Roundtable Discussion
Committee Members, Speakers and Discussants

DR. McGRATH: Thank you, Judith. That was a mind-full.

I'm really happy to open up the table now to use this time to address questions to our guests, who traveled far and wide over short notice to come here and talk with us.

DR. TUCKSON: I just want to give one question as you do this, just for you all to think about. I was trying to follow the discussion as best I could. I just want you to be thinking. This is not necessarily right and you probably have your own constructs or your own notes that are better than mine.

But I'm asking myself over and over again, is there a problem. Is there a problem here that deals with genetic exceptionalism or with just medicine? So, is there something about genetic exceptionalism here, and is there a problem.

Is there a problem with availability of expertise by disciplines, by diversity? Is there a problem because of the integration of expertise into daily clinical practice, whether that is the individual doc or professional level or the infrastructures of coordination across disciplines. Is there a problem.

Is there a problem because of compromised patient care. Do we have any evidence to find out whether this is a big enough issue because somebody is not getting good care as a result of this not being as optimal.

The second big set of questions for me is, is there something, then, that the Secretary can do. Is it through connectivity to others of our reports, like the Oversight discussion we are going to have, or through the Coverage and Reimbursement Report, which has a whole section on this whole idea of who should get reimbursed for what and that whole big thing which I opened up around that genetic counseling deal.

Is there something the Secretary can do around CMS in terms of payment. We will not pay for thus or so unless you have proven that you have kept up with your level of education. In other words, it may not be that you need to beg and plead somebody to go to a CME course or whether they have gotten their boards. If you don't have your board certification and you haven't kept up with the board certification for family medicine that includes a rational genetics one-on-one, you don't get any bucks. You could be that draconian. I'm not saying this is what we should do.

The last area is, is there something that the profession should be doing themselves and that you use our bully pulpit to urge the profession to do, whether it is, again, the specialty societies, the boards, and what not.

Anyway, I'm not sure it was helpful, but I just wanted you to have something in your mind as you try to think through this now. You are supposed to be asking questions that get you to a conclusion. So don't ask questions just because you are interested. You only get to ask questions that will take you to, in your own mind, a yes or no about whether you want a subcommittee and what you are going to charge that subcommittee to do if you do it.

MR. MILLER: I would just add two questions focusing on framing it. What is the role of SACGHS in this discussion. Also, I'm always interested in what are the metrics that we look to

in terms of education. You talk about a dearth of folks, but what are the metrics that we should be looking at.

I have two points, I guess. One is, a number of you mentioned diversity issues, and I acknowledge and agree that that is an important piece, but none of you mentioned it. I would encourage you, when thinking about diversity, to also think about people with disabilities in the profession. In fact, people with disabilities are a medically underserved and underrepresented population when it comes to the health care professions and particularly with respect to genetics.

And, to think of people with disabilities as simply patients as opposed to a community that brings something to the table in terms of understanding the experience of living with a disability or having a disability is something that is very unique and equally valued with respect to diversity issues with respect to race and gender.

My question, though, is I want to piggyback on something else that Reed said in terms of this question that he asked right before the break in terms of who is qualified to make judgments about this. We have all these different groups that are thinking about genetics in different ways. I'm wondering whether there is a role for the SACGHS in bringing groups together and talking about what are qualified genetic professionals. How do we think about genetics in health care and whether the overarching group of this Committee is something that can bring the individual groups together to talk about that issue.

DR. McGRATH: Muin.

DR. KHOURY: First, I would like to thank everyone for this wonderful tour de force this morning. I'm glad to hear Judith's talk about the reorientation of ACMG to translating genes to health. That was really music to my ears. I can go home and be happy about that.

But as we make this transition, so to speak, from the paradigm of taking care of people with genetic diseases and their families and their communities, et cetera, to how to deal with genetic information in general, whether it is microarrays or gene expressions or taking drugs in the practice of medicine and public health, I would like just to get your thoughts about how you think this should be done given the lack of work force.

The numbers are not going up, if anything. The information is going up. I like your statistics. One lawyer per 17 residents of D.C. seems a bit steep. But if you think about how many geneticists you need per snip --

[Laughter.]

