purposive sample of women coming to the cancer genetics risk clinic for the first time. Questions were asked about family relationships, how relatives talked about cancer in their family, the client's use and dissemination of counseling information, what they thought was the likelihood of their developing cancer, and how they might try to prevent it. The interview material was supplemented by 5 months of weekly participatory observation at the clinic.

Results: The interviewees did not have any strong family stories about breast/ovarian cancer that came down through the generations, but there were a few misconceptions, e.g., cancer skips generations, men cannot carry the cancer gene, and birth order is related to the chance of developing cancer. Regardless of whether the family was close or not, there were differences within almost every family in their members' willingness to obtain and use genetic cancer risk information. The four heuristics focused on (1) representativeness, (2) availability, (3) anchoring, and (4) illusion of control. For most of the women, their prior experiences with cancer in family members or friends exerted a greater impact on their feelings than did the statistical risk that they faced.

Discussion: Most of the interviewees, while aware of cancer in their families, did not realize until fairly recently that these cancers might be hereditary, which may account for the dearth of family stories. Family communication patterns around familial breast cancer were extremely complex, varied, and, frequently, emotionally laden. The psychosocial context that the client brings to the cancer genetics risk clinic needs to be incorporated into the counseling process.

Racial/Ethnic Differences in Prenatal Diagnostic Test Use and Outcomes: Preferences, Socioeconomics, or Patient Knowledge?

Miriam Kuppermann, Elena Gates, A. Eugene Washington

Background: Prenatal diagnosis of chromosomal abnormalities, first introduced in the late 1960s, has become common in care of women who will be age 35 or older at the time of delivery. Reports documenting differential use of prenatal diagnostic services across racial/ethnic groups surfaced soon after amniocentesis was made available. However, at the time of this study, no comprehensive analyses comparing the relative rates of prenatal diagnosis use and Down syndrome-affected births in a racially/ethnically diverse group of women (including African-Americans, Asians, Latinas, and whites) had been published.

Objective: We conducted a study to determine whether use of chorionic villus sampling and amniocentesis varies by racial/ethnic group and, if so, whether this variation leads to differences in the prevalence of Down syndrome-affected births in women ages 35 and older, the ages at which prenatal diagnosis is currently offered.

Methods: We reviewed medical charts of 238 women ages 35 and older presenting for care at University of California, San Francisco, by 20 gestational weeks in 1993-94 to assess prenatal diagnostic test use. The Birth Defects Monitoring Program provided us with data on the prevalence of Down syndrome-affected births in California from 1983 to 1989.

Results: We found that Latinas and African-Americans were much less likely to undergo prenatal diagnosis than were whites and Asians. Odds ratios (OR) and 95 percent confidence intervals, with white as the reference group, included Asians 1.16 (0.57-2.36), Latinas 0.19 (0.08-0.43), and African-Americans 0.19 (0.07-0.49). Trends persisted, at diminished magnitude, after adjustment for socioeconomics (ORs = Asians 1.77 (0.78-3.98), Latinas 0.28 (0.09-0.83), and African-Americans 0.33 (0.10-1.10)). Non-white women ages 35 and older were significantly more likely than whites to give birth to a Down syndrome-affected infant (risk ratios = Asians 1.81 (1.61-2.03), Latinas 3.00 (2.74-3.28), and African-Americans 1.86 (1.63-2.11)).

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Conclusions: Racial/ethnic differences exist in prenatal diagnostic test use and associated outcomes in women ages 35 and older. Socioeconomic factors are partially responsible. Further research is needed to determine the roles of structural issues in the delivery of care, provider attitudes, patient education, patient preferences, and other factors.

