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Georgia Wiesner
Case Western Reserve University

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GENOMIC MEDICINE: THE HUMAN GENOME PROJECT FROM A HEALTHCARE PROVIDER'S PERSPECTIVE

GEORGIA WIESNER¹

I'm delighted to be here today, and I want to thank Professors Davis and Engel for inviting me. The other thing is, I want to apologize for my voice. I'm hoping that my laryngitis that I developed yesterday will allow me to complete my talk and my comments.

I also just wanted to parenthetically say, whenever Dr. Zahka was mentioning something that I might have an opinion on earlier this morning, it wasn't that I was being too shy to mention and make any comments, it was that my voice wouldn't have carried, and if you all know me, you know how frustrating it was to sit back and not be able to have this discussion.

Anyway, I wanted to begin today by telling you what my role is, and what I thought my role would be, because we've begun with a clinician and we've ended this morning with another clinician, and it's really to give you another viewpoint about how genetics actually works within healthcare, but with a specific goal of talking about what those consequences might be, and how those consequences really affect both the patients, their family and society as a whole.

So I thought it would be best to frame my comments back on some scientific information and frame it against the Human Genome Project that we've actually discussed several times this morning, but to put it within context as to what this genetic information actually tells us and how we actually then can use this information to move forward.

The Human Genome Project really began a number of years ago as an international consortium where the goal was to sequence and identify all of the genes in the human genome.² So that was the major goal, and as a matter of fact, this year, they were able to publish the first draft sequence, so all of the genes within the human genome at least have been published in what we think will be a preliminary fashion. But there have been some surprising events of this particular endeavor, and some of the surprises have been just in our number of genes, where the estimates of genes had been between 60 and 100,000 genes that made humans "human," and we now estimate that it may be about 35 to 40,000, and as my slide shows, these estimates change almost on a daily basis, so it depends on which study comes out at the latest time to actually see how many genes do we have.

The other important point is that all human groups really have a great deal of similarity in our genomes. So as you can see, I use the phrase "human groups" rather than "racial groups," because the concept of race is now under a lot of

¹M.S. Biochemical Genetics; M.D., University of Minnesota. Ms. Wiesner is an Associate Professor of Genetics and Medicine at Case Western Reserve University, and is the Clinical Director at the Center for Human Genetics and the Medical Director of the Cancer Genetics programs at University Hospital. Dr. Wiesner's research includes the study of the genetics of cancer and the use of genetic testing in clinical practice.

²[Http://www.science.doe.gov/ober/hug-top.html](http://www.science.doe.gov/ober/hug-top.html).

discussion, because of the Human Genome Project and our understanding of what makes people people is one of the most exciting things that we've been able to do at the end of this century and going into the next century.

But the Human Genome Project from my point of view as a healthcare provider has really been on what advantages and what advances can we be able to provide from that. So we really learned a lot about how genes work, how they talk together and how we actually have both health and disease as a consequence of the Human Genome Project. So really understanding this complex interaction is one of the most exciting things as well.

What this really has done for healthcare is allowed us to individualize our healthcare. To be able to say for one person against another that their genes might, in fact, give them a basis to be healthier or actually, might have more problems with illness. So this individualization is what's really new and what's been important.

So I want to introduce another concept, and that concept is one of genomic medicine, so it may not be something that you've heard before, but it's been used as a way to try to describe, how do we use this genetic information that we get from the human genome, and then actually translate that into patient care.

So what I have up before you is just a small quote from Dr. Art Beaudet's 1998 presidential address to the American Society of Human Genetics, where he essentially defines genomic medicine as the routine use of genotypic analysis, usually in the form of DNA testing, to enhance the quality of medical care.³ So it's not testing for testing sake, it's testing to actually use that in an active way to take care of our patients and to even advance medicine.

So what I wanted to talk a little bit about was what does this actually promise for us, where do we go with that, and then talk about some of the problems that we've encountered in that promise.

So genomic medicine is now to be used to say for an individual person that they may or may not have specific risks based on their gene type or what their specific (inaudible). So why do we want to do that?

Well, in one case it's a way we can now translate medicine into a preventative medicine rather than reacting to disease after it's happened. We're hoping we can reduce the incidence of diseases. We can improve diagnosis and we can develop therapeutics based on the genomic and proteonomic pathways that we now are discovering on the basis of the Human Genome Project.

Proteinomic actually means the study of the proteins or gene products and how they interact together. So the question right now is, what is the problem and do we have a problem? The answer is, yes, we do have a problem, because this little man is actually pondering why he's doing a genotype now instead of an x-ray of his patient. He's looking at these genes and pondering what he can actually do for this individual.

But our real issue is how do we provide the quality healthcare that really balances the risks and benefits we've been talking about today in providing and translating the genetics, and this is important both for the individual we're talking about, it's

³Art Beaudet, *ASHG Presidential Address: Making Genomic Medicine A Reality*, 64 AM. J. HUMAN GENET., 1-13 (1999).

important for the family member of that individual, and it's important for the society as a whole.

