Public Policy Issues Surrounding Personalized Genome Services
Kathy Hudson, Ph.D.

DR. TEUTSCH: The public comment period will begin right after lunch. I expect that will be around 1:25 to 1:30. I apologize to all of you. Hopefully you can all stay so we can be the beneficiaries of your thoughts.

MS. AU: Our next presenter is a frequent visitor to SACGHS, Dr. Kathy Hudson. She is the founder and director of the Genetics and Public Policy Center, located in Washington, D.C. Dr. Hudson's presentation will explore the public policy considerations for this emerging field of direct-to-consumer personal genome services.

As soon as we get her talk cued up on the screen, she will drive us through this.

DR. HUDSON: I want to thank you for inviting me. I recognize that many of you are probably suffering from low blood sugar, and so snack carts will be coming through the aisles. Snack boxes are available for $5, wine and beer for $3.

[Laughter.]

DR. HUDSON: What I would like to do in the next 15 minutes -- I will go as quickly as I can -- is to put a little context around what you have heard already today. I will talk explicitly about some of the tensions that have been coming up recurrently over the course of the last day and a half. Then I will talk about some of the critical policy issues that I think we are facing today.

The whole genome association studies and the kinds of companies that we heard from today really fall at the end of an evolutionary continuum in the kinds of tests that have been available directly to consumers over the last decade or so. Of course, the focus that we have heard today has been on health-related genetic testing. Beyond looking at SNPs, we can sequence entire genomes, as we heard from George earlier today.

The companies that we heard from are really only a subset of the companies that are offering health-related testing services in this space. This is a now outdated slide. We have a more updated version available now, I think, on our website where we categorize the companies in terms of what kinds of tests they are offering and focusing mostly, again, on health-related testing.

So, how do we look at this new paradigm in genetics. I think what we have heard recurrently and what we read about in the newspapers and in the medical journals is sort of this tension between the old precepts of genetic medicine and this new concept of personalized genomics. I want to go through these precepts really quickly because I think they are important in what we have been hearing over and over again as some of the key stress points.

The first is that genetic testing requires pre- and post-test genetic counseling. The second is that you need a healthcare provider in all genetic testing. In the olden days genetic tests for non-actionable conditions were considered the highest risk. So when the predecessor to this Committee put tests into categories of what was most high risk and therefore warranted the highest degree of oversight, it was highly penetrant tests for which there was no intervention available. I think we actually might flip that today. And, that genetic information is special.

I think we can challenge all of these precepts today. First of all, in terms of pre- and post-test genetic counseling, there are too many genes, not enough genetic counselors, it is too expensive, and a model that was really built on reproductive genetic testing and Huntington's disease may not fit with the kind of testing that is available today.

In the olden days we had odds ratios of greater than five and they were extremely rare. Today we have these teeny tiny odds ratios and they are proliferating like crazy.

The second precept, no testing without a healthcare provider as an intermediary. As Ryan mentioned earlier, not all genetic tests are created equal. They pose different risks for interpretation and for intervention. It may not be a viable model given high levels of consumer interest. There are inconsistent state laws, and I will come back to this, about who can offer a genetic test.

Third, about actionable or non-actionable information, we have heard a reference to the Reveal Study in which results were provided back to people about their Alzheimer's risks and in fact there was no demonstrable increase in anxiety, jumping off of bridges, et cetera. So people can handle this information where even there is nothing they can do about it.

In fact, I think we really need to focus our attention on the validity of tests where there is something you can do about it. If you are going to take a drug or not take a drug, have surgery or not have surgery, the validity of those tests is of utmost importance.

Lastly, genetic information as being special. I think we in genetics of course think genetics is special, but we worry that the public thinks it is special, genetic exceptionalism or determinism. I think that while that may have been at one point true among the general public it is no longer true. When we did a survey recently of nearly 5,000 people and asked about genetic information versus risk information, generic information, there were no discernible differences between people's appetite for that information and concerns about that information.

In the old days we had this very systematic way of translating genomics or translating biomedical research into health impacts, and now we are leapfrogging over some of those essential steps, or what we used to view as essential steps. We are in the midst of a wave of creative destruction which is making many of us uncomfortable.