DR. KHOURY: -- or per base pairs, those numbers could be a little bit too much.

What is the role of the new genomic person or genomic specialist, whether it is a genetic counselor or a genomic counselor, nurses in genomics, the medical geneticists? What do they have to do to translate genes to health? What is the role of evidence-based practice and how can the new geneticists embrace the concept of evidence-based medicine and use it in translation?

Without that, there is really no payment for services. Let's face it, that is the current model here. It is not the traditional model of genetic information as valuable per se. We have to sell why is it valuable to improve health, or at least metrics of health.

DR. FRIES: Can I comment a little bit on that? I am an epigenemian as well as a geneticist, so I can look from the different roles of the different fields.

Evidence-based medicine, for example in OB/GYN, is largely based on large studies that have been made of practice patterns, whittling down things that have been done from just simply "We have always done it this way" to actual evidence that this makes a difference.

Part of the difference with that in genetics is that when you are looking at it as a residency-based specialty, since 1992, it has really not got that body of practice information.

So you would have to incorporate some of all specialties' practices and incorporate genetics into those in order to assess that. To ask genetics to screen itself for what its best practice guidelines have been is going to be based on a limited number of experiences on patients. So it is going to need a different sort of developmental pattern.

However, that doesn't mean that it can't be done. I think the way that it has to be done is the way that it is done anywhere in medicine. Look at innovative strategies and then set up large sponsored trials of those innovative strategies as a comprehensive group. I think that that is an area where this group could be very influential both in funding and support of those kinds of strategies.

DR. McGRATH: Mara. I'm sorry. I was going to go Mara, then Joseph, then Julio. Oh, I'm so sorry. I missed that. Of course.

DR. KAHN: There are two questions on the table. There is Dr. Tuckson's question and there is Dr. Khoury's question. This may be the only thing that I have to contribute to this conversation, is the answer to this question.

You asked what is the problem and what can the Secretary do about it. I would give you a simple take-away. I think the problem is integrating genomics into the daily practice of whatever our professions are, the daily practice of health care. It is about taking care of people.

What can the Secretary do about integrating genomics, which is not well integrated, into the daily practice. I would suggest to you that the most important thing the Secretary can do is focus on decision support.

Now, there will be a component of education that is necessary. If decision support is integrated into electronic health records or even into office practices that don't have electronic health records, there are other ways to get decision support through the Internet. They have Internet connections.

The clinicians are going to need to be educated on what is in that decision support, and that is really critical, but that clinical decision support at the point of care is where people are being taken care of. That is how you really integrate this information into clinical practice.

MS. BENKENDORF: I just wanted to make a comment about decision support. I think our newborn screening ACT sheets and algorithms were mentioned, and I just want to tell you where that is going. The idea was, obviously, to get these into the hands of the primary care providers. So the newborn screening laboratories do send those out with all positive test results. The next round of ACT sheets and algorithms that are going to be developed are going to be for genetic tests commonly ordered by non-genetics physicians, again to be disseminated by the laboratories.

We are doing two things. One is that AHIC identified the ACMG and these ACT sheets as a prototype, and they are going to be integrating them into medical records as a point of care education tool through their genetic and genomic testing initiative. So we are going to be evaluating how that works.

The other thing is that the ACMG has obtained funding to convene a meeting, which will probably be this spring, of all the EMR industry folks to talk about decision support tools.

DR. TUCKSON: As the Committee continues to deliberate on this, recognize that we will be hitting these issues again under the Oversight Committee conversation. So there is a considerable part of the things just discussed in the Oversight Committee, so know that we have more than one chance to go after this.

MR. RACKOVER: There is a quick comment I need to make as an educator. We need to get the GINA bill passed. I'm tired of being able to talk with students but they won't get past that. So everybody at this table, before we leave Washington, should make a phone call. Without that, the students don't have to hear that they are concerned about the ethics behind the genetics bill.

DR. TUCKSON: I just want to make sure that you know. Thank you for that. You are preaching to the choir. We have been fighting this a long time. By the way, you can't make that phone call today. After today you can call.