Now, remember that the Human Genome Project was one of the only projects that has set aside funds to be able to look at the ethical issues and social issues that have come out of our genetic discoveries, and so I wanted to put these promises and problems into some sort of perspective for us.

So first, we talked a little bit about the promises, and then I want to outline some genetic myths, things that we all carry around with us that we think are true when, in fact, may not be, and then talk about how those myths have really translated into barriers for people to be able to provide care and uses them on the basis of genetics.

The first genetic myth that I want to talk about is the concept of genetic determinisms, and I want to speak a little bit about this because I've heard a lot of deterministic talk about our genes, our genome today.

One thing that we've always thought of is that there is a genetic defect and defects are called mutations when you talk to geneticists, that a defect when it occurs in a gene, that that will inevitably lead to a disease or a disease process, and we know, in fact, that is not true. But (inaudible) what we think about that sort of deterministic information into our general talk and our general understanding of what our genes do, so we need to be very careful to go back and say, "No, it's not just one gene. For most diseases, it's many. It's the interaction of those genes and it's the interaction with the genes with environmental influences.

This also says that we are not as able to make predictions about disease and health as we would like, so we really can't make those predictions about what the disease process is going to be and our ability to do that is very clouded. So that's one myth that I think we should all be very cautious about.

The next genetic myth that I wanted to mention was that the concept out in the community that genetic conditions are so rare and so unusual that it really doesn't touch most people in the population. The fact that medical geneticists will treat adults that have adult onset diseases is surprising to some people, that rare diseases of childhood are the things that we learn from. Well, we've been able to apply that to very many important diseases that occur later in life.

The other myth that people carry around with them and think about it in terms of their genome is that the diseases of adulthood themselves are caused merely by lifestyle, so it's our choices that we make, such as smoking and diet and lack of exercise and it's not our genetic makeup. When, in fact, this may be a true concept and it may not be true, and we believe now that the susceptibility towards health or disease is really one of those factors that we need to understand, and it's the interaction with the other environmental factors that are most important.

So the next slide is a list of common diseases, and this is from the CDC in 1998, and is a list by percentage of death of that year, and the most common causes of death within the adult population, and you can see heart disease is number one and cancer is number two, and then it goes on to chronic liver disease.⁴

The reason why I have injury, which is smack dab in the middle, which is about 4 percent, highlighted in green, is because that's the only one I think that we could

⁴Sherryl Murphy, *CDC Deaths: Final Data for 1998, 48 Vital Statistics Report, No. 11*, July 24, 2000, at 5, at <http://www.cdc.gov/nchs/products/pubs/pubd/nvsr/48/48-19.html>.

argue about that does not have a genetic component. All of these other diseases have some genes that we actually know about now that will contribute to the process of these diseases.

These are common diseases. These are diseases that we know many people have, and they're chronic in the fact that they need to be actually cared for, or those individuals cared for, over a long period of time. So it leads us to know that our genetic load is potentially quite large for the population, and if we're focusing on just one set of genes or a particular gene, we may be not understanding our whole scope of what our genome component and our whole genetic load might be on the population.

So these ideas and these myths that we carry around have actually led to several areas, and I want to talk about the genomic barriers next.

So I actually separated these barriers into two, just general groups, and what I'm talking about here is access of individuals to get a better genetic understanding of what their situation might be, and I put these down into medical problems and what I consider to be non-medical problems.

The primary medical problem that I see is really lack of knowledge, both by the provider and the patient, and I want to talk a little bit about that in a minute.

However, the reason why we have such a lack of knowledge is because there's a ton of evidence on the basis of whether or not genetic tests are efficacious or accurate or actually test what we believe they should test, and so it leads us to a problem in terms of knowing how much we can predict on the basis of a genetic test, when the studies that should provide us that information have not even been performed, nor are they even planned for the near future.

And in contrast to the medical types of problems and barriers, there are non-medical barriers, and I call these more societal barriers that the patient might bring to the table or the physician might have themselves.

One thing that we've been able to talk about this morning is whether or not genetic discrimination exists. I'm not able to really speak to that, but what can I speak to is the fear of genetic discrimination is great and it is across the board and in most patients that actually come to see us at the Center for Human Genetics. So this is a problem, whether or not it is actually fact, that we actually need to deal with.

The second non-medical barrier to genetic testing and understanding genetic information is a lack of insurance coverage for a genetic condition, or a genetic conversation, or for a genetic test, and the insurance process is one that's quite cumbersome as it relates to genetics. It's quite confusing for the patient.

And from a patient point of view, patients would like their insurance to behave like their insurance in which they go to the doctor and they get some form of payment or support for seeing most specialists and it's not always true with genetic conditions or as far as seeing a geneticist. And then the third barrier that I want to talk about was privacy of results and how people feel about their results and where they should reside and who should have this sort of information.

So let's go back a bit and talk about lack of knowledge. There are many, many, many, many studies, only one that I want to share with you, that points to the providers' misunderstanding of genetic information, and I thought that this particular study was particularly interesting in terms of genetic discrimination issues.

