

**Remarks of the Acting SACGHS Chair (continued)**  
**Cynthia E. Berry, J.D.**

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MS. BERRY: At this time I'd like to return to the Chair's introductory remarks, those that we glossed over when we first began in the interest of everyone else's schedule, and talk a little bit about the work that the ex officio agencies are up to in order to enhance SACGHS' currency and ability to stay abreast of developments. In August, you might recall that Dr. Tuckson asked the ex officios to provide us with updates on the relevant activities in their agencies and departments, and these updates, as was mentioned earlier, can be found at Tab 3 of the briefing books. Our thanks go out to the ex officios for reporting to us about these developments. The information will be very, very useful, and it's relevant to our work.

I know that in requesting these updates, Reed was hopeful that they would also be a resource to each of you by increasing your awareness of relevant activities across the agencies, and perhaps revealing opportunities for more interagency collaboration.

Now I'd like to take a few moments to highlight several of the agency initiatives. You may recall that at our meeting in October of last year, we learned about the Surgeon General's Family Health Initiative. The Family Health Initiative is a transdepartmental program aimed at increasing public awareness of the importance of family history and health, and providing the public with tools to be able to gather, understand, evaluate and use family history to improve individual health.

The Family Health Initiative is gearing up for its second big national event this coming Thanksgiving Day. We wish the Surgeon General and all the agencies involved in supporting this important health promotion message great success again this year, and we look forward to hearing how it all goes.

A few weeks ago, AHRQ sponsored an important meeting on gene-based discoveries. The agency's goal was to identify knowledge gaps and barriers to the clinical use of gene-based discoveries and develop strategies for overcoming the barriers and improving coordination of relevant federal activities.

Dr. Chesley, could you tell us a little bit more about the meeting and outcomes?

DR. CHESLEY: Sure, I'd be happy to. On behalf of Dr. Clancy, Dr. Goopernick convened this conference, whose objectives you mentioned. The title, though, is one important thing I do want to mention. It was titled "Genomics and Medicine I," and it really was titled that way to reflect the reality that we saw that as a first dialogue in an ongoing conversation both with our partners within the Department as well as key experts outside of the Department. The conference included representatives from across the Department, FDA, CMS, NIH, HRSA, as well as others.

The first day of the conference focused on genomics, and the second day focused on pharmacogenetics. I think it's important to point out that we'll have a detailed summary by mid-November, we hope, and that, of course, we can make available to this group.

One of the things that I think was key during the discussion and during the meeting is sort of pointing out some gaps between what we know and how we can use that information. One of the things we were looking for at AHRQ, as well as with our collaborators across the Department, is how to build on some synergies that may exist in AHRQ programs, such as our HIT program. We, for example, talked today about the need for an electronic medical record in the context of the study we were talking about this morning. So whether or not there's a role to develop or

facilitate such an electronic medical record was one of the things that we chatted about in our conference.

But also the intersection between, or I should say with, some of the evidence-based programs that AHRQ sponsors, like the EPC program, and others.

One of the things that I think was a key point made during the discussion by participants was their interest in having methods workshops and conferences to discuss issues involved in linking the information and data sets, both from genetic lab tests as well as clinical databases, in order to do research in this area.

MS. BERRY: Thank you very much. Appreciate it.

At our February meeting, we heard from Dr. Steve Groft, director of the NIH Office of Rare Diseases, and Dr. Joe Boone from CDC, about plans for a national conference on access to quality testing for rare diseases. The conference was held last month.

Dr. Groft, perhaps if you could come forward and give us a brief report on the outcomes of the meeting. Thank you.

DR. GROFT: Good morning and thank you for the opportunity to come back and report to you on what I felt was a very enlightening meeting with quite a bit of participation. We had over 150 registrants for the meeting representing clinical geneticists, patient advocacy groups, patients themselves, the clinical laboratories, federal government employees and program officials, and the professional organizations.