MR. RACKOVER: But tell your friends in Oklahoma to make the call.

MS. ASPINALL: Thank you. Again, this has been an incredibly impressive morning. I was struck by a couple of things. First, how much is being done across so many different organizations. It is great to see that.

That being said, [there are] two missing elements. One is I think there is one constituency not here, which is industry, and how much industry is doing in individual organizations and as a group to come together. I'm not just talking about funding, which has been a piece that several of you mentioned. By no means is a lot of this industry-funded, but I think that that is a component that is important to recognize.

The second thing I was struck by, Reed, is what you said. This may come up this afternoon. What I see is a window of opportunity with the coming of the electronic health records. Somebody mentioned that 40 percent of physicians have them and 12 percent more are on their way. I believe that there is a window of opportunity because I have heard too many times about great new practice guidelines, new tests, and new information that can't fit into a system.

If anyone has changed a system, you only want to do it once, or at least once in a lifetime. You don't want to have to them revisit it. So I think there is a window of opportunity now.

That being said in terms of context, my question is, do you work together? Is this the first time that all of you have come together or do you really share your best practices across the organizations and put together metrics so you and each other can see how you are doing against your own goals and against a broad, potentially set by this Committee, industry standard?

MR. McINERNEY: I don't think that we have worked together to the extent that you would hope we have in terms of being aware of one another's metrics and helping one another to assess those.

But I can say at least that we have worked with virtually every organization represented at this table, and that is not just because we have reached out to them, it is because they also have reached out to us and to the other people at the table.

So we do work together. It is a small community in many ways. Those of us who are interested in genetics education are sometimes, perhaps, a little too inbred, and geneticists ought to be aware of the danger of that kind of behavior.

But we welcome input from other groups as well. One of the things we do at NCHPEG, and I'm very happy to hear the affirmation for these kinds of approaches, is we reach out to the extent possible to a lot of groups that are not formally in the genetics community and we ask them what do you need. How is genetics manifesting itself in your practice. This goes back to Dr. Kahn's statement about looking at medicine through a genetic lens, or health care through a genetic lens, but then also looking at genetics through a nursing lens or a dietician's lens or a PA's lens.

So we do talk to one another. We try very hard to make the education relevant for the practitioners. Somebody talked about champions before, and that is very important in the Diffusion of Innovations mechanism. We are aware that there are few champions within some of these professions. Some of these professions are very large, like PAs. There is a handful of champions, and we have to clone them, or at least give them the resources to extend their impact somehow.

MS. ASPINALL: I think that is helpful. You don't have to recreate the wheel by sharing the best practices across the organization and, again, with industry. I think there may be a role for a convening organization to help you do that in an efficient way.

MR. McINERNEY: Thank you. That is why NCHPEG came into existence, to try to decrease the extent to which people recreate the wheels. I will tell you, however, that there is a fair amount of parochialism across all disciplines. Not just in health care, in all disciplines. There is some sense that if we didn't develop it ourselves it is not necessarily going to be right for us.

But we just don't have the resources to, I will use the word "squander," on that kind of approach. There is a certain set of core principles, perhaps core competencies in genetics, that are broadly applicable, not withstanding that they have to be elaborated differently for each profession. That is one of the things we help with as well. So we are on that page. We don't want to recreate wheels.

DR. McGRATH: Julio, do you want to go first?

DR. LICINO: I just had a comment, which is that for this meeting we received all this preparation package. Then I, coincidentally, was all out in The New York Times this past Saturday about the genetics companies offering the 1 million genotypes directly to consumers.

I work in this field, and I was kind of shocked because I know that people offer genotype directly and you can test for this or for that. But to see the whole 1 million [available] to consumers. I'm doing this for research and the price that they charge me for research is exactly the same that they charge for consumers. I think I'm being overcharged because they are making a profit.

[Laughter.]

DR. LICINO: But anyway, I see as a mismatch. I'm in Miami now, but until last year I was at UCLA and I was part of the Medical Genetics Training Program and I had someone who actually trained with me in the program as part of his research training. The research component to his training was all done with me.