So, what should our response be. We could get our genomes done, and some of us may have done that. We could start our own company, although we may be a little late.

[Laughter.]

DR. HUDSON: And as someone said yesterday, you have to be blond.

We could ignore the companies. As one well-known genomicist said, they are just a nuisance. We could insult them, and that has certainly been done in some quarters. Or we could support needed policy changes.

We have heard a lot about the promise of DTC genomic and genetic testing. I won't go through that. I want to focus a little bit on some of the concerns that have been expressed about DTC genomic and genetic testing, about consumers not being able to understand it, about consumers being especially vulnerable, about consumers getting tested without thinking about their family

members, and forgoing standard treatments or getting unnecessary treatments, as Francis brought up in the last panel discussion.

The essential point I want to make here is that this is all knowable information. Instead of speculating about this, this is knowable information and we should be supporting studies to actually get this information.

On the more policy side of the equation, there are concerns about the adequacy of privacy protections, about the validity of the tests, about the competency of the laboratories, about the evidence to support the claims that are being made, about the protection for research participants, and actually, an issue that I don't think has come yet in this meeting. With DTC testing especially with buckle swabs where you are sending it off and there is no person in front of you, there is a possibility for surreptitious testing of somebody without their permission.

This Committee knows well that there are enormous gaps in the oversight of the quality of genetic tests and has made some really fantastic recommendations in terms of policy actions that are now before the Secretary. I won't go over those because those are very familiar to you.

I will point out on the slide that I mentioned that there is no HHS authority over false claims being made by companies. There has been no FTC enforcement action, although I do understand that there are now some investigations underway. That is an interesting new development.

An additional problem that we have in terms of oversight of genetic testing is some lack of clarity in terms of who is authorized or should be authorized to order and interpret tests, the limited applicability of HIPAA, and the limited applicability of the Common Rule for Protection of Research Subjects.

I will spend just a second now talking about protection of research subjects. In the late '70s we put in place the Common Rule for the Protection of Human Research Subjects. That was really based on principles of beneficence, justice, and respect for persons. We may need to go back and reevaluate whether or not a model that was somewhat paternalistic and protective of physical harms really fits in today's biomedical research context.

But it is important to note that the common rule does not necessarily extend to all research that we would care about, including some that is being conducted by DTC companies.

Outside of the federal government, which I believe plays a very important role, especially in quality and in making sure that claims are consistent with the evidence, there is certainly an opportunity for professional groups and industry to develop guidelines. Those have the advantages of being flexible and those groups having the right expertise, although they may have some internal conflict of interest. Those guidelines would be voluntary. That means that not everybody has to participate and abide by the rules.

There have been a number of professional statements. I'm particularly fond of this one, since I was involved in its creation, from the American Society of Human Genetics which really says that some tests are appropriate for being offered directly to consumers but we need to make sure that the tests are accurate and reliable and that the claims that are made about them are also accurate.

ACMG, a few months ago, took a different position and recommended that healthcare providers be involved in ordering and interpreting all genetic tests. This was basically putting a stake in the ground, saying we, the medical geneticist community, need to be involved in this testing.

More recently still, the American Medical Association actually provided an interesting recommendation and provided some interesting clarification, I think, about what does it mean to have a doctor involved. The AMA recommended that states restrict the performance of clinical and laboratory genetic testing to individuals under the personal supervision of a healthcare professional. That is not somebody you have talked to that has signed off on your requisition. It is somebody who is actually overseeing your care.

The states have always had a role in laboratory testing and have recently made the entire scenery more interesting. They administer the Clinical Laboratory Improvement Acts, and they can impose higher standards than CLIA requires, such as New York. Ann Willey is here from New York. The states [also] determine who is an authorized person to order and receive laboratory tests.

This is an evaluation that we did some time ago on the state laws regarding who is an authorized person. The dark purple states are those where an authorized person is usually not defined, which means everybody is an authorized person. [That] means I'm an authorized person, so I can order my own clinical laboratory testing.

Lavender is limited, and you will notice that California and New York are both limited. I will come back to that. In the whitish, DTC is not permitted at all. A healthcare provider must be involved.

