HUMAN GENOME EDUCATION

MODEL PROJECT*

A
Project of
Georgetown University
Child Development Center-UAP
and
The Alliance of Genetic Support Groups

PROGRAM

Ethical, Legal, and Social Implications of the Human Genome Project: Education of Interdisciplinary Professionals Meeting - Georgetown University June 10, 1996**

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Alliance of Genetic Support Groups

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DISCLAIMER

HUMAN GENOME EDUCATION MODEL PROJECT

Ethical, Legal, and Social Implications of the Human Genome Project

Education of Interdisciplinary Professionals

Meeting on June 10, 1996

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Ethical, Legal, and Social Implications of the Human Genome Project: Education of Interdisciplinary Professionals

Georgetown University - June 10, 1996

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8:45	Introductions and Overview of Meeting	Joan O. Weiss, MSW
9:00	Surveys: UAP Health Professionals Speech-Language Pathologists Nurses - ANA	 E. Virginia Lapham, Ph.D. Sharon Willig, MA-CCC Colleen Scanlon, JD, MS
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10:40	Coffee Break	
11:00	Roundtable Discussion of What organizations have done Plans, ideas for future	 Moderator: Chahira Kozma, MD
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Surveys

UNIVERSITY AFFILIATED PROGRAMS
(Human Genome Education Model [HuGEM] Project)

E. VIRGINIA LAPHAM, Ph.D.

We would like to share with you some of the findings of our HuGEM survey of health care providers in 53 university affiliated programs. The respondents included education specialists, psychologists, physicians, occupational therapists (OTs) and physical therapists (PTs), social workers, audiologists and speech pathologists, nurses and 49 others that included dentists, administrators, and rehab counselors.

They were a fairly well-educated group with the mean number of years in the profession 17.3. Fifty-four percent had doctorates. Unfortunately, there were not as many different cultural ethnic groups represented as we would have liked. Three per cent were African-American, 2% Asian-Pacific Island, 3% Hispanic, and 91% Caucasian.

The first question we asked was, "Had you heard about the Human Genome Project before volunteering to participate in this survey?" Sixty-one percent of the consumers had heard of the Human Genome Project. Fifty-two percent of health professionals overall had heard of the Human Genome Project. Seventy-nine percent of the physicians, 61% of nurses, 52% of social workers, 50% of psychologists, and 30% of speech-language pathologists had ever heard about the Human Genome Project.

Where do people hear about the Human Genome Project? The media is still our primary means of education on this topic.

We asked, "How much have you heard or read about genetic testing?" Twenty-nine percent of the consumers and 26% of the health professionals said that they have heard or read a lot about genetic testing.

One of the questions we asked was, "Do you provide clinical services to individuals and/or families with genetic disorders?" Most physicians responded affirmatively. We asked, "Have you ever had occasion to refer a patient or client for genetic testing?" Most of the physicians and many of the nurses made these referrals, as did social workers, audiologists and speech pathologists. Nutritionists were also fairly high, 56%.

We also asked, "Have you ever had occasion to refer a patient or client for genetic counseling? The numbers were very high for physicians and nurses. Audiologists, speech pathologists and social workers frequently made referrals for genetic counseling. Then we asked the question, "Have you ever had occasion to provide counseling about genetic concerns for any client or patient yourself?" Physicians, nurses, and nutritionists frequently provided this counseling. The audiologists and social workers also counseled clients about genetic concerns.

Fifty-four percent of physicians have had one or more courses in genetics, 35% of OTs and PTs, 33% of nutritionists, 26% of social workers, 21% of educators, 20% of audiologists and speech pathologists, and 24% of psychologists.

E. Virginia Lapham, PhD

The top priority for education for both consumers and health professions was coping with the new genetic diagnosis in the family. The second priority for the consumers and the third for the providers was treatment for genetic disorders, including gene therapy.

Other priorities included access to genetic information and genetic disorders and health insurance and staying informed about new developments in the Human Genome Project. Additional educational priorities were health care reform and the Human Genome Project, new genetic information and the legal system, and advantages and disadvantages of participating in family studies of genetic conditions.

Our HuGEM survey identifies the strong need for education of health professionals. We highlight the importance of the media in educating consumers and health professionals about the Human Genome Project and new genetic discoveries. We document and are writing about the discrimination experiences of consumers in health insurance, life insurance, and employment.*

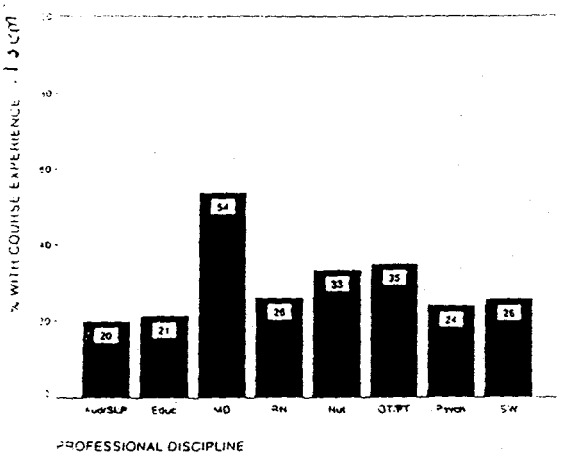
We gained new insights about similarities and differences between consumers and health professionals. The survey helped us establish priority topics for education and assisted us in developing new educational resources.

*(Editorial Note: An article, "Genetic Discrimination: Perspectives of Consumers", was published in the October 25, 1996 issue of Science.)

HUMAN GENOME EDUCATION MODEL (HuGEM) PROJECT

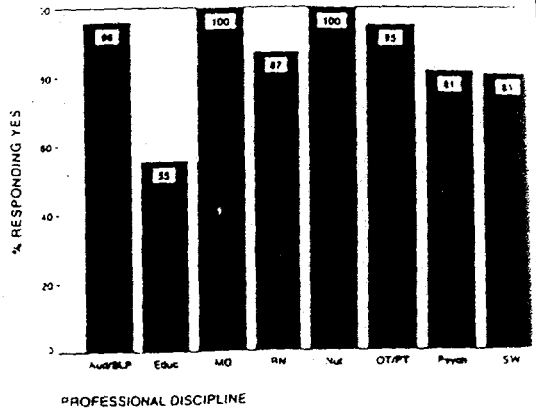
Survey of 329 Health Professionals in 53 UAP's