I think we tried to focus on a number of different areas, including infrastructure, current models for test translation from the research laboratories to clinical applications. We also looked at quality assurance and quality control measures, including the international aspect of test flow and sample flow. A major focus was on the need for educational efforts to assure and promote quality in patient testing and in the test translation process. So it was a rather busy couple of days and couple of evenings as we started, and some of the outcomes -- you've received, I think, a copy of the set program, the Collaboration, Education, and Test Translation Program. I think that was provided to you. You got that okay?

That's something that is under development, and we hope to have it implemented and open for business by January of 2006. Dr. Giovanna Spinella, Andy Faucett, Dr. Bonnie Pagan, Dr. Susanne Hart from Human Genome were involved in developing this, and we'll be going through processes that are identified there in the description of the project, and we hope to start to stimulate the development of genetic tests for the rare diseases. I think four or five years ago the feeling was that nothing much could be done, there wasn't much interest in the rare genetics disorders. I think by the last two meetings that we had, the first one in Atlanta and then here in Washington, there is considerable interest. It's just a matter of bringing the people together, focusing on the issues and the concerns and the needs, and then having individuals who are committed to finding answers work together to get things moving, and I think we've been able to do that.

As all good groups, you always want room for another meeting, so we are planning another meeting in 2006. I don't think we can get away from that. But there are going to be presentations at the American Society of Human Genetics and the American College of Medical Genetics. We

SACGHS Meeting Transcript  
October 19-20, 2005

are distributing the results and the findings and looking for more input from different people as we go along.

Another recommendation related to education, we felt an awareness campaign about genetic testing and genetic counseling services was necessary here in the country. There just seems to be a tremendous absence of adequate information to the public, to clinicians, to the researchers about the requirements and the needs related to genetic testing. So I think we'll be focusing on that however we can with whatever partners we can gain as we move forward.

There's considerable effort already devoted to development of international quality assurance and quality control guidelines, and I think that will continue. The OECD group from Europe and others, Joe Boone is intimately involved in this, and we will continue working that area and just facilitate the development of the genetic test across borders.

Currently, the focus has been on molecular DNA-based tests, and we're hoping to expand or consider the development of new and expanding networks to focus on the biochemical and cytogenetic procedures for the development of genetic tests. I think it was two groups that sort of felt that maybe they were on the periphery, but after the last meeting a feeling of inclusion I think is there, and we're hoping that they either will form new networks or we'll just incorporate them into the existing network.

We base many of our proposed activities for the set program on activities that Dr. Bill Gall, the clinical director from the Human Genome Research Institute, has been involved with in developing genetic tests. We use that as a model or pilot to see if we really could utilize commercial laboratories, academic laboratories to develop genetic tests. During the past two years we've developed 21 or 22 different genetic tests. We're using this as a model.

So we'd like to extend this a little further to see if we can really expand this out into the community further and a couple of years from now see what the possibilities are for maybe a little bit larger initiative throughout the entire NIH structure.

So what's about it. Do you have any questions? There are some more qualified people in the audience who were there than I am that can answer questions. But if you have any questions, we'll try to answer them.

MS. BERRY: Thank you so much, Dr. Groft. Appreciate it.

Next I'll attempt to report on the activities of several SACGHS members and staff, folks who have been up to some very interesting things, and I'll start with Dr. Telfair who, as you know, is the SACGHS liaison to the HHS Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.

What is the acronym? How do you pronounce that?

DR. TELFAIR: I'm only a liaison.

(Laughter.)

MS. BERRY: All right. Well, the committee held its fifth meeting in July, and we'd be interested to hear a brief update as to what transpired at that time.

SACGHS Meeting Transcript  
October 19-20, 2005

DR. TELFAIR: Well, in the packet is a summary, a condensed version of a much larger report. So I will highlight a few things, just the bolded parts of this report. So there's much more discussion there.

