

RECORD, Volume 28, No. 3*

Boston Annual Meeting
October 27-30, 2002

Session 53PD

Impact of Genetic Research on Health Carriers

Track: Health

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Summary: The impact of genetic testing on the cost and delivery of health care has recently attracted public attention. Scientific advances in the understanding of human genetics will affect the various public and private systems for financing medical care and risk classification for health insurance. Panelists discuss potential effects on the health insurance system and implications of future developments in genetic technology.

MR. BRUCE PYENSON: Cecil Bykerk, who is listed in your program as your moderator, is not able to be here today, so I'm filling in as moderator and speaker of this session.

We're very lucky to have guests from Washington, D.C. with us today, who have been active in the political and in the technical area of genetic testing. It's unfortunate that Cecil can't be here today because, as you know, he's a senior actuary involved in Society and Academy activities and he has been very involved in testifying on Capitol Hill on behalf of the profession and on this issue in particular.

We have with us today Kathy Hudson and Craig Burton. Kathy is a biochemist by training, with a Ph.D. from Berkeley. She was involved in the Human Genome Project for about seven years at the National Institute of Health (NIH).

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Now she's at Johns Hopkins as the director of the Genetics and Public Policy Center, a part of the Berman Bioethics Institute. She's heavily involved in public policy issues, especially as related to human reproduction and fertility. She's been involved in genetic testing and related issues for quite a while in both the technical arena and with their connections to public policy. Craig is also from Washington. He is the lead health advisor to Senator Frist (R-TN), the ranking Republican on the Senate Subcommittee on Public Health. Craig served on Senator Frist's staff for a number of years in Tennessee, and he is now involved on a day-to-day basis in supporting the subcommittee on anything related to health that comes before it.

I'm a health actuary with Milliman USA in New York City. Most of my work is related to managed care. In recent years, we've gotten involved in a number of projects related to things like disease management, where you value things like medical intervention (such as prevention and detection of cancer) and return on investment, using the traditional actuarial techniques like we do for rates or reserves.

DR. KATHY HUDSON: This morning, I'd like to talk for a few minutes about genetic testing, where we are today and where we're likely to go in the future. Then I'd like to focus on some of the ethical and policy issues being raised by advances in genomics.

It seems like genetics is virtually everywhere today. It's in cartoons, including one in which a gentleman sees on TV that the genetic code has been cracked and he's dismayed because he can't even program his VCR. We see DNA in cosmetics and leisure advertisements as well.

Certainly our fascination with DNA and the genome was heightened in February 2001, when two groups announced they had completed a working draft of the human genome sequence. One was the federally- funded Human Genome Project, with which I was associated, and there was also a private effort by Celera Genomics.

Why all of the fuss about DNA and the genome? What's likely to come about as a consequence? The reason the federal government invested in sequencing the human genome, and is investing heavily in genetic research generally, is that virtually all diseases have some genetic component. The contribution of genes to disease can vary across a large spectrum. For some diseases, such as cystic fibrosis, the genetic component is very strong. If an individual has a mutation in the cystic fibrosis gene, then it is likely he or she will be affected by cystic fibrosis, though the severity of those symptoms may vary because of environmental circumstances.

On the other end of the spectrum are diseases we think of as infectious diseases. We know that AIDS, for example, is caused by an environmental agent (human immunodeficiency virus— HIV), but even for AIDS there's also a genetic

contribution. Specific genes can increase or decrease the likelihood that HIV will infect us. But most common diseases fall into a middle category, where neither a single gene nor a single environmental factor or infectious agent causes the disease, but rather a number of genes interacting with one another and with complex environmental factors give rise to the disease. Examples include adult-onset diabetes, heart disease and many forms of cancer.

The hope is that with the powerful tool of the genome sequence in hand and the advances in other areas of genetic research, we can identify individual genes, make the association with disease and other disorders and then ultimately develop and make available effective therapies, interventions and prevention technologies.

Once you have the gene in hand and you know what its association is with a given disease, it's a relatively straightforward matter to develop a diagnostic test, a genetic test, to determine whether or not someone has a misspelling in that particular gene. This has been the focus of considerable research and effort in the biotechnology and pharmaceutical industries. That has caused what some people call the "therapeutic gap," that is, you can have this information about a mutation in gene or genes, but an effective intervention is not yet available.

Of course, we want to have effective therapies. Those therapies can be drug-based therapies, development of smart drugs based on understanding the specific molecular underpinnings of that disease and what goes awry when a gene is not functioning properly, or gene therapy, using the gene itself as the drug. Gene therapy has been in the research setting for many years with only very, very modest success. Also, there is the whole new area of pharmacogenomics—understanding how an individual's genetic profile makes him or her either more likely or less likely to respond to a particular drug, and also whether or not he or she is likely to have an adverse consequence as a result of taking a specific drug. There are a number of examples where specific genes have been found to be associated with specific drug response characteristics.