COURSE EXPERIENCE IN GENETICS



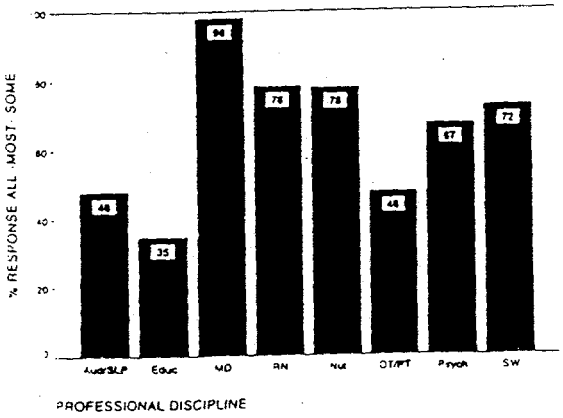
PROVIDE CLINICAL SERVICES

PERSONS WITH GENETIC DISORDERS

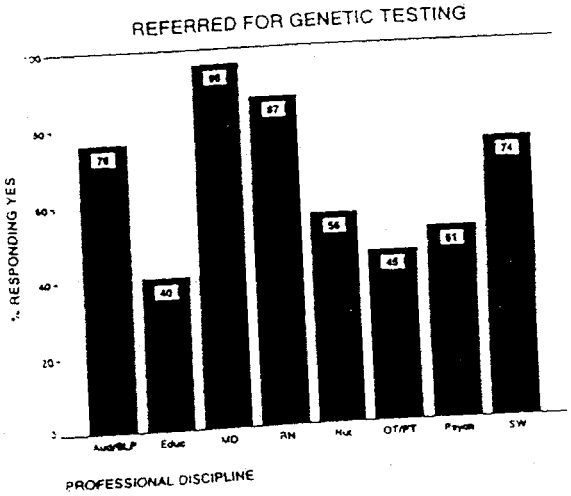


DISCUSS GENETIC COMPONENT OF

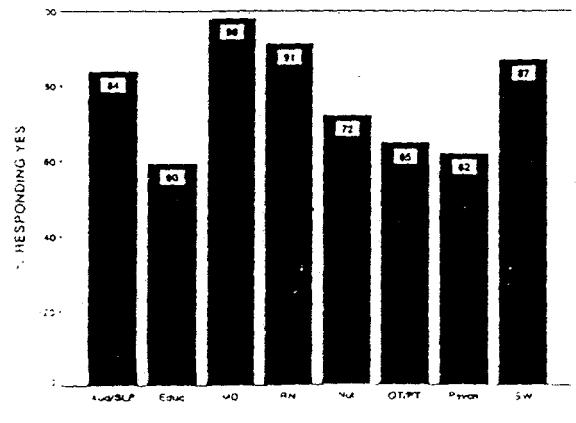
MEDICAL/DEVELOPMENTAL PROBLEM



REFERRED FOR GENETIC TESTING

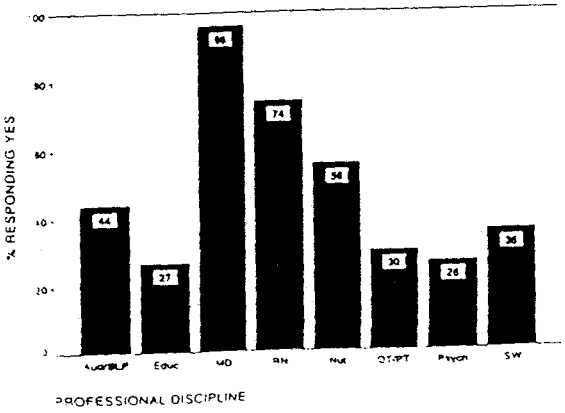


REFERRED FOR GENETIC COUNSELING



PROVIDED COUSELING ABOUT

GENETIC CONCERNS



HUMAN GENOME EDUCATION MODEL (HuGEM) PROJECT

Survey of 329 Health Professionals in 53 UAP's

Professional Discipline:	#
Audiology/Speech and Language Pathology	25
Education	56
Medicine	52
Nursing	23
Nutrition	12*
Occupational Therapy (18) Physical Therapy (22)	40
Psychology	54
Social Work	31
Other (Administrators, Dentists, Counselors, etc.)	36

Mean number of years in professions: 17.3

Degrees:	
Ph.D.	35%
M.D.	15%
Other doctorate	5%
Master's degree	37%
Bachelor's degree	4%
Other	5%

Employment Status:	
Full time	86%
Part time	14%

Culture/Ethnicity:	
African American	3%
Asian/Pacific Islander American	2%
Caucasian American	92%
Hispanic American	3%

Religion:	
Roman Catholic	21%
Protestant	36%
Christian (Other)	9%
Hindu (1)	
Judaism	13%
Muslim (1)	
Other	2%
None	18%

Genetic disorder/disability in the family: 29%

*11 additional questionnaires were completed by nutritionists recruited by K. Camp and are included in the bar graphs on the other side.

Surveys

SPEECH-LANGUAGE PATHOLOGISTS

SHARON WILLIG, M.A.-C.C.C.
Georgetown University Child Development Center

In 1995 we received responses from 329 health professionals in the United States who were surveyed to investigate their knowledge of new genetic advances and what implications these advances might have on their practices. Of this group, 18 were speech-language pathologists (SLP) and four were audiologists. All respondents worked in university affiliated programs (UAPs) generally associated with medical centers and provided diagnostic and treatment services to individuals with disabilities. Nearly half of the SLPs and audiologists had doctorate degrees and therefore did not constitute a representative sample of the profession. According to the American Speech-Language and Hearing Association (ASHA), only 4% of the SLP membership hold doctorate degrees.

To achieve a more representative sample, the same survey was distributed to 350 public school SLPs in the suburban D.C. area. Public School SLPs were chosen because they represent 53% of the ASHA SLP membership. Ninety-six surveys were returned.

Only 6% of the ASHA membership report they belong to a racial or ethnic minority group. As this information is voluntary for ASHA members, it may not be representative. However, 29% reported minority membership in the local sample. Educational status and gender differences appeared similar for both groups. The local SLP group had more years of professional experience on a whole than the ASHA group.

What is the significance of surveying SLPs?

Facts:

*42 million Americans have some form of communication disorder (ASHA, 1996).

*Most children with developmental disabilities have speech-language disorders.

*Over 400 known genetic syndromes are characterized by hearing impairments (McKusick, 1994).

*Cleft palate, a common oral anomaly affecting speech production, is associated with 139 syndromes (Gorlin, Cohen & Levin, 1992).

*Fragile X is the most common inherited form of mental retardation and speech-language problems are a primary characteristic.

In looking at what SLPs know of the Human Genome Project and how they receive their information, look at the graph, "Surveys - Public School and UAP". The dark line represents the local sample, the light line represents the UAP sample of SLPs and audiologists. Focus on what percentage indicated they worked with individuals with

genetic disorders (82%), how many referred for genetic counseling (15%), and where they primarily obtained their information on the Human Genome Project (media). Very few public school SLPs got information from lectures or workshops.

Most SLPs agreed the most on the importance of finding the genetic causes of Alzheimer disease, breast cancer and autism.

Some of the issues which public school SLPs would like addressed in continuing education programs are treatments for genetic disorders, who has access to genetic public information, and coping with a new genetic diagnosis in the family.

HUMAN GENOME EDUCATION MODEL (HuGEM) PROJECT

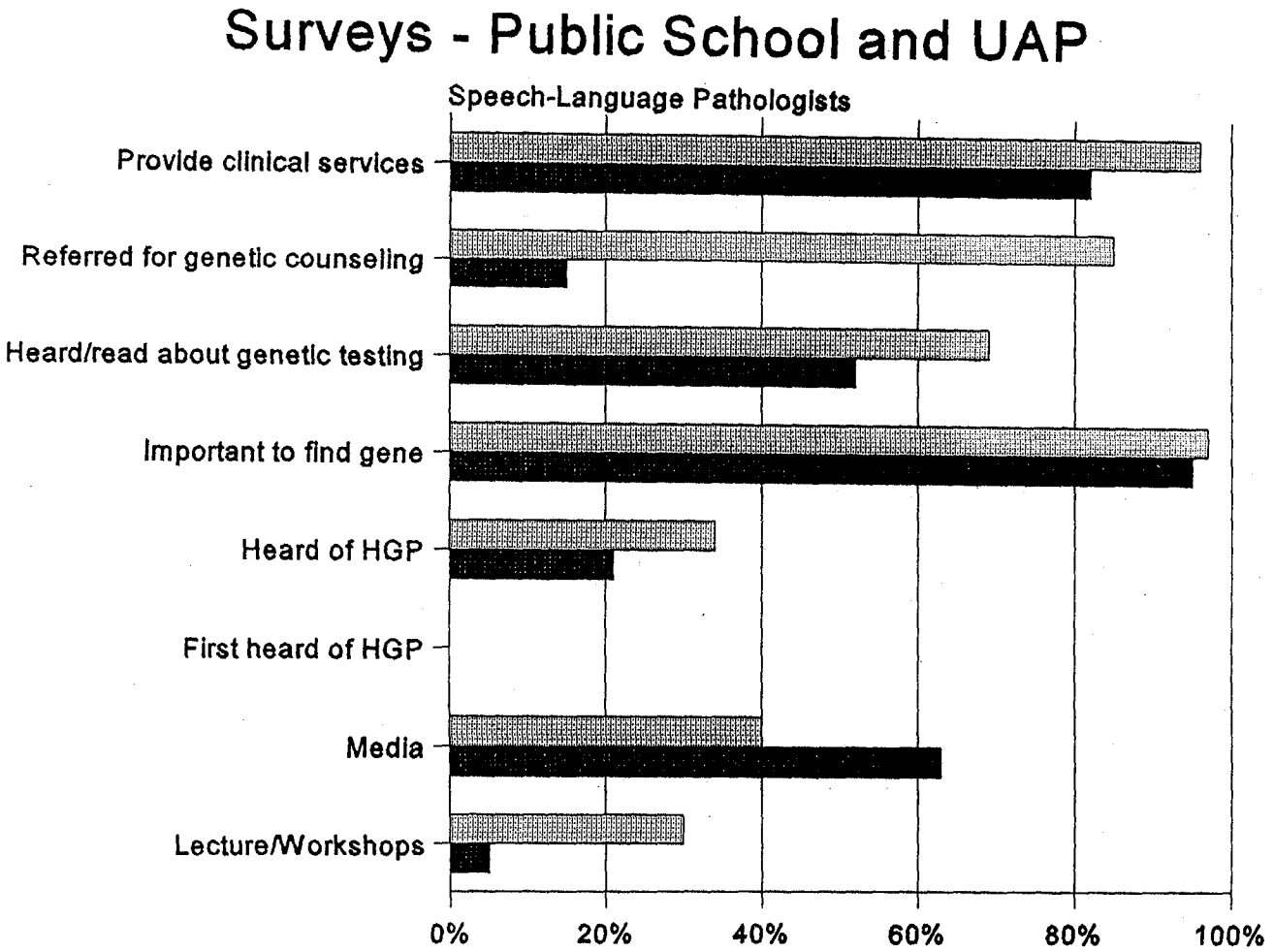
SURVEY OF
PUBLIC SCHOOL SPEECH-LANGUAGE PATHOLOGISTS

N = 96 (350 surveys were distributed)

Respondent Categories:

	Metro Area	ASHA
Ethnicity:		
African American	25%	(6% reported
Asian American	1%	other than Caucasian)
Caucasian	71%	
Hispanic American	1%	
Native American		
Other	2%	
Education:		
Bachelor's Degree	5%	3%
Master's Degree	91%	93%
Doctorate	4%	4%
Professional Experience:		
Mean:	16 years	12 years
Median:	17 years	10 years
Range	1 - 36 years	
Gender:		
Female	94%	96%
Male	6%	4%

HUMAN GENOME EDUCATION MODEL (HuGEM) PROJECT



SURVEY OF PUBLIC SCHOOL SPEECH-LANGUAGE PATHOLOGISTS

It is "very important" for scientists to try and find the genetic cause or gene(s) for:

	Public School SLPs	UAP SLP/Aud
• Alzheimer's Disease	90%	81%
• Breast Cancer	90%	73%
• Autism	88%	89%

SURVEY OF PUBLIC SCHOOL SPEECH-LANGUAGE PATHOLOGISTS

Genetic Course Topics Considered "Very Important" for Inclusion in Education of SLPs.