Consumers' Perspectives of Genetic Discrimination: Professional, Industry, and Policy Responses and Impact

E. Virginia Lapham, PhD, Joan O. Weiss, MSW, Chahira Kozma, MD, Mary Ann Wilson, BA

In a study of the perceptions of 332 members of genetic support groups with one or more of 101 different genetic disorders in the family, it was found that as a result of a genetic disorder, 25 percent of respondents believed that they or an affected family member were refused life insurance, 22 percent believed they were refused health insurance, and 13 percent believed they were denied or let go from a job. Fear of genetic discrimination resulted in 9 percent refusing to be tested for genetic conditions, 18 percent not revealing genetic information to insurers, and 17 percent not revealing information to employers. Following publication of the data in the 25 October 1996 issue of *Science*, the media publicized it widely both nationally and internationally; the health insurance industry (HIAA) attacked it as biased and used its own biased analysis to declare that genetic discrimination does not exist in health insurance; and professionals cited it in more than 58 articles in professional journals sometimes alluding to weak methodology. Public officials at State levels used the data to pass legislation on genetic discrimination in more than half of the States. Senator Kennedy's staff used it in drafting the Kassebaum-Kennedy Portability Act. Congresswoman Louise Slaughter's staff used it in drafting Federal legislation that is still pending, and President Clinton cited the *Science* article as one of two studies underlying his historic action to ban genetic discrimination in the Federal workplace on 8 February 2000. Thus, 332 telephone interviews intended to provide information for genetics education of consumers and health professionals were catapulted into the public arena through publication in a scientific journal. NHGRI-ELSI funds have made a major contribution toward policies and legislation to prevent genetic discrimination in health insurance and the workplace. Was the criticism worth it? Would the grantees do it again? Both the study and the responses that continue today will be considered and members of the audience will be invited to share their views.

Genetic Variation and the Category of Race in SNPs Research

Sandra Soo-Jin Lee, PhD, Joanna Mountain, PhD, Barbara A. Koenig, PhD

Despite its lack of biological validity, the category of race and assumptions of racial difference continue to influence the trajectory of medical research and health care practice in the United States. Recent developments in human genetics research, most notably in both the privately and publicly funded Human Genome Project, have led to a heightened search for genetic variation among populations. Although 99.9 percent of the genome sequence is identical in all humans, the remaining 0.1 percent varies among individuals. There is increasing interest in the correlation between these variations and the onset of particular diseases. An important focus of this search for genetic differences has been on single nucleotide polymorphisms (SNPs). SNPs represent variations in single base combinations that are believed to occur in approximately every 1,000 of the 3 million base couplets of the genome. In discovering the location of SNPs in different populations, research scientists hope to understand the genetic etiology and relative incidence of disease among groups. The creation of the DNA Polymorphism Discovery Resource by the National Human Genome Research Institute represents an effort to further this trajectory of SNPs research. The resource contains the DNA samples of 450 anonymous U.S. residents who were chosen to represent the diversity of the United States. In an effort to achieve racial and ethnic diversity, the resource contains samples from individuals with ancestry from Europe, Africa, Asia, and the Americas. While samples distributed by the resource are combined in ways to ensure this diversity, information on individuals has been stripped of all identifying information. In an effort to identify the significance of both cultural and scientific understandings of racial classification on SNPs research, this paper examines both the decisions leading to, and the implications of, eliminating racial identifiers from genetic materials. Of particular interest is the impact this decision may have on the research questions posed by scientists interested in genetic variation. In addition, this paper addresses the racial categorization of genetic materials practiced by other cell repositories and the implications of the use of such taxonomies for research on health disparities between racially identified populations.

A Population-Based Study of Ashkenazi Jewish Women's Attitudes Toward Genetic Discrimination and BRCA1/2 Testing

Lisa S. Lehmann, MD, MSc, Jane C. Weeks, MD, MSc, Neil Klar, PhD, Lois Biener, PhD, Judy E. Garber, MD, MPH

Context: The Human Genome Project continues to produce an increasing number of genetic susceptibility tests. Some of these

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genetic tests target social or ethnic groups that are at increased risk of developing a disease. The Ashkenazi Jewish community is one ethnic group that is an ongoing source of genetic investigation.

Objectives: To assess the attitudes of a population-based sample of Ashkenazi Jewish women toward breast-ovarian cancer susceptibility testing (BRCA1/2). In particular, we assessed concerns about group discrimination and perceptions of the advantages and disadvantages of BRCA1/2 testing and assessed the relationship between concerns about discrimination and the potential benefits of genetic testing.

Design: A population-based telephone survey of 200 Jewish women.

Participants: Adult women (n=200) who had at least one parent of Jewish descent.

