Future SACGHS Priorities: Issues and Planning Process
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DR. TEUTSCH: I think there is more coffee back there, so even if you are not energized you can reenergize. So, as folks need a break, I understand that you may want to take rolling breaks.

You heard yesterday from Reed about the major projects that have been completed and the many of them that are actually nearing completion. What I want to do now is to devote some time today to a brainstorming session on new priorities that might be appropriate for the Committee to take up.

The point is not to come to any final decisions today but simply to come up with a list of things that we need to consider, and then we will work through them probably at our next meeting in July. So the goal today is really just to identify some of the issues that we might want to take up.

The priorities that we have been working under and which Reed went over with us in some detail yesterday were established through a systematic process that the SACGHS undertook in 2003 and 2004. In Tab 5 you will find a summary of the process that was used at that time, which in general seems to have worked very well. I want to walk you through my best understanding of what that was, since I wasn't there. Fortunately, I think a few people are still around the table who actually were there and can talk to us, and we will get some of their input on how it worked.

So what was done is initially the Committee members and the ex officios came up with a list of about 19 topics that were suggested during a brainstorming session somewhat analogous to what we are going to be doing this afternoon. A taskforce was formed to narrow down that list and investigate the remaining issues in preparation for a meeting which they had in March 2004.

Now, between the meetings the taskforce, with input from the Committee members and ex officios, narrowed the list from 19 to 11, which is what you see there. In narrowing that list to 11, the Committee considered a variety of questions. I'm not going to read them all but the criteria, if you will, for winnowing down the list.

Four of them I want to highlight here, and the first one is, does the government have jurisdiction or authority over the issue. So, do we have an audience that we are supposed to be talking to. Second, does the issue that is under consideration raise concerns that only the government can address or would government involvement be duplicative of other efforts. Third, is another body addressing the issue or actually better equipped to address the issue. As we all know, we are only one of many bodies that advise the HHS. Finally, has a policy solution to the issue already been worked out. That is, do we have something to contribute.

Once the list was winnowed down from the 19 to the 11, issue briefs were prepared on each of the 11 issues that were selected. Hopefully, some of you had a chance to look in Tab 5 at those issue briefs.

At the next SACGHS meeting then, members and ex officios deliberated on each of the 11 issues and organized them into three categories. They were issues that required in-depth study, issues that required short-term action or monitoring, and overarching issues that would be considered within the context of all the other issues.

You will see on the far left, under the category of "Short-Term Action," the vision statement, or roadmap as it eventually came to be called, was written to describe the 11 priority issues, the

reasons they were selected, and the process for identifying them. It includes the issue briefs that were prepared as background for the priority-setting deliberations, and all of that is in Tab 5.

Genetic discrimination, genetics education and training, patents and access, and oversight were categorized as issues requiring monitoring. The coverage and reimbursement, large population studies, pharmacogenomics, and DTC market were categorized as those requiring in-depth study. Then the overarching issues are access, public awareness, and genetic exceptionalism.

The next slide shows the priority issues which were first established in 2004, and some of those issues have actually changed categories, the ones that are there in blue. For instance, oversight was initially categorized as requiring monitoring but was subsequently elevated to requiring in-depth studies.

So in brief, that is how the priority-setting process was done in 2003 and 2004. Before we jump into all of this I would like to ask for a few volunteers to actually serve on a committee that is going to help work through this process so that some of you can take very close notes as we go through the subsequent discussion.

The purpose of this group is to continue the brainstorming on the issues suggested today, to develop a plan for the priority-setting process, and to identify what we need to do in terms of materials and background for that July session. The point is, get folks identified so you can be particularly vigilant as the ideas flow this afternoon.

So, could I get some volunteers to serve on that taskforce? Paul, great. Paul 2. Paul the Lesser, Paul the Greater. Jim and Mara. Ex officios are fine. I got Muin and Gurvaneet and Mara. I think that is a pretty good group.

Anyone volunteer to be chair of this group? I will participate as well.

Now, I think it would be helpful to actually hear, how many of the folks who actually were on that planning process back in 2003 and 2004 are actually in the room? Some of the ex officios actually were there.

DR. FROSST: I was in the audience taking copious notes, but yes, we were involved.

DR. TEUTSCH: Perfect. Muin, were you here for that?

DR. KHOURY: I think I was involved in the group, but --

MS. ASPINALL: Muin has the tenure.

DR. KHOURY: I'm the longest-serving person at this table.

DR. TEUTSCH: Anyway, since no one is willing to volunteer prior experience, I would like to open the discussion to the rest of the Committee just in terms of the process because clearly we could follow roughly the same priority process that we did before.