I'd like to talk about a few concepts that are important for understanding genetic tests and the genetic contributions to disease. They are: penetrance, age of onset and the availability of an intervention. For certain genes, if you have a mutation in the gene, you will get sick. The example I gave earlier was cystic fibrosis. If everybody who has a misspelling in that gene becomes sick (100 percent in this particular example), that's called "high penetrance" or "complete penetrance." Those genes were largely the ones that were identified early, are easy to identify from a genetic research perspective and are mostly very rare childhood diseases.

More recently, genetic research has focused on finding those genetic contributors to the common, complex diseases: stroke, cardiovascular disease, diabetes, etc. In these later-onset diseases, penetrance is often incomplete or low. A gene mutation is low penetrance if, for example, only 11 percent of the people who carry that misspelling are sick by the age of 45.

Today, more than 900 genetic tests are available or in development. Approximately half are available clinically today. Additionally, the majority of those are for single-gene disorders. The other concept is the availability of interventions. This is a very important concept because it's very straightforward to develop the diagnostic, and yet developing the effective intervention is a much slower and complex process. The spectrum ranges from effective intervention to no effective intervention available. Huntington's disease is on the far right (no effective intervention available) of the spectrum. If an individual's father or mother had Huntington's disease, he or she has a 50 percent probability of contracting the disease. A genetic test can positively detect or rule out the presence of the gene mutation, yet there's little one can do medically to avoid getting the disease.

Further down in the spectrum is cystic fibrosis, where there are now an increasing number of drugs available to ameliorate symptoms and increase life span. BRCA1 and BRCA2, genes that increase the probability of developing breast and ovarian cancer, fall in the middle of the spectrum. There are now surgical prophylactic techniques and some pharmacologic methodologies being employed to reduce the risk caused by mutations in these genes.

We have greater intervention capabilities for hereditary nonpolyposis colon cancer, a disease that falls closer to the left (effective intervention available) of the spectrum. For a number of hereditary colon cancer syndromes early colonoscopy and regular colonoscopy can detect polyps early. The polyps can then be removed, massively decreasing the incidence of cancer.

Hemochromatosis is one example of a disease with very high intervention capabilities. Hemochromatosis is probably the most common genetic disease among Caucasians and is massively underdiagnosed. Currently, there's a large NIH-sponsored clinical study looking at the frequency of hemochromatosis mutations in the population. In this disorder, the body is stuck in the "on" position for absorbing and taking up iron. So the body takes up iron, takes up more iron and takes up even more iron. Because of iron overload, the affected individual ends up having damage to many organ systems. The effective intervention, very low-tech, is phlebotomy (bleeding), getting rid of excess iron in the blood, thus averting the devastating consequences of this particular gene mutation.

Genetic medicine has enormous promise. Medical professionals and genetics professionals are hopeful that a large number of diagnostic and predictive genetic tests will become commonly available as a part of routine medical care, that illnesses will be detected early by molecular surveillance, that gene therapy will be increasingly successful, that we'll have gene-based designer drugs and that pharmacogenomics will increase drug efficacy and decrease adverse reaction.

Despite this promise, the public is troubled about advances in genetics. There was a *TIME/CNN* poll taken about the time of the announcement of the working draft of

the human genome sequences by Celera and the Human Genome Project. When asked whether they would want information about the risk of genetic disease in the future, a slim majority indicated that, yes, it would want that information. When asked whether or not the Human Genome Project would be harmful, nearly half said, "Yes, it's going to be harmful." Last year, in a less scientific measure of public opinion, *Vanity Fair* in its "Thumbs Up, Thumbs Down, What's In, What's Out," section indicated that in the category of scary science, Sally, the cloned sheep, was "out," and the Human Genome Project was "in" as scary science. If the same evaluation were to be done today, I suspect that the Human Genome Project as "out" as scary science and human cloning would be "in."

What are these concerns? The public has a number of concerns about the privacy and fair use of genetic information and how it will affect their families. The following stories illustrate some of the aforementioned concerns. This story is about Judy, her two brothers and their father. Judy's father has been affected by early-onset presenile dementia. He requires the constant care of Judy's mother. He is unable to do many of the simple life tasks in order to take care of himself. Judy's brothers have early signs of presenile dementia, both with short-term memory loss and difficulty with new learning. These were symptoms that Judy's father had early on in the progression of his disease, and Judy and her mother are suspicious that the two young men have the same disease.

Judy's mom learns there is a research trial looking at the genetic contributors to early-onset presenile dementia. The trial looks at three genes that have been shown to be involved in presenile dementia: presenilin 1, presenilin 2 and amyloid plaque precursor protein (APP). Judy's mom is interested in having the family participate in this particular research protocol, despite knowing that the family members themselves will not benefit. There is no intervention available for early-onset dementia. Nonetheless, Judy's mom talks with her family members about whether or not they'd like to participate in this research trial. The affected brothers are interested in participating. The father, who really isn't able to consent to participate in research—which brings up a whole host of other issues—also agrees to participate. But Judy is not sure that she wants to know. She hasn't noticed any of the symptoms in herself, and she's not sure that she wants to have this information.