Results: A minority of women (17 percent) in this study expressed concern or discomfort with Jews being offered BRCA1/2 testing. Most women believed there were scientific reasons for testing Jews (71 percent), and only 4 percent of women felt that research that focused on Jews was bad for the Jews as a group. Increased concern about genetic discrimination was associated with women who were highly educated (OR, 2.68). Of women surveyed, 40 percent were interested in BRCA1/2 testing, 40 percent were not interested, and 20 percent were uncertain about whether they would obtain BRCA1/2 testing. Increased interest in genetic testing was associated with a desire to obtain information about children's risk of disease and valuing information for its own sake.

Conclusions: The majority of a population-based sample of Jewish women did not express concerns about group discrimination resulting from genetic testing. Women who are highly educated are more concerned about genetic discrimination. There is significant variation in Jewish women's interest in breast cancer susceptibility testing.

Implications of the Geneticization of Health Care for Primary Care Practitioners

Mary B. Mahowald

This project targeted primary caregivers with no special training in genetics with the goal of facilitating their understanding of human genetics and its implications for their practice. For each of five areas of primary care, general medicine, pediatrics, obstetrics and gynecology, family medicine, and nursing, we identified three individuals trained in that area: one with added expertise in genetics, another with added expertise in ethics, and a third who was a fellow or trainee. Each team of three had responsibility for developing a transferable educative program for practitioners in their field. Each genetics expert presented grand rounds, outlining what the general practitioner in his or her field needs to learn to bring the fruits of advances in genetics to patients. The role of the ethics experts was to provide a similar function by identifying the relevant ethical and policy issues. The role of the fellow was to work with the genetics and ethics experts to develop an educational program for others in the field in order to facilitate its carryover to the broader community of generalists. Fellows involved in this project were pursuing their fellowships in the MacLean Center for Clinical Medical Ethics and the Robert Wood Johnson Clinical Scholars programs at the University of Chicago. These individuals have since moved to training programs at other institutions throughout the country and beyond.

The topics we addressed included scientific and clinical issues (e.g., principles of genetics, genetic counseling), generic issues (e.g., concepts of health and genetic identity; impact of cultural, gender, and class differences), issues specific to each discipline (e.g., testing children, susceptibility testing), and policy issues (e.g., insurance risk). All were presented at a national conference and have led to a collection of readings on the topics, *Genetics in the Clinic: Clinical, Ethical and Social Implications for Primary Care*, edited by Victor McKusick et al. and scheduled for publication by Mosby, Inc., in 2001.

Privacy and Fairness in the Use and Interpretation of Genetic Information: The Example of a Workplace Notification Program for Chronic Beryllium Disease Susceptibility

Babette L. Marrone, Geoffrey Lomax

Los Alamos National Laboratory (LANL) performs beryllium metal work that exposes several hundred workers to beryllium dust and fumes. Exposed workers can contract chronic beryllium disease (CBD). Research suggests that specific gene combinations confer a thirty-fold risk for development of CBD. Our challenge is to develop a responsible program for informing LANL workers about their genetic risk for CBD which also incorporates privacy, fairness, and the right to work. The development and implementation of such a program creates important research and policy questions for the Department of Energy and the University of California (UC), which operates LANL. A collaborative research study between LANL and the University of California at Berkeley (UCB) has been initiated to address these questions. These questions include (1) how should the results of genetic testing for CBD susceptibility factors be communicated to worker participants, (2) what is the impact of participant notification, and (3) how does this experience inform future LANL and UC policies regarding worker access to genetic information? We will describe our approaches to answer these questions and

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present a "strawman" strategy for the integration of genetic testing for CBD susceptibility into the ongoing Beryllium Medical Surveillance Program at LANL.

Breast Cancer Survivors' Attitudes About Educating Their Children Regarding Breast Cancer Risk

S. Miesfeldt, W.F. Cohn, S.M. Jones, J. Weinstein, M. Ropka

Introduction: We explored the attitudes of a group of breast cancer survivors concerning whether, how, and at what age children from a breast cancer-affected family should be educated about their potential genetic risk for cancer development.

Methods: Women with a history of breast cancer, recruited from 34 Virginia hospitals, responded to two mailed surveys: (1) The Family History Questionnaire ascertained information allowing for the separation of participants into women with suspected hereditary breast cancer (HBC) and women with presumed sporadic breast cancer (SBC), (2) a subsequent survey included questions addressing participants' perceptions of their children's concerns about their breast cancer risk and participants' preferred information sources for their children about HBC.