	Public School SLPs	UAP SLP/Aud
• Treatments for genetic disorders including gene therapy	83%	73%
• Who has access to genetic information	81%	81%
• Coping with a new genetic diagnosis in the family	81%	92%

AMERICAN NURSES ASSOCIATION (ANA)

COLLEEN SCANLON, J.D., M.S.

The American Nurses Association received grant funding from the Human Genome Project, ELSI Branch (1993-1995), to look at the unique experience of nurses and practice and attitudes related to genetic advances that are impacting them in clinical practice. We focused exclusively on nurses who are in the clinical arena as compared to looking at some of the experiences that educators, administrators or non-clinical services persons might have.

The grant was a two-pronged project, the first part of it focusing on the development of a survey which would help us obtain some information about what was actually occurring in the clinical reality of nurses. We were looking particularly at their experience related to the management of genetic information, i.e., eliciting it, transferring it and using genetic information in the care of patients and families whom they served. The second part of the grant was really to focus on the development of some resources that might be helpful as they continue to deal with this area of practice.

Not surprisingly, we found that nurses were generally quite ill-prepared to deal with this aspect of clinical practice. We surveyed a thousand nurses from across the country in a variety of clinical settings, the majority, staff nurses. But some were clinical specialists, nurse practitioners and nurses in leadership positions within the clinical arena, including head nurses and nurse managers.

The majority of nurses had had no formal course in genetics, nor did they feel that genetics content was adequately taught throughout other content areas of their professional education. They had significantly perceived their confidence and their competence in the area of genetic services as markedly less than other areas of nursing practice that we would consider important, whether it be communication skills, interpersonal skills, family dynamics, or anatomy and physiology.

They realized that they really didn't have the competence in this area. It was also clear that as we asked about particular practices and particular interventions that might be in the scope of their practice related to patients and families with genetic concerns, the more complex the intervention, the greater the sense of lack of competence. The more basic the intervention, the more confidence they had.

The majority of the nurses surveyed said they encountered patients and families who had genetic concerns. Either they wanted information about a genetics problem, wanted a referral, or they were actually going to be engaged in genetic testing and screening.

We also looked at some of the attitudinal issues and concerns and some of the ethical dilemmas. We used some case studies and tried to find out how nurses would respond if they were confronted with some of these scenarios in their practice.

The majority of our respondents had never heard of the Human Genome Project. Only 17% had heard of it. Yet, the great majority of them also realized that this emerging area of genetic science and biotechnology was impacting their clinical practice. And there was a strong willingness to engaged in education programs. The nurses believed that there should be mandatory education in professional education and practice. They also believed that continuing education should be made available to clinicians.

Although there was not a lot of awareness of the Human Genome Project, there was a very strong sense of awareness and a commitment to what was actually occurring in their environment, along with a desire to participate in preparing themselves to more adequately deal with that dimension of practice.

The second part of the grant was to look at these results and try to develop some resources for nurses. We developed a booklet called Managing Genetic Information: Implications for Nursing Practice, which provides some introductory materials and a look at some of the issues such as informed consent, privacy and confidentiality, truth telling and disclosure and non-discrimination issues.

We have received a tremendous amount of positive feedback from that publication. However, as you are certainly aware, it's just a very preliminary step in what is a huge undertaking for all of us. We need to be much more aggressive about educating health care professionals. The question is how best to do that. There are numerous dialogues going on now about different alternatives. I hope we will come out with more direction for all of us.

Presentation

HUMAN GENOME PROJECT UPDATE
SIGNIFICANCE TO HEALTH PROFESSIONALS

FRANCIS S. COLLINS, M.D., Ph.D.

Priority issues need to be addressed about how we are going to deal with this knowledge deficit on the part of many health professionals. It is a knowledge deficit which is increasingly one that those professionals are aware of and are seeking solutions to. Breast cancer is an example of a situation where gene discoveries that have sprung out of the Genome Project are already now presenting clinical dilemmas for health care professionals requiring their increasing sophistication in areas of genetic testing and counseling.

The HuGEM survey already indicated that most people get their information about genetics from the media. You can hardly pass the newsstand these days without seeing some article of this sort. Recently, U.S. News & World Report in their issue on health, focused on genetic testing for cancer as the topic that they thought was of highest interest to the public in 1996. In a recent issue of Glamour, on the list of seven things to worry about now that you never had to worry about before, down there at the bottom is genetic testing for cancer, how to handle the bad news. The implication is that you only get bad news from testing and that there is no way you ever get good news. Obviously that's not true.

Notice, by the way, that another issue relevant to our discussion today is getting sick and having your insurance company decide it's a pre-existing condition. Of course, if you accept the idea that virtually all conditions are genetic, and virtually all of us are going to get sick from something, then we all have pre-existing conditions written into our DNA. And that is a very serious issue.

This is an important concept in the thinking process about medical disorders. I would challenge you, regardless of the field you work in, whether it's nutrition or speech pathology or whatever, to come up with examples of conditions that you deal with which are truly non-genetic. It's very difficult to do so. Trauma is perhaps an exception. But for most medical conditions, be it cystic fibrosis or diabetes or even AIDS, we are increasingly becoming aware that there are genes that contribute to who gets sick and how sick they get. Even a disorder which is essentially an infectious illness has a genetic component.

Just to sum up the progress scientifically of the Genome Project, things are going well. We have the genetic maps done. The physical maps are approaching their completion. The DNA sequence is just beginning to ramp up in seriousness. I believe that we will reach our goal by 2005 at the latest.

What have the consequences been already for an understanding of medical diseases as a consequence of the success in gene mapping? They have been substantial. The first positionally cloned gene was found exactly 10 years ago in 1986. And it was another three years until any more were found from those first three in '86.

The Genome Project in 1990 began providing the kinds of maps that people needed to go from families to fighting the specific gene. Then things really began to break wide open. In 1993, there was beginning to be an explosion in the number of genes that were being found. Increasingly, gene mapping is focusing on more common disorders such as breast cancer and Alzheimer's Disease.

Clearly the progress in identifying genes that are responsible for medical conditions is rather spectacular. Are we ready for that? Are we ready for the consequences of that in the clinic? And for that purpose, I'd like to give the specific example of breast cancer. If this is not a condition you deal with in your particular specialty, then I ask you to think broadly about this. This is only an example. The same kinds of issues that I will refer to here and the kinds of cases that I'm going to tell you about could be also brought up for conditions across the range of medical problems that you encounter in your own practice.

After all, breast cancer until five years ago was largely considered to be a disorder where the genetics were going to be so complicated and potentially so weakly contributing to risk that they would not be something that would invade medical practice any time soon. And here we are now in 1996, facing the likelihood that commercial testing for BRCA-1 and BRCA-2 may be offered to large numbers of people before the year is out.

I think many of us are deeply concerned that we're not prepared in terms of the counseling and education that need to go along with that. Health care professionals, for the most part, have not been provided with the information that they will need to participate effectively in that kind of a genetic medicine.

If you have a mutation in BRCA-1 and you're a woman, your risk of getting breast cancer is quite dramatically elevated. This risk factor, however, is based on families where large numbers of individuals have had breast or ovarian cancer. So there is some possibility that there's a bias here. And the risk may not be quite this high.

But in individuals where there is a strong family history of breast cancer and where a mutation is found in BRCA-1, the risk of getting breast cancer approaches 90% lifelong. The risk of ovarian cancer, which is not shown here, is about 45 to 50% for those women as well. Obviously, the question that comes to mind is: Do we have an obligation as health care professionals to find the roughly one in five hundred women who is in this category and warn her of this risk in order to give her the opportunity to take advantage of prophylactic procedures, be they surgical or medical, in order to try to reduce that risk of dying of a terrible disease?

In order to answer that question, there are many things that we wish we knew that we don't know. Are the interventions that are available, such as mammography or more drastic surgical procedures, for both breast and ovarian cancer beneficial? How beneficial are they? What's the likelihood that they will fail? Furthermore, we'd like to know what is the likelihood that somebody who goes through that kind of testing and gets a result will have that information used against them so that the risks associated with the procedure turn out to exceed the benefits. I am talking about discrimination.