So what they did was survey of group of what we call "primary care practitioners." Most of them were OB/GYNs and some were internists, and they surveyed them about their use and understanding of genetic tests and susceptibility

for cancer, and one of the good things about this study was that over 70 percent of this group took a family history for cancer and actually felt comfortable in providing that information, but there is a but to that sentence.

Only 21 percent of these people that took that family history and said they could recognize that would actually send theirs on to a medical geneticist to actually talk about testing and talk about evaluation. So a minority of these providers would actually send them on, and I think the next two points will explain part of why that is.

First of all, almost 70 percent of them were concerned that the genetic test results might have a negative affect on their patients, and I think most people don't know how to handle that. Nearly 80 percent of these providers were concerned themselves that the genetic test result might be used by the employer or the insurers to discriminate against the patients.

So if the physician does not feel that it's a safe situation for their patients to go forward and have this sort of conversation, they're not going to be for their patients unless it's a barrier to care for patients who actually need that information.

So what about geneticists that actually deal with this problem, what do they think? That is a study that was done of 163 cancer genetic counselors from the National Society of Genetic Counselors. This group actually has a list serv e-mail that people can subscribe to that is very, very active. Most of the genetic counselors that provide genetic counseling services for cancer belong to this list serv interest group, and so they're quite a knowledgeable group, and if anybody would know about this, they would be the group to go to.

And so of the survey they asked individuals whether or not they, themselves, would be tested under two conditions: One was for breast cancer testing for the BRCA1 and 2 gene; and, the second condition was for colon cancer risk for the HNPCC gene. An HNPCC is a common family syndrome that gives you risk for colon cancer.

So 85 percent of these genetic counselors that worked in cancer genetics said that they would be tested for are BRCA testing, and 91 percent of them said that they would be tested for the HMPCC, so there was an active understanding of how these tests might be used.

However, look at the next two points. Sixty-eight percent said they would pay directly out of pocket and would not involve their insurers, and 26 percent said they would use an alias when doing the testing. An alias obviously is renaming oneself, going into the clinic and then having the testing done out of pocket and out of the insurance eye.

So these are genetic professionals who, as they approach this problem, make such decisions about that.

Well, what is our experience at the Center for Human Genetics? When I was actually preparing for this talk, I went in to say, well, this is a good time for me to know what actually happened [inaudible]. I will tell you that there is little objective data. There's a lot of subjective data, but no real objective data on what people are actually doing. So let me just share with you a little bit of a retrospective study that we've done.

And again, I do the cancer susceptibility counseling, and that includes women that come for breast cancer counseling, and we reviewed 212 cases from 1995 to 1999, and then we've looked at a number of factors for those. But what I want to share with you is, of these women that came in, each and every one asked about

insurance and genetic discrimination. It is the number one question that I get in addition to, "Will I develop cancer?" So it's a very, very important issue for people, and everybody asked that question.

The other thing that we went and looked at was just to say how often did people pay out of pocket or change their name in order to come through, and it's been pretty constant over the years. About 10 to 12 percent of those individuals will pay for their services out of the pocket, which include our services for counseling, the testing and then the post-test counseling. So for some women, that's actually a quite high dollar amount.

Now, when I went back and looked and said, "Yes, I want to go through testing," and then they would go through the insurance process to say whether or not they would test, about 36 percent of that group, a little over a third, ultimately decided not to test. So these are women who said, "Yes, I want to be tested," and ultimately didn't go through with it.

And so I have some answers as to why that might be. Some people came back and said that their insurance coverage was not adequate enough and they were not able to bear the costs themselves. Other people said they really thought about the issues of genetic discrimination and they weren't ready to have the test done when it was such an unsure field at this point. Some people came back and said they didn't quite know how they were going to use the test result, and then other people came and said, "Well, my mother didn't want me to, my sister didn't want me to," and it suggested a lot about family dynamics.

As a matter of fact, this is just a listing of what I know some of my patients have said, and what we're doing right now and Michelle Merrill, who's a genetic counseling student, who is in the audience today, is doing a project where we're looking at these issues and looking at the insurance issues as they relate to genetic testing.

So I have one more slide, and just to summarize, number one, there's been a wide impact on healthcare dealing with concerns about genetic discrimination, both on the cost issue, "I can't get that test because I don't have the money," "I don't have insurance," and also, on, "I don't want my insurer to know what the information is."

The problem with all of this is it alters the physician and patient relationship, and remember, that's essentially a contract between two people, in which the physician says, "I will work with you to take care of your healthcare problems," and the patient says, "I'll kind of do what you tell me to do and then we'll work together," so it really is a contract.

What the problem is, is they come to me and say, "Don't tell my surgeon, don't tell my OB/GYN, don't tell these other people about my genetics." These other people are hampered because they actually cannot provide the level of care that needs to be because they don't know what the risks of those individuals would be. So that, I think, is a real issue.

So my last point is, we all need to become more literate in genetics and our understanding of genetics, both in the healthcare and in the education so we can improve our understanding of our genome and our understanding of genetic health.