The affected family members (Judy's dad and her two brothers) get tested and they are all found to have a C instead of a G in the amyloid precursor protein. That is the causative misspelling that is causing their disease. This is very real now for Judy. She knows now that there's a 50 percent chance that she has inherited this mutation from her father. She's thinking about whether or not she wants this test. She's thinking about how this information might affect her personally, what her reaction would be to knowing and what future implications lie ahead for her. One of her concerns is how this information might be used against her. She is concerned about how this information could be used by her employer. Does she really need to be worried about this?

In February 2000, President Clinton signed an Executive Order that prohibited the federal government from using genetic information in making hiring, promotion and other job-related decisions. In signing that Executive Order, which applies only to the federal government as an employer, he said he wanted to set an example for the private sector in not using genetic information in making job-related decisions. One year and one day later, it was discovered that the Burlington Northern Santa Fe Railway had surreptitiously tested employees for supposed genetic susceptibility to carpal tunnel syndrome. (It turns out that the genetic test for carpal tunnel syndrome is a completely nonvalidated test). The union brought suit and the Equal Employment Opportunity Commission got involved. That case was later settled. To illustrate the bipartisan interest in encouraging employers to use the ability of an individual to do a job and not his or her genetic risk factors in making job-related decisions, President Bush, in a June 2001 Saturday radio address, indicated that he would support legislation prohibiting the use of genetic information in employment and in health insurance.

If Judy is a federal employee, she doesn't need to worry whether her employer knows she's at risk of developing this disease or if she has a positive test result. However, Judy still might be worried about whether or not this information could be used by her health insurer to deny her coverage or limit her benefits. In 1996, Congress took an important step in the passage of the Health Insurance Portability and Accountability Act (HIPAA). This Act includes limitation on the ability of insurers in the group health insurance market to use health-related factors, which include genetic information to determine who will or will not be granted coverage. "Genetic information" was *not* completely defined by HIPAA, but HIPAA specifically precluded considering predictive genetic information? information about a healthy person's future risk of disease. The Act said this type of information could not be regarded as a preexisting condition.

Although HIPAA did not define "genetic information," the definition of "genetic information" is very important and very difficult. Many states have enacted legislation to prevent the use of genetic information in either the workplace or in health insurance, or both. The definitions of "genetic information" included in those state laws are incredibly variable and many of them are very unclear. This reinforces the notion of how difficult it is to come up with a coherent, robust definition that works. Since Judy is a federal employee, she gets her health coverage through the federal group health insurance plan. She's protected by the 1996 HIPAA law.

What else might Judy be thinking about? Judy's married and thinking about having kids. Having a family is important to her sense of fulfillment and meaning in her life, but she's concerned that if she has this mutation, she might pass it along to her children. It's the contemplation of building a family that causes her to get the genetic test. She finds out that she does carry the mutation.

What should she do? She could take the roll of the dice and have a child, knowing that the child would have a 50 percent risk of carrying that same mutation. She's heard about some new technologies that might allow her to have a child that would not be affected by this genetic disease. One relatively new procedure is preimplantation genetic diagnosis, in which in vitro fertilization is used and a genetic test is performed on the resulting embryos to select only those that are free from a mutation. Then the selected embryos are placed back into the woman's womb to initiate a pregnancy. This might allow Judy an opportunity to have a child that she would know would not have that particular genetic disease.

Preimplantation genetic diagnosis has been done for 100 different genetic diseases, most of them early onset. There's significant controversy about using this technology for late-onset diseases where there's variable or low penetrance. There is active discussion about what are the limits of this technology and if there should be limits on this technology. How do people make those kinds of choices?

These are some of the issues that I'm exploring in detail at Johns Hopkins University's Genetics and Public Policy Center. We are looking at genetics in human reproduction, looking at all of the issues surrounding the advances in genetics and looking at the combination of advances in genetics and human reproductive medicine. We've been fortunate enough to be funded by a substantial grant from the Pew Charitable Trust. Over the course of the next several years, we will develop policy options? not a single set of policy recommendations, but a set of policy options that may guide the development and use of these important technologies.

Genetic science is hurdling along the track, and it will require the active involvement of all of us to make sure that the policy and the science move down the track without colliding. As Thomas Jefferson said, "But I know also that laws and institutions must go hand in hand with the progress of the human mind." Certainly in the field of genetics, the human mind's progress has been incredible over the last several years, and it's important that our laws and institutions keep pace.

MR. J. CRAIG BURTON: Thanks, Kathy. Actually, I liked that last cartoon of yours of the trains, but I would disagree a little bit with the concern about a railroad collision. I think what we see most often is science, in this case genetics, hurdling forward and public policy almost in a horse and buggy, so to speak, trying to catch up. That's the kind of challenge that we face.

Kathy, you touched on a lot of issues that are raised by these genetic testings. I think definitely we face regulatory issues regarding safety and efficacy, and cost and access issues. These issues aren't necessarily unique to genetics. These come into play any time we have advancing science and medical innovation. However, there are clearly some unique issues raised by genetic research that are specific to genetic research.

One that you didn't mention that I would just throw out as food for thought are some of the intellectual property implications of genetic research and how our patent system addresses some of these issues. I'm sure that most folks have heard what sometimes come off as hysterical claims of scientists "patenting life," which obviously no one likes that thought, but at the same time, that's an incentive that is important to getting this research done and published.