Now Muin has a comment.

DR. KHOURY: The reason why some of these things are on the board is sort of a gestalt of what was happening back in 2004. For example, the large population study was a very timely issue

that Francis Collins brought to the table because of the desire for NIH to do a large cohort study, which now has been rephrased.

DR. FROSST: For the U.S. to do one.

DR. KHOURY: Yes, for the U.S. A big cohort study around gene and environment. Now, that led to an in-depth analysis and now there is an initiative called GEI, the Gene-Environment Initiative. So there has been some movement. That was brought to that table because of the fed bringing that to the table.

The direct-to-consumer marketing was brought to the table because --

DR. TEUTSCH: Muin, I think what we really want to know is just about process right now. We will walk through some of the actual topics and what is on the table as a holdover. I think the question is, is that a process that we are comfortable with in general or are there other suggestions about just how we are going to sort through this in general.

The process was the generation of the brainstorming list, forming the committee to winnow that down, developing issue briefs. Then there was actually a voting process in there. The staff developed these, and they are in your folder so you get a sense of what they look like. Then, bringing that back to the entire committee to review and make some decisions about.

Yes, Mara.

MS. ASPINALL: I have a question. It sounded like it was somewhat part of the '04 piece, but shouldn't we ask the Secretary and his staff what priorities they have?

DR. TEUTSCH: Right. I'm going to walk you through at least some of the ones we have gotten from the Secretary. I think Greg is here and he can help us with that.

MS. ASPINALL: What a perfect setup.

DR. TEUTSCH: I think that there are issues like the issue briefs, which are a lot of work. Is that something that is going to be helpful for our process going forward. That is the kind of thing that would be very helpful to get people's sense of. What were the important parts of the process.

MS. ASPINALL: Right now or for the next meeting?

DR. TEUTSCH: What I'm asking is, as part of a process for the priority-setting would you like to see those done again, realizing the work that is involved. Barbara?

DR. McGRATH: I know when I came on board you gave me the book of those, and that was critically important for me to read just to get a sense of the philosophy of the group, what was on that list and what wasn't on that list. So I think they are great, if we have new ones coming up, to have that similar sort of background.

MS. ASPINALL: I would agree. I think they are great. I actually think they are sufficiently good that I don't know if they could be useful in getting out more broadly to some of the public or stakeholders on some of these issues because I think that they are a great 80/20. They are not the comprehensive but they really, with a little bit of effort, outline the issues well enough that I would love to look for opportunities to use them more broadly.

DR. TEUTSCH: I think the Vision Report, which actually included those briefs, which was made available publicly, was done probably for a couple reasons. One is to let the public know what we thought we were trying to do, but also then to share that information broadly. So the extent we want to devote the energy to a similar exercise, that is one of the things that could be done.

Maybe it would be helpful, before we actually start brainstorming, to again look at the original priority issues, which you see up there. We have already completed the report on the vision statement, coverage and reimbursement, the large population studies. We are about to complete the report on oversight and pharmacogenomics. We have written letters to the Secretary on genetic discrimination and direct-to-consumer marketing, both of which we continue to monitor. We wrote a resolution on genetics education and training of health professionals, which we just talked about and just created a new taskforce to revisit the topic, and we are in the middle of our work on the gene patents.

Our newest priority, which we would call here "evaluation," which is what Reed explained yesterday, which really was looking at translational and economic issues, was added in March 2007 but was put on hold at that time because of the need to get on expeditiously with the Oversight Report.

We know that the IOM Roundtable on Translating Genomics-Based Research for Health is considering this issue as well.

We also have the three overarching issues of access, public awareness, and genetic exceptionalism.

So the genetics education and patents will remain on our list. I think one of the things for us to think about, then is are there any others that need to be revisited or require ongoing monitoring, and do any of the overarching issues actually stand on their own as stand-alone issues. Do we need issue briefs updating us on evaluation or on the work being done by other bodies.

So we will think about those as we go through. Let me turn now, partly to answer your question, Mara, about suggestions for new issues. We have received several suggestions already, and the Office of the Secretary provided the following topics for us to consider. Let me at least put them on the table.

One is on international genomics infrastructure for clinical research. The next is on primary care practice-based approaches for the integration of CME, curricular and medical boards. Third is on clinical research standards for biospecimen collection. The next one is on economic and diagnostic value of multiplexed genomic tests and how are costs integrated into the commercial development plans to determine what factors the developers actually use to assess value, and co-development of molecular targeted agents and diagnostic biomarkers.