Results: A total of 273 women returned both surveys. Participants' average age was 47.5 years; 91 percent were Caucasian, 7 percent were African-American, and 2 percent were Asian-American. A total of 231 (85 percent) participants reported having children. Of these, 82 percent of participants indicated concern about their children's breast cancer risk. A total of 127 (55 percent) reported expressions of concern from their children about their breast cancer risk because of the mother's breast cancer history. The mean age of children's reported expression of concern was 19.4 years (range, 6-38 years) for daughters and 18.6 years (range, 5-35 years) for sons. Of 257 respondents, 156 (61 percent) believed the appropriate age for their children's education about HBC to be the teens (up to age 18 years). Fewer (n=59) felt this to be 18-21 years, and a minority (n=12) felt this to be >21 years. When asked who should provide HBC information to children (more than one choice offered), of the 254 respondents, 214 selected the child's parent(s), 166 the child's PCP, 83 a gynecologist, and 48 an oncologist. Only 30 chose a genetic counselor and 7 a geneticist. There were no differences in responses when comparing those with presumed HBC with those with suspected SBC.

Conclusions: These data show concern among breast cancer survivors and their children about HBC and raise questions about the adequacy and availability of educational resources for daughters and sons of affected mothers.

Genetic Testing and Liver Transplantation in Japan: An Ethnographic Study of Familial Amyloid Polyneuropathy

Kaori Muto, Setsu Akutsu, Jiro Nudeshima, Shohei Yonemoto

Familial amyloid polyneuropathy (FAP) is a dominantly inherited, fatal, systemic, progressive amyloidosis. The major incidences of FAP are in Sweden, Portugal, and Japan. The purpose of this ethnographic study was to understand the present ethical, legal, and social issues surrounding FAP, specifically, predictive genetic testing and liver transplants. Liver transplants are available to patients with early-stage FAP to prevent further disease progression.

Method: The study was conducted in city Q between 1996 and 1999. In-depth interviews were conducted with 10 patients, 7 who underwent liver transplants, 4 at-risk persons, 6 family members, 6 medical professions, 6 nurses, 4 social workers and 5 reporters of local media. Documents such as diaries and letters were also reviewed. The data were analyzed using the principles of grounded theory.

Results: We found that patients and family members suffered from marriage discrimination, defined as a breach of engagement contracts, or marital separation based on rumors of a "family with an awful and cursed disease." Such discrimination caused patients to hide their disease to afford children the opportunity to marry. As a result, people with FAP experienced isolation within their community and refused health care services. Insurance and employment discrimination was not found. Available since 1984, predictive testing has been used by clients considering marriage. In 1990, the introduction of liver transplants served as the only hope for such clients. Transplants lessened marriage discrimination, but brought about other problems. Due to the lack of national legislation, some patients traveled abroad to acquire livers, which resulted in international backlash. The lay FAP organization was split between patients who could not afford to undergo transplants and patients who lost their identity as FAP patients as a result of undergoing transplants. Transplants between living donors have become more popular, but Japan's Transplant Act does not regulate such procedures. Liver transplants have changed the purpose and meaning of predictive testing. Testing is now used not only to detect the presence of the disease, but also to identify potential donors. We argue that regulation of liver transplants between living donors and ethical guidelines on predictive testing for the purpose of identifying donors are necessary.

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Matching Old Banked DNA with Current Knowledge: A Present Day Challenge

Linda E. Nee, MSW

Over the last two decades research groups banked blood and skin samples as a resource for DNA research. Large families with dominantly inherited Alzheimer disease (AD) were selected for research study, and now many of those families know the mutation that runs in their family. Small families were often not ideal research subjects, so many of those families do not know their unique mutation. This has presented a challenge for clinicians. For example, a request was recently received from a newly married daughter of a deceased woman who had been diagnosed with dominantly inherited AD. The mother's DNA sample was banked in the 1980s. The mother and her deceased sister were the last family members affected with dementia. The daughter is now faced with a situation of potentially wanting prenatal diagnostic testing regarding family planning, but such testing may be useless or present false negatives. Analysis of the mother and sister's DNA could give this family valuable genetic data and broaden their knowledge base for decision-making. Identifying a mutation could not only assist the family but also increase the value of the samples for research purposes. The commercially available diagnostic services, as far as I have been able to determine, test only fresh blood samples.