I think the most immediate public concern is the misuse of genetic information for discriminatory action in health insurance or employment. But the public interest in genetic testing comes from the tension between two interests. There are really two major reasons why folks have come around to this. One is a broad societal desire to prevent discrimination, but there's also the very basic, practical desire to see this research optimized and have us gain as much as possible from this knowledge.

Kathy touched on the concern that genetic testing of what individuals may be at risk for in the future may be used against individuals. There's the case that people have cited, Burlington Northern, where I think it was almost a Keystone Kops attempt at genetic discrimination. As Kathy mentioned, the test has been invalidated. Really, Burlington Northern was looking for some type of genetic link in their workers who were on disability for carpal tunnel syndrome to try to get out of paying workers' comp benefits. In any event, that case was settled, but it did highlight some of the concerns that people have.

There's definitely the goal to protect genetic information, but one of the other things that we've seen is that if we're going to optimize this research, we also need to protect individual's ability to take the tests. It is not enough to keep that information private and make sure it cannot be used as the basis for discrimination. We also have to make sure they can't be discriminated against simply for going and taking a genetic test. Like I said, one of the biggest concerns is that this fear may prevent individuals from participating in research, and thus it may prevent us from increasing the quality of care. This idea of improving health care quality, as we look at it, it requires both patients and practitioners to have the best information available to them—this means both through better research and the ability of patients to take that tests and share the results with their provider without the fear that it could be used against them.

In some cases, you're talking about if an individual has a certain gene misspelling, then he or she will develop a disease. But in more cases than not, that misspelling just puts them at some level of higher risk, and with its interplay with the environment, that could result in developing a disease or not. The goal is to reassure the public that if individuals or families want to undergo genetic testing, they can, and they won't be subject to genetic discrimination.

Kathy touched on HIPAA, which prohibited group health plans and insurers from adjusting premium or contribution amounts or establishing enrollment restrictions for individual members of a group on the basis of health status, which included

genetic information. But as Kathy noted, HIPAA did not define "genetic information." HIPAA also only applies to the group market. Following that, people really looked at this and said that we need to do more.

In the 105th Congress, two Congresses ago, more than 10 bills were introduced that dealt with this. Some bills focused on health insurance, some bills focused on employment. There were a number of negotiations. Following negotiations between members on the Health, Education, Labor and Pensions Committee and a number of the interested parties, Senators Snowe (R-ME), Frist (R-TN) and Jeffords (I-VT) introduced in 1998 a bill that focused on prohibiting discrimination in health insurance.

The bill was focused on healthy, asymptomatic people in the group and individual markets. It would have applied to all employer group health plans and state-regulated plans in the group and individual markets, but bipartisan agreement and the ability to move this bill was prevented by continued disagreement over three issues: definitions, which Kathy raised; remedies, which is a recurring issue, and whether we're going to allow lawsuits or how we're going to enforce this. Also, a new focus was raised on the importance of also addressing employment discrimination.

In the 106th Congress this issue was debated by the Senate. The Snowe, Frist, Jeffords bill passed the Senate on three occasions. A modified version also passed the Senate in the 107th Congress, but the House has never acted on the bill. The amendment was stripped out of the bills it was added to, so we really are no further along than we were six years ago. During this congressional session, Senators Snowe, Frist and Jeffords reintroduced their Bill S.-382. Senators Daschle (D-SD) and Kennedy (D-MA), and Representatives Slaughter (D-NY) and Morella (R-MD) in the House, who are the sponsors of the main opposing proposal on this, also reintroduced their bill, but there appeared little impetus for action.

I think people were pretty strongly entrenched, and I really credit President Bush's address, which came as a surprise to everyone, for sparking new interest in this. Specifically, the president called for Congress to pass fair, reasonable legislation prohibiting discrimination that is consistent with existing discrimination statutes. Following this, the sponsors of the Snowe, Frist, Jeffords bill started to craft a new section to add to their bill regarding employment discrimination. The goal here was to provide safeguards that are consistent with those already available through other discrimination statutes, such as Title VII of the Civil Rights Act or disability discrimination under the Americans With Disabilities Act (ADA).

At the same time, we tried to update and streamline the health insurance discrimination sections of the bill to reflect the latest developments. The bill that we had introduced was written in 1997, and the definitions and other parts of the bill hadn't been updated since then to take into account new science and new regulatory action. In March of this year, Senator Snowe reintroduced her bill as S.

1995, the Genetic Information Nondiscrimination Act of 2002. Senator Frist, Senator Jeffords, Senator Gregg, and Senator Ben Nelson (D-NE) were cosponsors, as well as a number of other members.

The bill prohibits the use of genetic information in employment decisions. It also imposes restrictions on the obtaining of such information by employers and protects the confidentiality of that information if employers happen to have it. It places the responsibility for these protections with the EEOFC, which has already been addressing this issue through the ADA, which is who we rely on for enforcing other forms of employment discrimination.

On the health section of the bill, we wanted to update the definitions to reflect the state of the science—post-mapping of the genome. Because it has a privacy component, we also wanted to amend the bill to conform with the new federal medical privacy rules.