We don't have terribly good answers to any of those questions right now, which is why many people feel that this kind of testing still belongs in the research arena. But great pressures are coming to bear on that. It seems reasonably likely that testing will go on in an expanded way over the course of the next few years. We need to be ready.

BRCA1 was first mapped to chromosome 17 using genetic markers back in 1990. And then the gene was identified. Like many genes, BRCA-1 has the potential to be misspelled in many different ways. This is not surprising. This is the usual thing in genetics. When you find a genetic condition and you identify the gene that's responsible, when you look at one family you might find that they have a glitch in another part.

The question, I think, for many genetic tests is this: Is having information, seeing into the future in some way or another, going to benefit you or hurt you? And that's what people want to know.

Here's an example of a family recently counseled at the NIH. There is a grandmother with breast cancer at 46 who died of metastatic disease. Her daughter developed ovarian cancer at 39. It turned out that there was a mutation in BRCA-1 in this family affecting the grandmother and her daughter. And, therefore, the other two sisters, age 44 and 38, now have the ability to find out their risk if they want to know. They each have a 50% chance of having inherited the altered gene from the grandmother.

Do they want to know? The 44-year old who has a daughter of her own is currently undergoing a regular screening anyway because she's been tipped off to her own risk by her family history. She was very anxious to find out and came forward, was counseled, went through with the testing, and turned out not to have the mutation which affects her sister. In that situation, there was both good news and bad news. For her, that obviously reduced her risk and reduced her daughter's risk.

What has happened, however, is that she and her sister, who had been close prior to this time, now have a rather strained relationship. This difference in their genetic status has turned out to be very significant in terms of how they deal with each other. The 44-year old, the older daughter ... older sister ... feels uncomfortable talking about this topic, with some "survivor guilt." The middle sister, feels like she can no longer talk about her own feelings about the genetic aspects of ovarian cancer with her older sister because it doesn't apply to her.

Family dynamics can change substantially, and many times, require counseling and intervention. Those of you involved in social work and psychology, I think, will see a fair amount of this.

The youngest sister, the 38-year old, had had a very difficult previous experience with HIV testing. She, as a young adult, had engaged in high-risk behavior. She had eventually decided that she was potentially at risk and had gone through anonymous HIV testing, remembering with great pain that interval between being tested and getting the result.. Despite a great deal of pressure from her other two sisters to be tested, she decided she just didn't want to know.

An important point is that her decision needs to be supported and that there are good reasons why some people choose not to have this information. The general gold standard of genetic counseling is to be non-directive and, in this situation, to provide information about risks and benefits and to help that person think through the outcomes of being tested or not being tested. Then it is important to support the decision as long as that decision is well thought through. For this sister, this seemed to be the right decision, although this created considerable strife in the family because both of her older sisters were convinced that she was making a terrible mistake by not
being tested. All of these issues become complex and require a good grasp of psychosocial issues as well as of DNA.

We cannot give any assurances that if a woman tests positive for BRCA-1, that information will not somehow find its way into the hands of her health insurer, despite the best efforts to keep that from happening. If that happens, a woman might very well find herself uninsurable. Obviously, one would like it not to be the case that this kind of situation becomes the factor that causes someone to make a particular decision. However, at the present time, all too often that is a very serious part of the decision-making process.

I am happy to say the National Cancer Institute, recognizing that we are in a difficult situation where these kinds of barriers would in many ways mitigate against opening the doors wide to offering clinical testing on a large scale, has decided to set up a cancer genetics network to increase access to testing, but continue to have the testing done through approved research protocols that have been overseen by an institutional review board.

I think this will be a nice pilot project to see how we can include more health care professionals in the business of providing genetic medicine services to their patients, without losing the opportunity to collect the information as part of a research project to find out who comes in, who decides to be tested, and who doesn't. For the people who are tested, what results are obtained? And then what happens afterwards? Five years from now, hopefully, we'll have better answers to these questions.

The Cancer Institute has been very thoughtful in deciding to invest some of their resources in this model. But I don't know that we can do that for all genetic diseases. Maybe we can do that for breast cancer and colon cancer because it's such an urgent matter. But we're going to have to have other solutions, I think, for the long list of diseases that are coming along quickly, for example, diabetes and prostate cancer.

The Ethical, Legal and Social Issues (ELSI) Program has been set in place to try to deal with the long list of dilemmas that come out of the successes that are being now realized in genetic discoveries. It is appropriate for once for a scientific project that's attempting to make these discoveries to also take this kind of responsibility for figuring out what that's going to lead to.

And that's what the ELSI Program is all about. Five percent of the Genome Project's budget has been devoted to this program from its outset.

Privacy and fair use of genetic information is of great concern to the public. The HuGEM survey showed that. A Harris Poll showed that as well. Eighty-six percent of a statistically valid sample of the U.S. population indicated concern about health and life insurance companies using genetic information against them.

Another effort which has spun out of the ELSI working group, the group that has been working since the outset of the Genome Project on these issues, is a national task force on genetic testing. This group has recently issued a draft series of principles, and those are available to you through the Internet. This task force has a range of different participants which includes consumers as well as primary health care providers, biotechnology companies, geneticists, insurers and government agencies. They have been working very hard to come forward with not only principles but implementation of those principles by the end of this year. Much of that does deal with health care professional education.

The public and professional education part of the ELSI Program has been in many ways reflective of what's going on in the grants program. We have funded 21 education projects through NCHGR. There is among them a couple of surveys. HuGEM is one of the ones that's funded through this effort.

While you might be pleased to learn that 86% of the public is sensitized about genetic information and health insurance as shown in a recent NSF survey, only 9% of their sample could define what DNA is. So we have a distance to go.

The concern I have is that the time table for getting information effectively into the hands of professionals and the public is really getting telescoped. The ELSI Program recently reviewed the education projects that we're supporting and decided that, from the point of view of the National Center for Human Genome Research (NCHGR), our primary target should be health care professionals.

We've been trying to fund projects that dealt with curriculum development for K-12, projects that developed TV programs for public broadcasting, projects that looked at curriculum development in colleges. And I think the conclusion was that we have limited resources. The most pressing issue is to educate health care professionals. That's where most people are going to get the information when they are really at that teachable moment when it's going to matter to them.

What we want to do, if we really want to be effective here, is to provide a setting where when somebody needs the information, they have access to it. Deluging people with information about genetics when they are not really interested right then is not going to have a long-term effect. And for most people, when they're really interested, it is because there's something relative to themselves or their families. They are going to go to a health care professional and ask, what should I do? My brother has this condition and I heard something in the newspaper that it might be genetic. Is there a test? Should I be tested?

If we don't have a situation where the health care professional can answer the question or know how to get the answer, we're in trouble. And we are, I think, rapidly approaching that point. Thinking about this over the course of the last year or more, we at NCHGR have come to the conclusion that what we really need is a coalition of health care professionals devoted to advancing education in genetics.

We have, over the course of the last three or four months, been having a series of very productive discussions with the American Medical Association and the American Nursing Association about establishing this coalition. This coalition would have a steering committee which would be consisting of leaders from health care professional organizations that have a major stake in this business -- getting genetic information into the hands of their members so that they can provide good genetic services. A membership of this coalition would be much broader and basically involve all organizations that represent professionals involved in medical and psychological care of patients. That coalition hopes to have its first steering committee meeting in the relatively near future.

We need to try to basically co-opt the enthusiasm of the leadership of the organizations that represent health care professionals. And I'm talking about all of you. Rather than the other model where the geneticists stand up like I'm doing now, shake their finger at you and say, You should know about this stuff or you're going to get in trouble! That doesn't really work well.

If we're going to succeed at this, I think we all have to band together. The issues that an occupational therapist or a social worker or nurse or a physician are going to need to get familiar with are all related to each other and they shouldn't be thought of in isolation but rather in a group. If we can come up with models to get that information effectively into the hands of people who are out there in practice, as well as integrating that effort into curricula, that's where we need to be.

The goal of this coalition is to achieve that. It is still in its early stages. Consumers will be a very important part of that coalition. I'm happy that the Alliance of Genetic Support Groups has indicated their enthusiasm for it, as have the American Society for Human Genetics and the American College of Medical Genetics representing the genetic .... medical/genetics professional community who will need to be heavily involved in providing information. I am also pleased about the enthusiasm from the National Society of Genetic Counselors and the International Society of Nurses in Genetics, all of whom have much to contribute to such a coalition.

I think that's where we need to go. And I think we need to do that pretty quickly. The consequences of gene discovery are often first in the diagnostic arena, and may, in many instances, as hopefully will be the case for breast and colon cancer, lead to good preventive medicine strategies. But the ultimate goal is gene therapies and drug therapies. Unfortunately, time does not allow those things to happen as quickly as you might want them to. You have a lot of very difficult science to follow up cloning a gene before you have come up with a therapy that's going to succeed. And for many diseases we're going to live in that window between effective diagnostics and effective therapeutics for the next five or ten years. We really have to struggle with that.