This is a committee where the last meeting I went to was their fifth meeting, and actually this week is their sixth meeting. So they've been very active, and their primary focus is on newborns and childhood. The committee began its deliberations as a long-term follow-up for discussions with the public comment review from the American College of American Genetics report on newborn screening. The report itself was "Newborn Screening: Toward a Uniform Screening Panel and System." The notes from that and what proceeded on that is in the handout, but just three things that I want to highlight.

The focus was on the issue of improved access to services, especially to underserved and the most vulnerable populations. The other one was to ensure services of high quality, particularly those that have a high level of scientific merit. The other aspect of that was also to begin to look at issues related to culturally competent care. This includes things like health literacy and giving consideration to parents who have to make treatment decisions. The committee itself reviewed a very large number of public comments that came in. The public was given about a two-month time period to review the document, which they could get access to through websites and other means, and then to provide comments.

I forgot in my report to sort of research the actual number, but Dr. Mike Watson is here, and I'm sure he could tell you how many comments they got. So I would leave it up to that.

The committee itself actually had other business that it dealt with. It is because of its relationship with HRSA, and within HRSA it is the Maternal and Child Health Bureau. So Dr. Peter van Dyck, who is the associate HRSA administrator over that unit, has a high degree of responsibility and interaction with that group, and Dr. Michele Puryear, who is the director of the Genetic Services Branch, was within the DHHS, HRSA, Maternal Child Health Bureau, is in charge of that. So not being a fed, I have to get used to the acronyms.

But anyway, within that group, a major focus was on the issue of screening, and Dr. van Dyck's primary comment in his role was to discuss with the committee a means by which a letter will be drawn that will go to Secretary Leavitt, but at the same time how the Maternal and Child Health Bureau will be involved in communicating the information from that, and also looking at the recommendations from that particular study. I would encourage everyone to really review that report if they have not already done so, to look at that report.

Dr. Brad Therrell meets with this group, and Dr. Brad Therrell is the director of the National Newborn Screening and Genetic Resource Centers, and their main responsibility is basically to work with states to track the activities that the state health directors were involved with newborn screening there. Then the report, I refer everyone to the two handouts that he gave, which basically updates the status of the newborn screening at the states, both the number of conditions as well as the number of states that will do universal newborn screening in key areas. Also, he provides in great detail a detailed map to look at that as well.

There was an issue that came up in prior meetings related to the role of evidence and other factors that influence evidence in relationship to public policy decisionmaking, and several scientists were asked to come and give presentations on those, and those are listed in the report as well.

SACGHS Meeting Transcript  
October 19-20, 2005

Then there are several subcommittees that exist within the committee itself, and those committees are Education and Training Subcommittee that was led by Dr. Jennifer Howse, but now someone else will take over that role because her time on the committee ended; Follow-Up and Treatment Subcommittee; and a subcommittee that deals with laboratory standards and procedures.

Then there's a public comment period always, and I list a large number of the persons allowed to do public comment, but there was also a relationship with the American College of Obstetrics and Gynecology, which was given a little bit of time to discuss their perspectives on the ACMG report.

I tried to be brief.

MS. BERRY: Thank you very much, Joseph. Appreciate it.

Dr. Leonard is up next. She was recently appointed to serve as our liaison to the CDC EGAPP Working Group and just came from a meeting of that group and can provide us with a report on the meeting and the group's progress.

DR. LEONARD: Well, Muin, please feel free to jump in here because I feel like I'm usurping what has been done by you and Linda Bradley.

EGAPP is now a year old, so a steering committee selected a working group of 13 individuals. Al Berg is chair of that working group, and the working group has had three meetings to date. In addition, there are subcommittees of the EGAPP Working Group that are working on various subprojects of EGAPP. So overall, just to bring everybody up to speed who may not know what EGAPP is, it's Evaluation of Genetics in Principle and Practice. Is that right? What is it?

(Laughter.)

DR. KHOURY: Evaluation of Genomic Applications in Practice and Prevention, double P.