Let's walk through a summary of the bill. In the group market, the bill prohibits group plans and insurers from adjusting premium or contribution amounts, or establishing enrollment restrictions for the group as a whole, on the basis of genetic information concerning an individual or family member of the individual in the group, including information about a request or receipt for genetic services.

In the individual market, the bill prohibits issuers from using information to adjust premium or contribution amounts or using it as a condition of eligibility. The bill addresses privacy and confidentiality by requiring that a group health plan comply with existing medical privacy regulations that were issued as part of HIPAA governing the use and disclosure of individually identifiable health information. It also prohibits group health plans from requesting or requiring genetic information, including information about a request or receipt of genetic services. The point here is that there's a big fear out there that a health plan may require me to go get a test and that they might use that, so it's to say, if folks want to get the test they can, but a plan can't require them to do so.

The exception here allows health plans to request this information that is necessary for treatment, payment and health care operations as defined by the Department of Health and Human Services (HHS) privacy rules, to the extent that these reasons are consistent with the legislation. Note here that medical underwriting would not be consistent with the legislation—it would not be permitted. The point here is that if plans were being asked to pay for an individual taking a genetic test, they would not be able to have access to the results of the genetic test to use those results in underwriting or for other purposes. But there may be purposes for which they need that information or when it's useful to have access to it. They need it simply if they're asked to pay for the test. In some quality operations, as we try to have genetic information integrated into the mainstream of the practice of medicine, we do want that information as part of operations, as long as it's not being misused.

The enforcement remedies are the same remedies as for discrimination participants, or the Department of Labor can sue for benefit recovery under ERISA and the IRS can impose tax penalties of \$100 per day per person.

Kathy touched on the number one key issue, which is definitions. Definitions are the key to the bill, but because the science is rapidly evolving here, they're also difficult to nail down. A definition that is too narrow may not adequately prevent discrimination. On the other hand, a definition that is too broad can bring current health status into play and have major consequences, mostly detrimental, on how providers treat patients, on how health insurance functions, on the scope of existing discrimination laws, such as the ADA, and on a range of employment practices. Moreover, many large employers offer work site wellness benefits such as cholesterol or diabetes screening.

These employers probably don't realize that the information they're collecting is genetic information. The key here is to accurately define "genetic information." The intent is to protect healthy people. The Snowe-Frist bill does this. It seeks to distinguish between current health status and an individual's genetic predisposition. Protections are applied only to genetic information and not to an individual's current health status. If someone is sick today, insurers can use that information to determine coverage and set premiums according to HIPAA. The bill doesn't interfere with current insurance practices that allow carriers to set premiums based on the risk. An insurance carrier would typically charge a higher rate for a person who currently has cancer than for one who doesn't. The point here is that a carrier cannot charge a higher rate for someone who has the BRCA 1 or BRCA 2 gene who has no symptoms, but who simply has a higher risk for developing cancer.

In contrast, here the Daschle-Kennedy bill uses an overly broad and circular definition that doesn't clearly exclude current health status. Their bill would essentially pre-empt state laws that have historically had the exclusive jurisdiction to regulate individual insurance, and it would establish new federal rate-setting rules that would apply across the board. It would essentially get rid of all medical underwriting based on current health status, on the basis that current health status is genetic information. That's the key issue.

Another key issue is structure. The Snowe-Frist bill is built into the HIPAA framework. It allows states to exceed those protections. What we're establishing is a federal floor of protections. If states want to act on their own to establish stronger protections, they can do so. That's a decision that should be made on a state-by-state basis.

Another key issue is remedies. The Daschle bill creates an unlimited private right of action for all violations. The Snowe bill, as I said, tracks current HIPAA remedies. We deal with this with every piece of health insurance legislation. I referred to it the other day, I think, as a demon that the people on the other side of dial from us

should need to exercise—to get over their desire to have a new right for individuals to sue. The Snowe bill tracks current law and tracks the Kennedy-Kassenbaum law, which does not allow for lawsuits. Some folks have raised the concern that current HIPAA remedies aren't adequate, that \$100 per day per person isn't adequate, and that may be the case. If so, that's the case for all of HIPAA. It's not unique to genetic information.

My boss and other members on our side of the aisle who are working on this issue are very concerned about taking legislation to prevent genetic discrimination from becoming an opportunity to create new manners to bring suit on these issues, or using this legislation to create a new precedent for us to go back and relook at certain issues such as privacy.

The third issue is privacy and how we treat genetic information as part of the medical record. Privacy is critical. It's really part of the larger debate concerning the privacy of an individual's medical records. Rather than parceling out pieces of the medical record and treating each of them differently, the sponsors of the Snowe bill have tried to look at the medical record as one piece, and address the privacy and confidentiality of the medical record, insuring that this is done thoughtfully and carefully in its entirety. Therefore, we incorporated the HIPAA safeguards and added new requirements only to the extent that there are gaps in the privacy rules today. The Daschle bill establishes extensive new privacy rules for genetic information that overlap, and in some cases conflict, with the medical privacy rules governing the use and disclosure of medical information. It would really be difficult for providers, and in the long run harmful to patients, to have different rules for the flow of genetic information versus other medical information, particularly when we're at a point today and even more so as we go forward, where most medical information contains genetic components.