So I'm kind of familiar with that structure. I don't know how to say this, but from what I have seen, it has a background and it has evolved from traditional medical genetics. You have now this kind of collision of this explosion of information, which is really not about the risk for typical genetic diseases. It is like susceptibility alleles, some of which may contribute very little to the disease risk.

How equipped are the people who go through these different training programs to deal with this kind of information and be able to interface with the patients? Because the person gets this test and they are going to go to a professional. They say, "I tested for this. What does it mean? Can you digest this for me?"

Are the professionals equipped to do this? Should they be equipped to do this or not? What is the situation? How are you going to handle this kind of a direct-to-consumer, very aggressive effort with traditional medical genetics?

I could see you taking a lot of different positions and justifying them very strongly. You could say we have nothing to do with this. It is not ours. It is not genetics. Or you could embrace it, or you could be cautious, or anything in between. What is your perspective on this?

MR. McINERNEY: I will jump in quickly. I think it depends on how you define "professional." If you are talking about the genetics professionals, I think they are equipped to handle this. I think the average primary care provider is going to be absolutely clueless when confronted with some of these test results.

But the educator in me sees an opportunity here. In fact, we will be meeting with the 23 NV people soon to talk about, I hope, complementary activities to what Judith Benkendorf was discussing from the college.

But if I were trying to integrate genetics, for example, into medical education or into education of PAs or nurses or dieticians, for example, I would take one of those test results into my class and say, look, someday soon when you are in practice, one of your patients is going to walk in with this. What are you going to do. What do you need to know. How do you expect to respond. How are you going to handle this in your practice.

So I think there is a real opportunity for us here because maybe these companies are pushing us faster than we were willing to push ourselves.

DR. CASHION: If I could respond to that also, I actually have an NIH-funded study that is looking at gene-environment influences of weight gain in renal transplant recipients. We are doing adipose gene chips and we are looking at blood as well as dietary nutrition and exercise.

So I have that part of me, that mind-set, that is working on this. I also teach undergraduate nursing students genetics. People say, well, what do you teach. What is the content in that. Over the last six years, I have found that the most influential content you can teach them is how to be the lifelong learner. We really focus on websites. We go to websites every day and look at them.

We do the Family Health Initiative. They have to do it on their own families. We didn't have the gene chip information that is now out there by the three companies, and I was actually thinking I need to pay that $1,000 and let me go ahead and get this done on myself. But those are the examples that are meaningful to the undergraduate students.

I also teach advanced practice nurses, and they are the ones who are coming in and wanting the BRCA1s and BRCA2s. So again, it is how to look for the knowledge. Whatever I teach them today is not useful two or three months from now. So I really do not teach content as much as I teach how to learn, how to maintain your skills.

MS. PESTKA: I would like to add to that as well. I think much of the answer to this question is not so much in our hands as it is in our patients' hands. As you pointed out, patients are learning. The video snip that I showed of James, James was pretty exuberant in his hopes for genomics. But almost all of our patients come in and they have hopes and they have expectations. We really need to be prepared to deal with those expectations.

I believe there certainly is a place for our experts, our medical geneticists, our genetics counselors, but obviously there are not enough. So we need to have all healthcare providers educated, and then we need to have our body of experts that we can refer to with the really complex cases.

DR. TUCKSON: We have to close out. Was there one last one, Barbara, you had? Toby, go ahead.

MR. CITRIN: I just wanted to at least tie a few of the comments and a couple of the questions together from the public health perspective. It seems to me that Dr. Kahn's comment on the need for integration is very true of public health. It actually addresses Dr. Khoury's question about the size of the work force.

We don't need a larger faculty in our school of public health to incorporate genomics in what we teach. We just need people who are teaching the subjects they teach to incorporate genomics in those subjects. To some extent we have been successful in moving in that direction.

The same is true of public health departments, people who specialize in chronic disease. People who are doing studies of risk factors, who are administering behavioral risk factor surveys need to incorporate family health history in what they do. So it seems to me that it is not a work force question. It is very much a training and education question.

I think this also relates very much to Dr. Tuckson's question about is there a problem. I think the problem is that if we do not move the educational process forward this way, the public will be seeing genetics as segmented because the private sector is moving things in that direction.