Wayne Gretzky, the hockey star, said that the reason that he's so successful in his particular sport is that he skates where the puck is going to be, not where it is now but where it's going to be. That's what we should all be trying to do today.

ROUNDTABLE DISCUSSION SUMMARY

American Academy of Pediatrics (Franklin Desposito, MD)
AAP Genetics Committee - Distributes the American Society of Human Genetics' published general principles and medical student core curriculum in genetics (1995) and their EduShare Report of 139 North American medical schools surveyed as to their courses in genetics; provides educational materials and seminars on genetic topics for practicing pediatricians as part of their educational and continuing medical education mission; develops health supervision statements for specific genetic disorders (7 developed and published to date).
American College of Obstetrics & Gynecology - Has a newly formed Committee on Genetics which publishes technical bulletins and practice parameters for obstetricians.
Maternal and Child Health Bureau - Funded 9 Genetics in Primary Care SPRANS Awards in FY1995.

American Dietetic Association (Ronni Chernoff, PhD, RD).
Currently, the ADA has no genetics education program for its 50,000 members. Steps are being taken to develop education in genetics.

American Medical Association (Michael J. Scotti, Jr., MD)
The AMA was founded on the education of physicians regarding standards, quality of practice and ethics. Currently two products in genetics are available: a) a videotape on Fragile X from their ethical series; b) education packet for high schools.

American Occupational Therapy Association (Penny Kyler-Hutchinson, MA, OTR-L)
AOTA is in its infancy in developing areas in genetics. The philosophy is not to glamorize one genetic condition over another. They do not know how much their 56,000 members know about genetics. A curriculum in ethics which includes genetics is being developed. Their dissemination of genetic information can be made through their many publications, weekly e-mail message to leaders in the field, workshop over the internet, semi-annual and annual conferences.

American Nurses Association (Colleen Scanlon, RN, MS, JD)
The ANA is an umbrella of 2.2 million nurses which disseminates information already gathered. Within the umbrella 70 specialty groups, such as the International Society of Nurses in Genetics (ISONG) and Oncology Nurses, have their own programs. The ANA is looking at its core curriculum and trying to find a time slot for genetics. At its 100th year meeting June 17, a session is being conducted on genetics. The ANA is on the steering committee for the Coalition of Professionals for Genetics which is being established.

American Psychological Association (Jo Linder-Crow, PhD)
The APA was at the conference to listen and to learn because they need solid resources in genetics for their members. They are looking forward to collaboration with other professional organizations. They have a strong model in their AIDS Continuing Education (ACE) project which might be helpful.

American Speech-Language-Hearing Association (Diane Paul-Brown, PhD, and Evelyn Cherow, MS)
ASHA is a credentialing association that encourages incorporation of information about genetic disorders in pre-service training in communication disorders and science. They offer continuing education units required by most states for licensure and develop their own educational programs in genetic disorders in communication disorders. In 1991 a day-long conference "Genetics: Progress and Promise for Communication Sciences and Disorders" was held. At the 6th Annual Research Conference November 19-20, 1996 in Seattle, Washington "Genetics: Communication Sciences and Disorders" with four workshops will be featured. Teleseminars, annual conventions and several scholarly journals keep their 85,000 members informed.

Council on Social Work Education (Donald Beless, PhD)
They do not have a lot to report. Dr. Julia Rauch at the University of Maryland and Dr. Susan Taylor-Brown at Syracuse have developed models for genetics education in social work. In 1995 one west coast and one east coast conference included a workshop on genetics. Very few articles on genetics have been submitted to their journals. They are pushing interdisciplinary approaches to working with clients.

Department of Energy (Daniel W. Drell, PhD)
The Department of Energy awards grants which are reviewed once a year. They are looking for innovative projects which can have impact beyond the intended audiences. Grants have been funded on intellectual property, non-traditional inheritance, a pamphlet for junior and high school students entitled "Your World," and a Human Genome Project primer now on the world wide web. The "Human Genome News" is free and is published every 2 or 3 months. It is also on the internet.

National Association of Social Workers (Ann Johnson, PhD)
A session on genetics was held at the NASW International meeting in 1994. Dr. Ann Johnson and Dr. Susan Taylor-Brown are drafting a policy statement and a clinical practice update which will include the Human Genome Project. In their courses taught at Catholic University and Syracuse University, genetic issues are incorporated into their inter-generational lectures.

National Center for Human Genome Research (Elizabeth J. Thomson, RN, MS)
Of the 110 grants funded by the NCHGR, 105 are educational projects. They are looking to fund developing programs, particularly in professional education. The Center has increased interest in the ethical, legal, and social implications of genetics around NIH. They have collaborative projects with the Food and Drug Administration and with the Center for Disease Control. One-half of their budget is in clinical areas and 20% of their budget is spent on educational projects.

National League for Nursing (Beverly Raff, PhD)
The mission of the League is for education, practice and research and emphasizing consumerism, community and diversity. They are politically active and hold an annual conference.

National Rehabilitation Association (Ann Tourigny, PhD, CAE)
The association is primarily interested in training for employment, and the counselors' training is spotty and inconsistent. They have an internationally respected journal and they hold 3 national conferences and 72 regional meetings annually. Their future plans would be to include genetics in their continuing education.

Rehabilitation Information & Technical Services (Janie Scott, PhD)
This organization is not doing anything specifically on genetics. The University of Baltimore has a legal and ethical program and is developing a course in healthcare ethics. Genetics is also expected to be included in the women's issues in healthcare courses.

Uniformed Services University of the Health Sciences (Edmund G. Howe, MD, JD)
Dr. Robert Murray, Director of Genetics at Howard University, teaches a summer course on genetics. Dr. Howe chairs the panel on ethics for all the armed forces, chairs the Institutional Review Board at the USAF, and is editor-in-chief of the magazine "Clinical Ethics"
No articles on genetics have been submitted to the journal. An annual conference on research and genetics is to be held in the near future.

Panel

PSYCHOSOCIAL ISSUES

LARRY ALLEN
Virginia Sickle Cell Awareness Program

My wife, Michelle and I have three children. Two of our children have forms of sickle cell anemia.

For the past eighteen years, we have struggled to keep our children alive and as healthy as possible, both physically and emotionally. We have fought to keep them insured, to keep food in the house, and to keep our utilities on. We have come through bankruptcy and loss of employment as a result of their illness. And as if having sickle cell is not enough, our daughter is facing open heart surgery at Hopkins at the end of this month.

Our initial experience with insurance companies started approximately eighteen years ago when the reasons for denial included "pre-existing condition" and "experimental treatments". If there was payment, the amount would be reduced drastically. As a result, by the time our daughter was five, we had already experienced bankruptcy.

Today, the insurance companies deny or reduce payments by saying, "the hospital did not follow their admissions procedures", or by holding the bill for six months to one year, after which the insurance company says they are not legally responsible for the bill. For example, in 1993 we almost lost our youngest son due to complications with his liver and gall bladder. His stay in the hospital came to approximately $30,000. For one year we received no bills, no letters from the hospital or insurance company stating there was a problem. Then one year and three months later, we began to receive statements from Blue Cross stating that they would not pay any of the bill and that we were responsible for the entire amount. We also began to get threatening calls from the hospital regarding payment on their bills.

Our calls to Blue Cross were very insightful. The first thing the customer service representative told me was that after one year, the insurance company was not legally responsible. It did not matter that we were not made aware the bill had not been paid. All they would promise us was that they would review the bills again. They informed me that the hospital did not follow their admission policy and that was the reason for delay, although the hospital stated they had done everything they were supposed to do.

That summer, while still working on this problem, we were asked to ride the bus and participate in the Health Security Express Project which was trying to get universal health care for all. It was during this trip we told our story to a local newspaper reporter who contacted Blue Cross. Blue Cross told him we were perpetuating a fraud and that they had paid all of our bill. My wife had brought copies of all the bills and letters, and we were able to fax them to the reporter who then called Blue Cross. This time, however, they stated there was a communication problem and they would get in touch with us about the bills. By the following day, the bills were paid. To make matters worse, they told the reporter that the bill had been paid months earlier and they had mistakenly forgotten to notify us. This happens every day - to someone.

Unfortunately, families such as ours often reach a point where we must literally start all over again. Employment is a major problem at times. We always notify them of our family condition. However, everyone is not as sensitive as they could be. Some people don't understand what it is like to care for someone with a chronic illness. Sometimes you face ridicule for placing your family/child before your employer. That has always troubled me, but we have learned from people like this. We recognize them very quickly nowadays.

I realize I cannot speak for each family that has a genetic disorder - I can speak for some. As different as each day is for all of us, there is always a constant for us. When you have lived in fear as long as we have, each day brings about doubts and apprehension. Your successes, no matter how great, are short-lived due to the constant pressure you are under. You have no time to savor the happiness, not time to appreciate life, no time to release the fear. When you feel you have met your match, not much matters any more. When you have swallowed your pride, for some of us, there is nothing else to fight for.