Since Senator Snowe reintroduced her bill in March, the sponsors of the Snowe bill have been working with Senators Daschle and Kennedy in a good-faith effort to work through this. Kathy has been part of this as our sounding board on medical and scientific issues. The process has been useful. There's broad agreement on the policy intent. The process has also been slow. We started working on the definitions of the bill in March, and it's almost November, and we're still working on the definitions of the bill. I don't know how many drafts we've gone through here, but we are getting there. The key issues that are still outstanding are definitions, the privacy and collection issues and remedies. There are also lesser issues on unemployment, but I think those are lesser and we'll follow up if we get there—if we get the health sections resolved. The process has also been slowed down by other priorities. This has been a very strange Congress with because of September 11, and with the anthrax attacks that displaced a number of people, including myself, from their offices for several months. We're certainly understanding of that, but from Senator Frist's standpoint, and I think and the other sponsors of the bill, this issue has been out there for six years. Congress will be returning in two weeks for a lame-duck session to resolve some of the spending bills. I think we

conceivably could finish the bill up, at least on the Senate side, during that time. Will we? I doubt it. If not, Senator Frist believes this should be really one of the first things that we do when Congress reconvenes in January, so we will see where we go with this.

MR. PYENSON: Genetic testing is based on an emerging science, and it can profoundly affect the insurance industry. Public policy experts, ethicists and legislators have spent a lot of effort planning to regulate genetic testing, but the insurance industry and actuaries play almost no role at all in that public debate. For reasons I will describe, I fear that without actuarial input, we could end up with regulations that poorly serve both the public and the insurance industry.

The genetic testing debate has been going on for a number of years. Some of the material available through the National Institute of Health and on various Web sites is a decade or more old. Academics and other professionals are involved. The actuarial profession and the insurance industry hold almost no place in that debate. That's something we need to change. I hope this session will help inform you on this important issue, so the profession will have the support it needs to get involved.

This morning, I will try to create a view of genetic testing within the progress of medicine and the progress of health, particularly in the United States. This view of progress certainly applies to other countries. I will also talk a bit about what everyone here knows as "Insurance 101," which is the issue of risk classification, risk stratification and sound underwriting. I'm going to suggest a common interest on genetic testing between public health interests and the interests of the insurance industry. Perhaps that suggested common interest is also from Insurance 101—back from the days when insurance solvency and stability were a primary public interest concern of regulators.

In my opinion, genetic testing and, more broadly, the genomic revolution fits well in the amazing story of society's improving health. What we now call life science has fundamentally changed medical care. That's how I look at the development from roughly the middle of the 1800s? the great chemist Louis Pasteur and germ theory, the development of antibiotics? to more recent developments? hormone replacements (including insulin) and the emergence of specialized medical devices. Today the revolution continues with computer chips and microdevices, and new pharmaceuticals move from today's breakthrough enzyme-based medicines and into genomics and proteomics. The improvements in medical care and public health are truly amazing.

Figure 1

Remarkable Improvements in Life Span

Life Expectancy at Birth

<u>Year</u>	<u>Males</u>	<u>Females</u>
1900	46.3	48.3
1925	57.6	60.6
1950	65.6	71.1
1975	68.8	76.6
1990	71.8	78.8
1999	73.9	79.4

Same pattern for people age 65 and 75

National Vital Statistics System: *Health, United States, 2002*, Table 28, p. 116

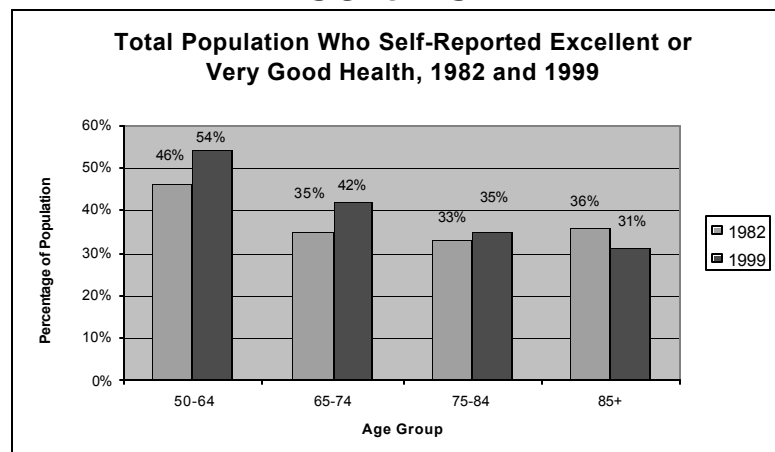
As Figure 1 shows, the life expectancy at birth has profoundly increased through the 20th century. Evidently, living in an industrial society is good for your health, and for both males and females, the life expectancy has increased dramatically. This, in fact, is the case throughout the world. The United Nations recently issued a statement saying that mortality rates are improving throughout the world. The same pattern exists in the United States for people in the oldest cohorts? age 65, age 75 and so forth. Life expectancy is improving across the board, and mortality experts predict it will continue to improve.