The public's consciousness is that genes are more and more responsible for their ills. The net result of that will be genetic tools, a worsening of disparities as these tools are available to some and not to others, a sense of genetic determinism that will result in more disparities, more stigmatization, and more sense of a rebirth of eugenics, all those horror stories that we are all very familiar with.

The integration of genetics with all the other factors of health and disease; in the public health sector certainly that is our defense against doing less and having the forces that are moving very fast distort people's views of their health and what causes it.

DR. TUCKSON: Eloquently stated. First of all, we have benefitted from a terrific panel. Not only did we get smart people but people who know how to express smart ideas very cogently. We really appreciate it. Let's give them a round of applause, please. This is terrific, just terrific.

[Applause.]

DR. TUCKSON: Now, here is our dilemma. We have a very power-packed session this afternoon on oversight. A lot of this material that we just heard, as I tried to intimate, is part of some of that. But there is still not going to be, in my way of thinking, even under the most blessed of circumstances enough time to get everything squared away.

I would propose that I think there is something here that is going to require a deeper deliberation by us. I would be surprised, but I'm open for someone to object, that would say that in the broadest of unformed terms that there is work that this Committee will want to pursue in this area.

Unless I hear someone scream out that this is all solved and there are no issues here and everyone return to their homes, I think that we are going to wind up creating a subcommittee to take another look at this.

What I cannot do, nor do I think we are prepared for, is to define the agenda or the scope of that work yet. So as the afternoon unfolds, we will be thinking about that a little bit and have something to present to you that will arise out of the oversight conversation.

Barbara, I know that you may not be able to be with us, which is a perfect way to draft you into continuing on, and we will populate the committee with a few interested people. I know that I'm going to draw Mara into this and a few others. Not Joe because he is disruptive sometimes.

[Laughter.]

DR. TUCKSON: But we will do that.

I want to telegraph quickly, before we go to lunch, one thing that is real clear for the new members. I think it was the right question and a great answer came from the committee: "Do you all talk to each other?"

One thing that I think is frustrating for all of us is when we try to ask who is qualified to do what, particularly when it comes to counseling and who then should be qualified to do counseling and what is the reimbursement that ought to go.

This issue comes up over and over and over again. We have asked over and over and over again for all of the factions to sit down together and figure it out, create an umbrella, lay out the issues, and then bring it back.

I think that that now, as I get more mature in this, is an unreasonable expectation. Therefore, I believe that it is a role that we might legitimately play to try to be a convener of the conversation. I don't need to get one more letter from one more organization that just rehearses exactly what they wrote five years ago. Clearly, we are not getting the message. We need to step up to the plate and convene.

That would be at least one thing that we would do. Secondly, I think it is very important that we understand and bring together very specifically those professional disciplines to tell us how they

are using their normal regulatory responsibility for who is qualified to do what and to determine the adequacy of those things. We shouldn't be trying to recreate the infrastructure of American medicine.

So I think that those are at least two low-hanging fruit. I think this third low-hanging fruit is real clear, and that is this decision support. We have been told pretty clearly that that has to be looked at. So the AHIC versus other mechanisms in terms of getting this review of what is going on in the EHR is something that is in the Oversight Report, so we will probably hit it there. But I think that is a third area that we are going to want to lock in.

So I at least see three things as a broad, general set of issues. Who is qualified to do what, is a real big thing, and that has to do with clinical practice and it has to do with counseling. Related to who is qualified to do what is who should get paid to do what. That is a little beyond the narrow confines of education, but it is so related that you almost have to look at them together because that determines a whole lot. Once you start asking the question "Who can get paid?" that starts to answer a whole bunch of questions. Then this idea of electronic records. I see those as being some of the things that are there.

With that overly long summary of putting down just a temporary marker, we are going to send you off to eat. Now, the dilemma is that we are at 20 minutes of, almost, and you are supposed to be back here at 1:10. So the dilemma is really yours, not mine. We will see you back at 1:10 because, whether you are here or not, we are starting at 1:10. Have a nice afternoon.

[Lunch recess taken at 12:38 p.m.]