DR. LEONARD: Practice and Prevention, okay. But you must have somebody who stays up like all night designing logos for you, because I was very impressed by the logo with the big E, and then G-A-P, and then a big P, with a DNA going between evidence and practice. I mean, who came up with that?

DR. KHOURY: We love to do this in the government.

(Laughter.)

DR. LEONARD: Francis, maybe you need a percentage of your budget for logos.

(Laughter.)

DR. COLLINS: It always results in a war between the staff. So you also have to put in some money for employee counseling.

(Laughter.)

DR. LEONARD: Sorry. I shouldn't have gotten sidetracked there, but it was quite impressive.

So the working group has spent time developing methodologies because they are approaching evaluation of genetic applications in a different way than some of the more stringent groups, and it's really delightful to see them considering some of the more social and knowledge-based aspects rather than strictly defining utility or benefit based on medical treatment availability. So they have developed an entire process, a process for selecting the genetic applications that they want to evaluate. So there was a whole group on how you choose these and prioritize them.

Then there's a request for task order, RFTO -- it's like an RFP -- that goes out to evidence-based review centers requesting them to do an evidence-based review on the particular topic that's been selected by the working group. Then there's a whole description of what is needed, and the evidence-based review center will provide that evidence-based review back to the committee in a specified amount of time. That working group will then take that evidence-based review, which will have its own conclusions, but the working group will then make recommendations based on that evidence-based review.

The recommendations. They are now developing how they are going to make these recommendations, and they realize that the recommendations have implications for physicians, as well as for individuals and how they tailor the needs of those different groups. So they're being very, very thoughtful about this entire process.

So they're looking at the benefits in terms of medical benefits, diagnosis/prognosis/treatment options, patient benefits, both medical and personal, family benefits, societal benefits, and public health benefits. So they are being very broad in the range of benefits that they're looking at.

So there are two evidence-based reviews that are far enough along that I think I can mention the specific topics. The first has gone out for request for task order, and I believe at this point the specific evidence-based review center has been selected, and that one is looking at cytochrome P450 testing for patients with depression who are being treated on SSRIs, either prior to or on treatment for SSRIs. That one will have a nine-month review process for the evidence-based review to be completed, and that will then come back to the working group.

The second topic is HNPCC testing algorithm from screening by Bethesda Criterion Family History through screening testing by MSI and immunohistochemistry to full gene screening for those that are positive, this entire algorithm. So since this testing is more complex -- and this would be for patients with newly diagnosed colon cancer. Since this is a more complex testing algorithm, there's a 13-month time frame being given for this, and this is about to go out for a request for task order response from the evidence-based centers.

Finally, the working group is considering if they can do fast-track options. The two that I mentioned are full-blown evidence-based reviews, but they're considering the possibility of fast-track topics when they want a narrower evidence-based review or if there's a much more limited amount of literature, and they're having discussions about how to do these. But those would be more on a time frame of three or four months.

So it's very exciting to be a liaison to this group. I think they're doing some really good things and definitely thinking outside the usual evidence-based review box.

MS. BERRY: Thank you, Debra.

DR. LEONARD: And since the meeting was Monday and Tuesday, you'll get my report later.

MS. BERRY: Okay. It's in the mail.

I also want to take note of an interesting policy research project being carried out in the U.K. on the evaluation of clinical genetic testing for complex conditions. The Wellcome Trust is funding the project, and scholars from Cambridge and Exeter Universities are leading the project. Last month the project team carried out focus groups with a number of U.S. experts to gather perspectives about how genetic tests can be evaluated before entering routine clinical practice, and how regulatory and health care systems can ensure the availability of valid clinical information for the interpretation of genetic test results.

Emily Winn-Deen participated in the consultation, as did a number of ex officio agencies.

Emily, could you give us a brief summary of how the focus groups went?