Health status seems to be improving along with longevity. About 20 years ago a controversial theory was aired, called the compression of morbidity. The theory says that we're living longer to actually spend less time disabled. The dismal, if common sense view is that we are living longer but spending more time disabled and sick. Happily, statistics support the compression of morbidity theory.

Figure 2 is based on Centers of Medicare and Medicaid Services (CMS) statistics. It shows the population who reported "excellent" or "very good" health, compared over a course of roughly 20 years. More people are saying they're healthier. The statistics for nursing home stays shows the same phenomenon.

Figure 2

Americans are Getting Healthier



AARP, *Beyond 50.02: A Report to the Nation on Trends in Health Security*, Source: National Center for Health Statistics, Trends in Aging Database, unpublished data, August 2001

There have been dramatic improvements in Americans' health, even recently, and certainly over the 20th century. I believe the role of insurers in that improvement has been very important and largely unappreciated.

These days, it seems to be part of popular culture to hate health insurers and pharmaceutical companies, but the fact is that a lot of the progress that has come in the course of the last 40 or 50 years has been due to pharmaceutical and insurance programs, including Medicare—the largest insurance company in the world (as it describes itself). I want to elaborate on how insurers, especially, have promoted better health and ask whether insurer use of genetic testing may also lead to better public health.

One example of insurers' public health role is disease management programs. Today, disease management is a billion-dollar industry—spent through insurance companies and disease management outsource companies. Disease management is performed directly for employers, sold to insurers or performed by insurers themselves. Over the course of the last 10 years, insurers have changed the chronic diseases they need to focus on. Asthma used to be a bigger issue than it is today. Proper preventive treatment for moderate to severe asthma today is well established? it includes use of inhaled cortical steroids on a daily basis. It was largely the pharmaceutical industry salespeople and the insurance companies that were doing disease management that educated the medical profession to adopt this standard of practice. Today, asthma does not appear to be as significant an issue as it had been. I believe that the actions of these two "hated" industries? the pharmaceutical industry and the health insurance industry? have changed the

treatment pattern for asthma for the better—toward evidence-based medicine.

Improving physician practices is also implicit in the Health Employer Data Information Set (HEDIS) measures. HEDIS measures include the appropriate use various pharmaceuticals and other basic evidence-based medical practices. These apply to chronic conditions such as asthma, coronary artery disease, congestive heart failure and diabetes. These quality measures represent a huge expenditure on the part of the insurance industry, employers and to some extent, the federal government. The HEDIS focus is one example of how the insurance industry promotes new technology and better physician practices.

The growing use of much medical technology is linked to reimbursement. This isn't the place to discuss the abuse of technology, which is real, but stable reimbursement has promoted beneficial new technology. Medicare has been fairly consistent about adjusting the Resource-Based Relative Value Schedule (RBRVS) as well as creating health care common procedural coding system (HCPCS) codes for new devices, and the American Medical Association has been fairly consistent about adding new common procedural terminology (CPT) codes for new technology. Funding has been available for new technology.

Payment for tests and vaccinations is an important public social issue. The government, through regulations and reimbursement policy, requires some services (such as child vaccinations) and encourages others through benefit mandates and reimbursement.

Insurers are even promoting genetic testing in a noncontroversial way. It is mostly associated with diagnosing particular kinds of cancer to fine-tune the chemotherapy or other treatment. Most people in the audience work in health insurance. If your company is paying claims for patients with leukemia or other kinds of cancer, your company is probably paying for tissue testing for genetic markers.

Let's apply the dynamics of adverse selection to genetic testing. Traditionally, we assume that the applicant knows of things that the insurance company doesn't know about and makes decisions based on that. The example everyone uses is the applicant knows his or her house is on fire, but the insurance company doesn't know it. The traditional ways of protecting are policy terms, underwriting, Medical Information Bureau, risk class patient and risk rating.

Genetic testing can have huge positive benefits, but information from genetic testing can threaten the stability of certain kinds of insurance. The adverse selection challenge is obvious—the people who test positive for diseases needing long-term care are more likely to buy LTC insurance. If that happens, the insurers could stop selling LTC insurance.

So, if you're a health benefits insurer, in the future you will likely be paying for genetic tests for someone who wants to see if he or she is inclined to get

Alzheimer's. The person likely to get Alzheimer's can then invest in long-term-care insurance, perhaps even from the company that paid for the genetic test! Insurance 101 teaches that a stable and competitive insurance industry is good for society, but genetic testing could sharply reduce the availability of LTC insurance.

I'd offer another kind of social benefit of underwriting, where underwriting serves the public by identifying hidden but treatable conditions. Currently, life insurers routinely test for hepatitis C –(HCV). HCV infects something like two percent of American adults under age 65. The disease can remain asymptomatic for decades, but it can progress to liver failure and death. HCV is largely undiagnosed because it's often asymptomatic for long periods.

Life insurance testing is one of the main ways people discover they have HCV. The disease is treatable and potentially curable through pharmaceuticals. Lifestyle changes, including giving up alcohol, can have a profound impact on infected individuals' health. This is an example of how underwriting and identifying conditions through life insurance, or through insurance in general, is in the public health interest.