We were brought up to believe that if you were good and worked hard, you had a right to happiness. One thing we have found is that for many people, suffering is one promise life always keeps. If happiness comes, we know it as a precious gift which is ours only for a brief time. We realize we are not alone with these feelings, and we can't forget the other families living this American Nightmare.

Panel

PSYCHOSOCIAL ISSUES

MARY BETH BUSBY
FRAXA Research Foundation

My children are not ill. They have Fragile X Syndrome, which, as I think most of you know, is the largest inherited cause of mental retardation. They each have an IQ of about 55. So, although they are not sick, they are very definitely limited in their expectations.

I just hope that the awareness of Fragile X will gradually rise. I learned about it in 1982, at which point my boys were already teenagers. And I think the only thing that makes my story unique is the family situation that evolved. I made the mistake of telling my brother about Fragile X before I told my parents. He reacted so strongly, and I guess many would say so strangely, that he absolutely refused to have me tell my parents about this. They, to this day, have never heard the term Fragile X. Although they know their grandsons are retarded, they have no idea why, and never will, clearly.

I was just thinking, if we could just imagine a world in which we didn't have to deal with family dynamics, wouldn't it be 'loverly." You know, you take a family where everyone has brown hair. And you have all these big family reunions and everybody has brown hair. Then there comes along this kid with red hair. People say, "Well, where did he get that?" Then if that redheaded kid turns out to be President of the United States, people in that family say, "You know, I always knew that kid stood out. I always knew there was something really a little different." But if that redheaded kid turns out not to be okay in any way, then those family members say, "Well, you know, I always knew there was something a little bit strange about that kid."

Families want people all to be alike, especially within their own family. And I think that the only thing I have to contribute to this group, as professionals, is to tell you that when you deal with families who have a child with any genetic defect, you need to be very careful in counseling those families about dealing with their families. I think you need to say to this mother or the father, "All right, who are going to tell first in your family?" "How are you going to do this?" "When are you going to do this?" 'When' is very important.

I think that at the time you learn about the diagnosis is not a good time to deal with family members. And, if I were a counselor, I would counsel that person to have a cooling-off period because it is truly devastating to be told that you are the cause of your child's defect. At the time I first learned about Fragile X, I was so dumb that I didn't even know that the boy could only get an X chromosome from the mother. When we went into this session with the geneticist, I was just, you know, cheerful. And I thought: well, we certainly have a problem, but we've known that for a long time. But when I learned it was my problem, really and truly, I just fell apart like a dollar watch. I truly could not deal with it. I'm not blaming the geneticist, but if I had been told, "Listen, don't deal with this with anyone else until you deal with it with yourself - let's see about helping you deal with this yourself," I would have been much better off. And so would my family, because my family eventually would have learned about Fragile X instead of <u>not</u> learning about it.

So I truly would just urge all of you to try to help people realize that it's not just dealing with the defect, it's dealing with the family that has to happen. And let the family then deal with the defect as a whole.

Panel

ETHICAL ISSUES FOR PROFESSIONALS

EDMUND PELLEGRINO, M.D.
Georgetown University Center for Clinical Bioethics

I'm glad to have an opportunity to discuss ethical issues as seen from the point of view of someone who is both a physician and a teacher of medical ethics. I'm pleased, as I think you all should be, that the National Human Genome Project has made provisions for the study of the ethical issues along with the scientific issues in the Human Genome Project. This may be the first time that a major scientific undertaking has been accompanied by a look at the ethical issues created by the production of new human knowledge. And certainly from the point of view of ethics, the issues that are arising now in the Genome Project are really fundamental and of the most complex kind.

First, I want to address ethics research and what ethics is so that your expectations of the 5% in the budget of the Human Genome Project won't be overblown; they'll be in contact with reality; what ethics, in fact, can do. Second, I will spell out actual ethical issues. And then, lastly, I will raise some fundamental questions which go beyond the practical questions that you and I face as health professionals.

Research and ethics are very different from research and science. Research and ethics deal with the questions that haven't been resolved in the past three thousand years and are not likely to be resolved in the future. What we can do with those issues is clarify them, step back, look at them, get our sights straight, get our compass bearings on what is right and wrong and good and bad in human behavior. That is what ethics deals with.

Ethics is a branch of philosophy. It is a formal, systematic and critical examination of the rightness and the wrongness of the things we do. What should we do? What is the right thing to do? What ought we to do with this new information? This information is being made available which has tremendous power for good, as you know, and tremendous power also for wrong, if it's not handled properly.

What the ethicists will do will be to look at the questions and the issues, to lay them before you. But, in doing so, let me point out that you, yourself, have to do the thinking as well as the ethicist. We do not come and say, "Here are the answers." This does not mean, by the way, that all answers are equally valid. But each one of you is accountable for what you do as a health professional and, therefore, must understand the ethical issues and come to some position which you can justify on ethical grounds.

Ethics research does not proceed by any empirical method. It does not have a laboratory method. All it uses is the human mind, in a critical, reflective way, looking at these questions of right and wrong. And it tries to find out if there are any things which ought always to be done. Are there any things which ought never to be done? Some people would say, "Well, there are some things you should never do." It also asks the question of, What do you mean by the terms and the words you're using? It looks at the logic you are using to arrive at your justification. And, above all, it looks at the assumptions from which you start.

Any ethical decision will have a structure to it. And when you are looking at an ethical decision in the clinical setting when you are dealing with human beings and trying to advise and help and work with them to resolve a human issue, then you need a philosophy of that professional relationship. Is it a relationship of trust? Is it a contract? Is it a business arrangement? Are you a businessman? Are you a physician? Are you a nurse? Are you a counselor?

You may say, Oh, yes, I know the answer to those questions. But I must warn you, the way our behavior manifests itself, it is clear that we do not always agree on precisely what constitutes a professional relationship. It makes a great difference at the very first step whether you feel that you are involved in a trust relationship in which the good and the primacy of the person you are serving is first, even at times above your own self interest. That is very different than seeing it as a business transaction or commercial transaction or looking at health care as a commodity today, which is the problem that you're trapped by.

Health care is a commodity. We are in the business of providing health care. If you're a businessman, we are not interested in your problems; we are interested in the bottom line. If you're a genetic counselor, what is the relationship?

You need to understand what theory of ethics you actually use. One principle in ethics is beneficence. One is always doing good for the patient. Then you say, What do you mean by that? Do you mean by that, not harming? Do you mean by that, preventing harm? Do you mean by that, actually doing good? Is it doing good when it costs you something? That's the crucial point. That's what makes a professional, for example. Where do you put the cut off on what you do: whether you do it for pay or not?

Most importantly, particularly in the field of genetics, what is the ultimate source of your morality? Where does it come from? When we push you back against the wall, you finally have to say, "It's right because it is what I arrived at by the use of human reason." That's philosophical ethics. And others will say, "No, it's this way in the Bible." And that would be a Fundamentalist interpretation. Or someone would say, "That's because a church teaches it." Or someone else would say, "Well, it's right because it's really built into your nervous system" It's part of evolutionary biology. And we know what part of your brain virtue resides in.

If you understand the structure, you will appreciate that research and the ethical aspects of the Genome Project are not going to come out with clean answers that everyone can say, "Well, we have arrived at that one; check it off. Let's go on to the next one." It's going to be a continuing dialectic and dialogue which is the history of ethics for the past 2,500 years.

Let us deal with the ethics of getting the knowledge itself, the ethics of the research which involves you because you may be part of that research project. And you have to ask yourself, "Are there any limits to this kind of investigation?"

Are there some things which ought never to be done? For example, some feel that we ought not to create human embryos for the purposes of getting genetic information, no matter how valuable that information is. Others would disagree. This is a very fundamental question and it needs to be raised. When we are dealing with gene manipulation, do we limit it to the somatic cells? Most people would agree that somatic gene therapy is acceptable. But when you're beginning to deal with the germ line, others would draw a line at that.

Should we use genetic engineering for eugenic purposes? There is going to be a tremendous push in that direction of creating better human beings.

There is a book out called 'Genethics,' which discusses the moral issues in the creation of people. What the author is saying is, How do we create a race that is superior or is rid of all the foibles and problems we have?

Let me simply say that the complexity of the genetic counseling process is one that should cause and give pause to these significant ethical issues. Can the counselor, in fact, be neutral? Can any human being keep his or her values out of the transaction? If they're in the transaction, what are the limits to which one should put? Should one step out of counseling if one has certain particular points of view? What is the degree of cooperation one has with something that is wrong if one participates in the counseling process and provides information that may be harmful? Where do we stop? What do we know? What is a family member's moral claim on that information? Other aspects to discuss are your moral claim to privacy and to confidentiality, access to information, ownership of the information, etc. Can we, for example, use genetic counseling for purposes of transmitting our own values? Is it not more honest to say who you are, what you are, and where you are coming from? And then give the patient or the person who is being counseled the information.

And then, finally, there is the big question of who owns this knowledge. You already have heard about a situation in which the insurance company is using this knowledge against a human being.