Where DTC is permitted, as I mentioned, the state laws are usually silent on the issue. Where DTC is not permitted, and one example is Georgia, they are very clear that it has to be a licensed healthcare provider who is authorized by law to use the findings. So I as a consumer am not authorized by law to use the findings.

Then there are the mixed states. Both California and New York fall into this category. This is an excerpt from the California statute. There are prohibitions with exceptions, and it is the exceptions that put it into the limited allowance.

In the absence of federal leadership in a number of areas in genetic testing, particularly oversight of quality, the states have stepped in. We have seen headline after headline after headline about the states trying to step in and protect their citizens.

What are the policy options as we move forward. We could take the stance that is a buyer beware marketplace and consumers should be informed about what to take into consideration as they consider buying these testing services. We could demand transparency, as was embedded in the Oversight Report in terms of a genetic testing registry which would include information and evidence that supports the test, how it is performed, and its characteristics. We could require third party review.

We could be taking action against false claims. I believe that we could do that both at the federal and state level and perhaps in a voluntary way outside of government.

We might want to think about creating a category of laboratory-developed tests that would be the moral equivalent of over-the-counter drugs. If we can now go into the drugstore, and we know

what we have, and buy that drug over the counter without a physician, aren't there also tests that should be similarly accessible to us without having to make an appointment, go see the doctor, and by the time you have actually gotten to the doctor and taken time off work the thing is resolved. I think we might want to think about this as a new mechanism.

HIPAA. People within the Beltway know what a HIPAA-covered entity is, which is sort of pathetic.

[Laughter.]

DR. HUDSON: Companies that do not bill electronically are not HIPAA-covered entities. So, at least federally afforded and state-enforced privacy protections usually don't go with the information. I think these companies, particularly 23andMe, selling a service at the same time that they are conducting research raise interesting and provocative issues about what we need to do to protect research subjects or research participants in this new age of personalized genomics.

That was really speedy. Now we can hopefully go to lunch pretty quickly. I want to thank my funder, the Pew Charitable Trust.

[Applause.]

MS. AU: Because Dr. Hudson was so speedy, we can take one or two questions. Kevin.

DR. FITZGERALD: You have to have quick hands around here.

[Laughter.]

DR. FITZGERALD: Thank you, Kathy. Again, as always, very thought-provoking, so I have a question. You mentioned in the statement that you were one of the authors of that safety should come first. You also mentioned that there is a great deal of evidence we need to have in order to make decisions that we don't yet have. Is it then logical to conclude that much of what is going on now you think shouldn't be going on because we don't have the evidence to decide what is safe and what isn't?

DR. HUDSON: I think it is hard to know. The actual answer is it is hard to know. If you look at some tests that are being offered, you can't tell what the gene is, you can't tell what the variant is, you can't find any publications. In some cases, the disorder that is being tested for doesn't exist in the scientific literature. It is just very difficult to know because we are not demanding the kind of transparency that we really need.

DR. FITZGERALD: Right. If you are saying safety first and we don't know, does that mean don't do anything?

DR. HUDSON: There are a couple of interesting models. One is, we could put in place this genetic testing registry tomorrow. It is not that complicated. I think we need to move ahead expeditiously with putting that in place.

Secondly, I think that we could have tests on the market where we either haven't had a chance to evaluate them or we don't yet quite have all the evidence and collect evidence as we go forward. It is approval with additional evidence collection.

There are ways where we could address the pursuit of the perfect not denying us some of the good tests that are available out there.

I should say there is a whole slew of tests that have been out in clinical practice and validated for a very long time that we need to figure out some mechanism of just grandfathering those in. We know what they do, we know how they behave, and we know the molecular biology and cell biology underlying them.

MS. AU: Thank you, Dr. Hudson. I'm sure we will see you back here again.

[Laughter.]

MS. AU: Back to Steve.

DR. TEUTSCH: Great. Thanks, Kathy. Thanks, Sylvia, to you for organizing this session.

[Applause.]

DR. TEUTSCH: Obviously, a stimulating and important area. We will be talking more about it later this afternoon.

For now, since you and Kathy graciously got us done by 10 to one, let's plan to meet back here at 1:20. We will have a half hour. There is a cafeteria down the hall for those of you who don't have a boxed lunch. Then we will take up the public comments. Thanks, all.