I believe that the overlap of underwriting and the public good for HCV may extend more broadly to genetic testing. Genetic testing can help identify treatable or avoidable risks, which sounds like it's in the public interest. We need to explore that potential commonality between public health and insurance activity. That commonality may very well exist, but if we don't discover it, we could face poorly conceived legislation that discourages genetic testing and hurts both the insurance industry and public health.

I think that genetic testing will be part of the routine physical of the 21st century. If I'm right, we need to add the results of genetic tests to the list of what the applicant can know about but the insurer doesn't. That can certainly lead to adverse selection, as individuals imminently facing some particular high risk choose to insure themselves against that risk. However, as described below, several factors may moderate the risk of adverse selection.

My view is that genetic tests are going to be followed fairly closely by effective treatments, given the rapid acceleration of medical technology and medical treatments. The connection between the two and the incredible acceleration in the progress of medicine suggests treatment or risk amelioration will quickly follow many of the conditions that are newly identified.

Can genetic tests really tell the future? I think for most diseases the answer today is "No," and it's going to continue to be "No." The public is likely to overreact to the results of genetic tests. That could actually be a good thing for the insurance industry, depending on what kind of insurance you're selling. I use the term "likely low specificity." Kathy used "low penetrance." That is, someone who gets genetic test results saying he or she is likely to die of heart failure or cancer (I could

probably guarantee that now for most of you) is likely to overreact and run out and buy insurance. Genetic testing could lead to a surge of insurance buying!

On the other hand, depending on what kind of insurance you're selling, this is potentially a huge threat. If you're in long-term care, if you're in long-term disability, people with the "clean" tests will avoid buying some kinds of insurance, especially if they also have healthy lifestyles. People who are positive for some of those conditions, or who begin to reflect on what their parents had or grandparents had, can cause adverse selection.

Particularly scary to me is the experience we've had of unintended consequences of well-meaning legislation. We're coming up on the 35th anniversary of the federal Medicare program. Medicare benefits are still based on the benefit wisdom of the 1960s. There's no prescription drug benefit in Medicare. What Craig described as the "horse and buggy" applies to amending legislation. The rules that are being set now for genetic testing, on an emerging science, could be with us for a very long time. I think that could profoundly affect what insurers are allowed to do, the kinds of products they sell and the profitability of different lines of business.

Our industry, our profession, needs to identify a common ground for this emerging science that is, I hope, an unqualified good for public health, the future of our individual health and the insurance industry. To identify people at risk for treatable diseases is a common public good.

We also need to emphasize in our industry the importance of avoiding instability as medicine and genetic testing accelerates. Americans are largely underinsured, particularly in disability and long-term care. Instability in the industry will likely make the underinsurance problems worse.

Please think about these issues with respect to the products that you work with. Think about the risks of adverse selection and the advantages of genetic testing. Please support the efforts that Cecil Bykerk and others have made in Washington on behalf of the American Academy of Actuaries to get actuaries involved in this great issue of our times.

Do we have any questions?

MR. MARK BROWN: Dr. Hudson, you talked about the speed at which different tests were being developed and so forth. A lot of those tests are purported to be replacements for existing testing and diagnosis, but practitioners don't seem to accept them at the same pace at which they're being developed. I'm wondering about your comments or feelings about acceptance of the newer tests, and how quick is the learning curve, so to speak. The second thing is that a number of those genetic tests are only done by certain labs, and is that due to patents or other things? I am curious as to the panel's take on future options, such as being able to get those lab tests done and diagnosed so there's competition for price.

DR. HUDSON: I think there are real issues in being able to ensure that health care professionals know the spectrum of options available and the strengths and weaknesses of those tests. There are a number of efforts ongoing to try to educate health professionals about genetics. Certainly that's being facilitated because of an educated public that's walking in the door saying, "Hey, I heard about it." I think that there are some genetic tests that are replacing and improving on old-fashioned or older technology, particularly those tests for the diagnosis of a currently manifested disease. If somebody walks in the door and is sick, a genetic test can be a rapid way of distinguishing between a range of possible etiologies for that particular disease. But I think increasingly within a particular disease, say a certain form of cancer, genetics is able to stratify so that colon cancer is no longer one disease; it is a number of diseases that can be distinguished by the underlying genetics. Underlying genetics can have important consequences for the appropriate treatment and also what the likely outcome will be for that individual..

Your other question was about labs performing the tests. It is certainly true that there are some tests that are only performed by individual laboratories. The control of those tests is governed in large part by who has the patent on the test and what their licensing procedures are, so BRCA 1 and BRCA 2 testing is being offered exclusively by Myriad Genetics, except in research protocols where they're providing that at a reduced cost or for free. I think that is an issue that providers need to know? who's offering the testing? and then that there's not a lot of competition in the marketplace for some of those tests.

MR. BURTON: The only thing I would add to that—I think some of what you see as a lack of competition is the fact that this is a new area, this is a new science, and when you're talking about "old" tests, you're talking about tests that are a couple of years old, and not things that have been fully integrated into the practice. I think some of this will be addressed as the science develops further, as the market expands and as there is more competition in there.