Updating stored DNA samples is of concern to researchers and clinicians working with families. Researchers want to continue the pursuit for mutations that might unlock the mysteries of AD. Updating samples for scientific purposes seemingly has greater sanction and financial backing at this time. Clinicians responding to the requests of families are faced with the challenge of finding the resources and funding to test stored DNA and a mechanism to activate the process. This is a challenge not only for AD clinicians but also potentially for clinicians working with other disorders as this present rapid pace of genetic discovery continues. The people/families we are trying to serve deserve to know the outcome of the research for which they consented to have their DNA studied. Their last chance to know today may have been banked 20 years ago.

Primary Care Providers and Genetic Testing: Are Health Care Providers Prepared to Implement Testing and Treatment Advances in Genetics?

Teresa Rhodes, BS, MS, LMSW, ABD

With the completion of the human genome sequence, during the summer of 2000, there has been a flood of new information. This growth is expected to continue for decades and will have a major impact on health care service delivery. Due to the health care delivery system changes that occurred during the implementation of managed care, the role of primary care providers has expanded. Primary care providers are often in the role of managing patient referrals. Consequently, they will have the responsibility of serving as the decision-makers and gatekeepers for genetic services. The largest percentage of genetic referrals and follow-ups are expected to be in the hands of primary care providers. It is very important that they are prepared to meet this demand.

This research project studies the preparedness of primary care providers in their ability to clinically implement genetic service in their practice settings through patient assessments, knowledge of when and where to refer for genetic services, and follow-up care. The project includes a comparison of differences between recent primary care graduates and those who graduated over 10 years ago. Additionally, the preparedness of primary care providers to develop and integrate clinical practice standards is compared in both metropolitan and rural communities. The findings are summarized into recommendations that are intended to be useful in the implementation of policy regarding education of primary care providers in topics that will enable them to better serve their patients.

An extensive literature review is included and covers related topic areas such as the need for future research regarding medical educational needs, recommended curriculum content changes, and continuing medical education concerns. While discussion is focused on primary care providers, options are discussed for enhancing the skills of other health care providers in dealing with the explosion of genetic knowledge and the resulting changes in health care services.

The Utilization and Impact of Forensic DNA Typing in Criminal Cases in Four Maryland Jurisdictions

David Wasserman, JD, Lois Tully, PhD, Victor Weedn, JD, MD, Joseph Peterson, DCrim, Valerie Prenger, PhD

This poster will report the findings of an ELSI-funded study of the routine utilization and impact of forensic DNA typing in four Maryland jurisdictions: Anne Arundel County, Baltimore City, Montgomery County, and St. Mary's County. The study addressed several questions on which there has been surprisingly little research: (1) How often, and in what offense categories, is DNA typing used? (2) What case, evidentiary, and defendant characteristics are associated with its use? (3) What impact does DNA typing have on outcomes of the cases in which it is used? The significance of DNA typing as an investigative and evidentiary tool is likely to depend on the proportion of cases where the identity of the perpetrator or the performance of a

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sexual act is at issue, on the availability of biological evidence in cases with those issues, and on the relevance to those issues of an association between the defendant and a particular residue.

The study examined all extant State Attorney's files for criminal cases in which DNA typing was performed through mid-1996. It compared the outcome, evidence, and other characteristics of DNA cases with non-DNA controls: cases involving similar offenses where biological residues were available but DNA typing was not performed.

A comprehensive coding instrument was used for abstracting information on each case, including arrest, disposition, and sentencing, demographic data, suspect's prior arrest record, confessions, prior acquaintance of suspect and victim, witness testimony, and laboratory results. Over 350 cases were retrieved, abstracted, and analyzed.

Significant associations were found between the use of DNA typing and several case and evidentiary variables, including weapon use, lack of prior acquaintance of victim and perpetrator, and the existence of circumstantial witnesses. Additionally, significant associations were found between case dispositions and both the use of DNA typing and specific DNA typing results. The poster will present these findings, as well as the results of several regression analyses now being completed. It will also highlight the impact of DNA exclusions — failures to match — on case disposition.

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