The fundamental question is, Who are we? What is a human being? What is it to be a human being? What are human beings for? Are we accountable if we have a genetic defect, at least a behavior disorder? Are we responsible? Can we be held responsible for the results of what happens in our genes?

We must not forget about nurture and environment, which interact with our genetic endowment and what ethics is and how that research differs from scientific research. We must remember that there are ethical issues involved in collecting the knowledge and using the knowledge and in owning the knowledge. The most fundamental questions are the ones that we cannot answer, but they are the ones that are most critical in answering the question, How do we use such powerful information as genetic knowledge?

Panel

LEGISLATIVE ISSUES AND UPDATE

KAREN ROTHENBERG, J.D., M.P.A.
University of Maryland Law School

I am going to focus on the use and misuse of genetic information in the contexts of health insurance, employment and privacy. In order to put public policy and legislation in context, I'd like to just focus on three themes. One is what I call 'genetics as a quick fix.' And society, providers and consumers have to be very sensitive to this. What I mean as a 'quick fix' is that we often look at the genetic test as an end in and of itself. We focus on the result of a genetic test rather than recognizing that a genetic test is just a means, hopefully, to make the quality of life better for that individual and their families. I think we have to be very sensitive to that. You will see how that has played in the making of public policy.

The second is an area in which ethicists, sociologists, psychologists, etc., and those that have been critical of the Human Genome Project have been very sensitive to what I call 'genetic myopia." We have our blinders on in terms of not being able to put genetics in the context of health more broadly. As a result of that, we could have *reductionism,* where everything is boiled down to the genes and a description of the genes, and *determinism,* where we may think everything is just determined by the genes. So we might as well just give up. And, again, I think, providers and consumers have to be sensitive of these things occurring in our society.

And then, lastly, is what I call a 'genetic underclass.' We have a society right now where we have 40 million people who have no insurance at all. If you don't get prenatal care, you don't get prenatal testing. If you don't get access into the system, you don't get providers helping you out. When we're thinking about public policy, we have to be very sensitive to an underclass that we are now layering on top of a social underclass that is going to be intensified and focused based on their genetics and their inability either to get the services or to be held accountable for the results.

The focus of a lot of our public policy is the struggle between whether or not individuals have a right of access to the information on the one hand, or whether or not we are attempting to just protect them against discrimination from their misuse of the information. So the access to the information and the misuse of the information are two different approaches to the making of public policy.

Since the beginning of the Genome Project in 1990, we have a sort of analytical framework for trying to get a handle on genetic discrimination in health insurance at the state level. There have been some states that prohibit the requiring or requesting of a genetic test or requesting the results of a genetic test, which is the privacy component of this legislation.

You can pass a statute that says 'You can't prohibit coverage,' which there is some attempt to do at the federal level right now. But if you do not have any prohibitions on the rates, the coverage, in itself, means nothing.

Here is the problem with these laws. First of all, with one or two exceptions, they focus on the genetic test - again, the quick fix - rather than focusing on the power of genetic information that can be obtained from other means, including from your medical record, medical exam or family history.

ERISA is a federal law standing for Employment Retirement Insurance Security Act. it basically allows those self-funded plans which are provided by your employers, and now over 45 million of us get our insurance this way, rather than through traditional insurance coverage regulated through the state. These employers are exempted from those laws I just spelled out to you. We might have this sense of security, and then you find out you are in a self-funded plan.

In 1991 Wisconsin was the first state to set the framework which has prohibitions to varying degrees. The last one is Oklahoma which appeared to have passed the law, then pulled it back somewhat and went with a task force to study the issue in more detail.

Virginia passed a statute this year, to a large extent activated by the National Action Plan for Breast Cancer and the breast cancer community. It has a two-year limit on it, but it does have genetic *information* rather than genetic tests in its prohibition. There have been a few additional states that are considering a statute: North Carolina and Michigan.

In 1995, the National Action Plan for Breast Cancer together with the ELSI working group had a one-day working group meeting in which we set out a series of recommendations as to what any legislation in this area should look like. These parallel but strengthen some of the state law approach:

- Insurance providers should be prohibited from using genetic information or an individual's request for genetic services to deny or limit any coverage or establish eligibility, continuation, enrollment or contribution requirement.

- Insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information.

- Insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information. This was the privacy component of our recommendation.

- Insurance providers and other holders of genetic information should be prohibited from releasing genetic information without prior written authorization of the individual.

The most notoriety probably came from this definition of genetic information which is information about genes, gene products or inherited characteristics that may derive from the individual or a family member. So it is not just a genetic test result. It is genetic information tied to the individual or family member. Most of the battles going on at both the state and federal levels are about this. The insurance companies argue that it's too broad. The life insurance companies have been the major fighters of the legislation at the state level.

Based on those recommendations, Congresswoman Slaughter and Senators Diane Feinstein (CA), Connie Mack (FL) and now Senator Olympia Snowe have introduced legislation: *Genetic Information Non-Discrimination in Health Insurance Act and the Genetic Fairness Act.* These two bills, which right now are not moving through the legislative process, do incorporate almost verbatim those recommendations I've just spelled out. The Hatfield and the Sterns bills are broader and include concerns about health insurance and employment. Actually, they are not moving yet either.

What is significant about these bills is that they have really raised the consciousness of the Congress in terms of the power of genetic information and their need to urgently attend to the issue. *The Health Insurance Reform Act* is the Kennedy-Kassebaum bill. The companion bill in the House is somewhat different. Amendments to both of these bills deal with medical savings accounts and mental health benefits, but the focus is genetic information.

Two bills have been introduced with respect to medical privacy. The Bennett bill in the Senate says nothing initially about genetic information, but it is now being marked up, and it will be amended somewhat in its second, eighth or ninth mark-up form. The second one has just been introduced a few weeks ago by Congressman McDermott: the *Medical Privacy in the Age of New Technologies Act*, which does say a lot about genetic information and issues. I understand neither of these bills this year look as if they are going to go. But again, the heightened awareness at the federal level is very significant.

The Kennedy-Kassebaum bill and the Archer equivalent in the House say that insurers may not establish eligibility enrollment, continuation or premium requirements based on health status. Within their definition of health status, they define genetics information.

Now, before we get all excited, Kennedy-Kassebaum says nothing and does nothing about the 40 million people who are uninsured. All this does is speak to people who already have group insurance coverage and may be going into another plan or may be going into an individual plan. It is a baby step, and it is significant in recognizing the importance of genetic information.

So what it does, as opposed to the state law, is apply to all health insurers whether they are state regulated or self-funded because this is a federal bill It has jurisdiction over those ERISA plans as well as all other insurance plans, and it includes genetic *information*. Although it does not define genetic information in the bill, it does define it in the Senate conference report. However, it does not prohibit insurers from requesting or requiring information. It has no privacy elements to it. It does not require them to obtain authorization before disclosing information. It does not restrict the premiums or the rates. And it does not impose requirements for reimbursement for genetic services. Consequently, it is narrow in what it really does do.

Clearly with respect to ERISA, the relationship of health insurance in our society with employment is very clearly intertwined. Some people who are employers might have an incentive to discriminate based on genetic information because it could have an impact on their health claims. Therefore, it can have an impact on your employment possibilities, on your availability or your access to health insurance, and what privacy you might have in the context of your workplace.

More specifically, in the workplace context there can be pre-placement medical exams and review of your medical records. Your employer might be paying your health insurance claims. They might, in fact, be your health care provider. Therefore, you have to be very sensitive about those issues as well. And many of you in your professional associations have members working in employment settings where, in fact, that's their job - to give this information to the employer about the employee.

Paralleling what I just showed you at the state level in health insurance are similar sorts of restrictions for employment discrimination. It is no accident that many of the states at the same time try to address health insurance and employment. They may be integrated with other anti-discrimination provisions. They prohibit the testing as a condition of employment. You have to get informed consent. It cannot affect the terms and conditions and the privileges of employment.

They just focus on the genetic test. Significantly, the employer is not prohibited from requiring a general medical release. The law does not restrict the access to the genetic information but focuses on how the test results will be used. The privacy issue is still very much there. However, the burden is on the employee to prove that the employer discriminated.

The Americans with Disabilities Act (ADA) prohibits pre-employment medical exam questions so you get a conditional offer. Once you get a conditional offer, it permits employers to do a medical exam of unlimited scope following the conditional offer of employment and get a release for all your medical records.

The EEOC, the Equal Employment Opportunity Commission, interprets the Americans with Disabilities Act. In 1995, they made a very significant statement. It is not the law. It is not a regulation. Rather it is in their compliance manual. It shows a recognition from the enforcement agency that genetic information is powerful and that a disability applies to individuals subjected to discrimination on the basis of genetic information, using the words 'genetic information' relating to illness, disease or other disorders. In its description, the Commission uses an example of an individual who had a predictive genetic test for colon cancer.

With respect to genetic privacy and confidentiality, you providers are well aware that we don't have one place to look. We have a patchwork of legislative sources at the state and federal levels that may or may not speak to how much protection you get for genetic information. It can be in confidentiality statutes, in data bases, in public health genetics programs, research regulations and anti-discrimination statutes, etc.