DR. WINN-DEEN: Yes. I'm not really in a position to summarize the focus groups because these were designed as a series of focus groups where each individual subgroup didn't really have access to what happened at the others. I think maybe Stuart would be a much better person to give a summary since he sort of ran all the focus groups.

I can say that in the focus group that I participated in, we raised a number of questions without coming to any clear answers, and part of what the group running this focus group was trying to do was to pull together the common threads and what things are common threads across both the U.S. and the U.K., other countries, what things are unique to a country like the U.S. that has diversified health care as opposed to nationalized health care.

So if you don't mind, I'd rather let Stuart give a little overview, if you don't mind.

MS. BERRY: Stuart, would you like to, or do you want to defer?

MR. HOGARTH: I must admit, I didn't come prepared to give a summary of our work in the focus groups.

DR. WINN-DEEN: Can you talk into a microphone and just give a little overview of what the point of it was and where you are in the process?

MR. HOGARTH: Yes. First of all, thank you very much to Sarah and Amanda for inviting me to the meeting. As I say, I hadn't come prepared to talk about the research, but it's a three-year project, and we are talking to all the stakeholders who have an interest in the evaluation of clinical genetic testing. So that's the government agencies, health technology assessment, regulators, clinicians, patient groups, and also industry.

We've run two focus groups in D.C. last month, and we had some really stimulating discussions with very diverse set of perspectives from those stakeholders. We're about to run out to U.K. Europe focus groups, which will be very interesting because we'll really start to see the differences in how the health care system structures and the different regulatory environments -- I mean, the way these issues are addressed is very different in Europe and the U.S.

I've just come back from Canada where I've been speaking to people there to try to get a take on that country's approach to these problems, as well.

SACGHS Meeting Transcript  
October 19-20, 2005

What I would say at this stage is that coming out of the two U.S. focus groups, there was a very strong discussion about infrastructure issues around the need for translational research and the lack of support for getting basic research findings through into clinical practice, and how there might be some kind of need for a change to the whole infrastructure where all the different points of control, the different gatekeepers involved, whether that's people in the reimbursement side, whether it's people in the regulatory side, whether indeed it's the professional bodies, which have a very important role to play in terms of clinical guidelines, can somehow actually be coordinated. That idea of actually coordinating the activities of different groups is, I think, a crucial one.

Aside from that, I think I'd probably stop rambling on, actually. Thanks.

DR. WINN-DEEN: Thank you, Stuart. Thank you for doing a much better job summarizing it than I could have from just one slice of the pie.

I think that the one thing that I just want to point out is that what Stuart made as one of his last points there, about the need for coordination, is something that our committee has also identified. So I think that's one thing that we should continue to have as an underlying theme for all of our deliberations on whatever topic, that we just need to continue to push for coordination, at least among the HHS agencies for whom we can advise formally.

MS. BERRY: And just to clarify, Emily, you participated as an individual, in your individual capacity.

DR. WINN-DEEN: I participated as a member of the diagnostics IVD assay community. So it wasn't as an officio or ex officio or representative of SACGHS.

MS. BERRY: One more item before the magic hour begins. In September, at the Western States Regional Genetic Summit, Suzanne Goodwin, our very own, gave a presentation on the SACGHS coverage and reimbursement report, and the summit was organized by Sylvia Au and colleagues at the Hawaii Department of Health. I wanted to make sure that we recognized that work and that summit.

There are other activities that we're going to talk about, and rather than going through all of them now, this relates to Reed's custom of putting up our priorities chart and seeing where we are, where we've been, we'll defer that until after lunch. So there is more work to talk about.

For committee members and ex officios, the lunches you ordered will be brought here. For members of the public, lunch is available in the hotel restaurant, as well as a number of nearby restaurants.

We will reconvene -- shall we say 1:05? -- 1:05, to give everyone a full hour.

(Whereupon, at 12:05 p.m., the meeting was recessed for lunch, to reconvene at 1:05 p.m.)