In the context of health insurance, these laws clearly state genetic information is the unique property of the individual, that it is privileged and should remain confidential. In and of itself, this means nothing. These are just proclamations within these laws, but they often they do not have any enforcements or fines associated with them

The challenge to providers is that even if we have protections in the research context, once things get out into the clinical setting, we have even less protections from this information being misused. So when is the appropriate time to move the research into market? When is it appropriate for certain tests to be moved from high-risk families to the general population? And then ultimately, what is going to be the impact of all this genetic information and this testing overall on our public's health?

Panel

EDUCATION ISSUES

MICHAEL J. SCOTTI, Jr., M.D.
American Medical Association

First of all, I do not want to come here under any false sort of false pretenses. I have been focusing on the educational issues having to do with the Human Genome Project all of six weeks now. Initiating educational programs are not new to society or to any of our professions or disciplines. I went about looking at the educational issues of the Human Genome Project based on many other educational programs, requirements, and needs that have been out there. I came away from the process with only one word in mind and that was 'cost.'

I think on some of these issues it affects all of the population, not kids only, adults only, geriatrics only, the unborn only, the sick. It affects the educational needs for the health professionals to enable them, us, to serve all of the population. It involves making intergenerational decisions. That is, the educational process will affect not only the people we're talking to, but through their decisions, their children and their children's children or, by the same token, there are no children and there are no children's children.

It's education and at a moving target. The amount of information is growing very fast and I think will continue to do so. Dr. Collins in his very appropriately well-received discussion talked about functional cloning and positional cloning. How we get down to where the gene is what is causing the problem.

The fact is, as we get more and more information, we're going to go the other way. We are going to start with the gene that is not exactly the way it's supposed to be and look for what it's doing. And I think we'll find a lot of other things.

The other difficulty in education about the human genome is that it is not compartmentalized. It affects all of us. It affects every single health provider, every single person involved in the system. When they put nuclear isotopes into the system as a means for diagnosis and therapy, people didn't have to know a lot about them because they were compartmentalized off, you know, in that nuclear medicine place in the basement of the hospital.

If you come up with an infection control business for surgery, you can compartmentalize it. And only those people that are up in the operating room really need to get into that on more than a superficial basis. But, the fact is, the information coming out of the Human Genome Project is not compartmentalized.

We are going to have to be fighting the market. And, that is, our education. Our education is going to be competing with not only the media, but with the people who are advertising their genetic products. I will tell you following the home pregnancy tests and following home HIV tests, there will be home genome tests. If something is possible, it will be done; it will be sold.

It covers education, covers every aspect of the clinical site. It is required for history taking, not only direct history taking, but past medical history and family history. It affects physical examinations in terms of requiring information, you have to know what to look for, and what is necessary for the diagnosis. It is increasingly necessary for treatment. It certainly is necessary for prognosis. Education in this area has a frightening potential for misuse. I'm not all that old, but in my life time, there were racial laws on the books about killing Jews, killing Gypsies, races not being able to marry, people with certain conditions being shipped off to isolation areas in Louisiana. I mean, this information, a little information is a very dangerous thing in some areas.

So for all those reasons, I think it is awesome when you look at the educational issues involved. There are some disadvantages in things working against a good educational program. For one thing, there's what I call, "the disease-of-the-month club" for the waft of the week. For every one of the recognized conditions there are coalitions of support groups, professional organizations, charity-giving organizations, research affiliates that make up a little package that needs to compete in the market place against everybody else.

As a result, there is a phenomenal amount of splintering in terms of the depths of education that is out there and the way to work it. We are also going to have to build a firm basic science base, and then on top of that, an applied science education for people who are coming up into the disciplines, into the professions. They are going to compete against everything else out there on the curriculum. They are going to have to be widespread, not as a separate curriculum, but be inculcated into a bunch of things out there. Which means, by the way, we have to educate some educators which is a whole different thing.

But then we have to retrofit those who are already out of school. I mean 90% of the health care provided in the next 25 years is going to be given by people who are already out of school. So that is a difficulty for continuing health education.

I do not think there's going to be a lot of money in this genome. And that's going to be a disadvantage.

There are some pluses. There are really some things that will help us as we deal with this. One is the fact that it has name recognition, that is, genetics, genes, things that run in families have recognition under one terminology or another. It is going to be demand driven. Whether we call them patients or consumers or clients or customers, there is going to be a phenomenal demand for answers to questions, specific questions about themselves and their family members.

This gathering and similar gatherings like it over the last several years which are interdisciplinary are very healthy. We may wind up not having to re-invent the wheel while all of us go out and develop new curricula and all of us go out and try to work through the system, because we may be able to do things together which will give us some economy of scale.

We have to be driven to do all this by our associates because there are a lot of medical/legal issues and stuff having to do with getting sued for wrongful death, wrongful life, failure to inform, lack of informed consent, and a million other things.

The fact is people are scared out there. And all of us are responsible. You know, because people come to us and they ask a question and we give them an answer. And that, ladies and gentlemen, is diagnosis and treatment. It does not have to be a formal session. We have some advantages because with this great amount of information that's pouring out, we also have some great technology that's pouring out in terms of computerization, interactive CD-ROMs, multi-media, all of which can be available to where we work and all of which can be available on the Internet. We can get information when we want it, where we are, when we need it. It may keep up with these three billion base pairs, the future of which can have one or two difficulties.

SUMMARY

AND

FUTURE DIRECTIONS

The field of medical genetics continues to witness dramatic advances, largely resulting from the Human Genome Project, in diagnoses, presymptomatic and predispositional testing, carrier detection, and improved treatment for a variety of genetic conditions. With the recent findings of possible genetic links to behaviors and life styles, the very definitions of genetic defects or genetic disorders are under scrutiny. While the potential to predict, cure and prevent disease with genetic technology seems limitless, the potential for abuses of genetic information by insurers and employers is also limitless. Both the advances in identifying the genetic component of diseases and concerns about some of the ethical and psychosocial issues of genetic testing have led to an increased demand for genetic services and have brought genetic issues to the forefront of health care.

It is often the allied health professional who is asked to explain or give more information about genetic conditions. However, the HuGEM survey, in addition to letters from leaders of professional organizations, revealed that allied health professionals have little knowledge of human genetics and the ethical, legal and psychosocial issues.

Participants at this conference on June 10, 1996 documented both the need and the motivation for allied health professionals to incorporate genetics and the implications of genetic testing and research into their professional and continuing education. The co-directors of the Human Genome Education Model (HuGEM) Project and of this conference, Virginia Lapham, representing the Georgetown University Child Development Center, and Joan Weiss, representing the Alliance of Genetic Support Groups, look forward to working with the leaders of the professional organizations who have expressed their enthusiasm and commitment towards going forward with educational efforts in genetics within their disciplines.

(Appendix)
Education of Interdisciplinary Professionals Meeting
June 10, 1996

HANDOUTS AND RESOURCES

Alliance of Genetic Support Groups **1-800-336-4363**
35 Wisconsin Circle, Suite 440, Chevy Chase, Md. 20815

Alliance Brochure
Alliance Publication List

American Medical Association
515 North State Street, Chicago, IL 60610

Video on Fragile X

Department of Energy **(301) 903-4742**
Human Genome Program,19901 Germantown Road, Germantown, Md. 20874

ELSI Projects, March, 1996
Mapping and Sequencing the Human Genome: Science, Ethics, and Public Policy
BSCS Innovative Science Education, 830 No. Tejon St., #405
Colorado Springs, Colo. 80903-4720
Your World, Biotechnology & You, Vol. 5, Issue No.2
Pennsylvania Biotechnology Assn, 1524 W. College Ave., # 206
State College, Pa 16801

HuGEM I

Two videos and manual

National Center for Human Genome Research **(301) 402-0911**
Office of Communications, 31 Center Drive, NIH Bldg. 31, Room 4B09, Bethesda, Md 20892

Genetic Information and Health Insurance
The Human Genome Project: From Maps to Medicine
NCHGR Fact Sheets
NCHGR; DNA Sequencing; Ethical, Legal and Social Implications of the Human Genome Project; Genetic Mapping; Physical Mapping; Position Cloning
"Medical and Ethical Consequences of Human Genome Project" by Francis S. Collins
Program Announcement: Ethical, Legal, and Social Implications of Human Genetics Research

Article about Elizabeth Thomson

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91 Summit Way. Syosset, NY 11791

National Rehabilitation Association — **Ann Ward Tourigny, PhD, CAE**
633 S. Washington Street, Alexandria, VA 22314

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c/o MD Dept of Health & Mental Hygiene, 201 W. Preston Street
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Uniformed Services University of the Health Sciences — **Edmund G. Howe, MD, JD**
13914 Vista Drive, Rockville, MD 20853

Ethical, Legal, and Social Implications of the Human Genome Project: Education of Interdisciplinary Professionals

Conference Participants - June 10, 1996

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