I can read them very well, or at least I can read them. Greg, we just ran through the list here of the ones that were contributed by the Secretary. Did you want to add any additional comments on any of those? I think one of the things we want to do today is get some clarity on what all of these things mean.

DR. DOWNING: I think, as opposed to how the oversight issue came as a specific charge, these were ideas in the journey of looking across the whole spectrum of personalized health care related to the initiative as to where further policy direction and activity the Committee might have some value.

There are aspects of this that I think touch on some of the other ongoing activities here. So we don't want to necessarily influence and move off any of the priority areas you are developing, but I can talk quickly through some of these if you would like.

Some of them reflect on some of the large population-based needs and resources for developing further research infrastructure, capabilities, and knowledge on some of the aspects of genome-wide association studies, for example, the aspects of biospecimens, and the characterization of them and the classification of them, the starting materials. The clinical resources behind the sequencing projects themselves has been something that has been pointed out to us. The Secretary was recently abroad in a number of countries and has brought back to us some specific follow-ups to ongoing international efforts.

It has been a while since I have really gone back through the records of what this Committee has done on international genomics projects, but it seems to me with the scope and the breadth of where a lot of the genome-wide association projects are going that there are multiple opportunities for international partnership. There may be within the agencies themselves enough activity ongoing there that this isn't something that has to be dealt with here.

But in terms of data sharing and the aspects of sharing the specimens and the characterization of the specimens, those are some of the things that we have stumbled across as being some obstacles that perhaps this group may want to weigh in on.

The extension of that to the clinical applications of genetic testing in other countries and that infrastructure and capacity is something. We are often contacted about where can health officials from other countries go to to find out more about how these are being integrated into clinical practice.

So if I had to say of the new things that he has brought to us recently, it has been the international domain of our context of this, not from necessarily just the research side but also into the translational side of it.

Can you go back? It has been a couple weeks since I saw these. Thank you. I apologize. I haven't followed specifically the one activity that you and Gurvaneet had tag-teamed last fall on as a potential area that relates more to the evidence development processes. A critical need that we see is that there are pieces falling into place about the aspects of being able to foster projects that support the evidence development. Not necessarily analysis of the evidence but where does all the evidence come from to support the clinical applications of these. That is a broad-ranging and complex issue but it is a big hole, as I'm sure you know from your experiences.

Then I think the primary care aspects. Some things that we have discussed offline with some of the staff are that the specialty areas have been dealt with and the educational components to this, but in terms of where we hear, frankly, a large amount of interest is from the primary care communities. They recognize they don't need to know all of the specifics but like to be able to start to plan to integrate from a preventive health perspective or be able to maintain current knowledge of understanding how to integrate various types of genetic tests into clinical practice and primary care.

I think Marc has shared a great deal of interest and information with us as well, so I think this is somewhat consequent with your last discussion.

We were asked to provide comments about directions and new things that we had seen through the Secretary's Initiative that might bring some interest to this Committee, but I want to be very careful to characterize these as not being of the same nature of the genetic testing oversight issues that we have brought forward.

I would be happy to elaborate.

DR. TEUTSCH: Why don't you stay here. First of all, are there some questions or clarification?

DR. WILLIAMS: I just think that as we were thinking about pruning the tree in our last discussion, Bullet No. 2 here would seem to be responsive to that particular thing and should fall under the aegis of the Education Taskforce.

DR. TEUTSCH: I think we are going to need to do some consolidation. We will get back to the taskforce group. But, are there clarifications of at least the issues that the Secretary has at least raised? Yes.

DR. FERREIRA-GONZALEZ: Greg, for the clinical research standards for biospecimen collection, NCI already has an initiative. I know it is oncology-oriented, but they already have an initiative to work in this area developing these standards. Do you know anything about that?

DR. DOWNING: Yes, probably more than I should.

[Laughter.]

DR. DOWNING: Nevertheless, we have been off to a number of communities with a number of different approaches to integrating genomic information into primary care or into health care overall.

One aspect of this that keeps rising to us are, great, we are working on all these health IT standards and all these other aspects of it, and recognizing that they are professional organizations as well that are behind this, but in terms of characterizing where does the genetic information come from in the literature, if you go back and find these studies there is not any way you can identify whether the lineage of these specimens and so forth has been credentialed.

I'm making this up on the fly, but the aspects of being able to say does this information come from a bona fide source of annotation and so forth. I'm not a clinical pathologist, but is this really what it says it is.

I think some of this is addressing areas where recently a lot of attention to characterization of other cell lines and others has come into question in terms of some standards aspects to it, being able to go back in the literature and say, well, I have the same cell line, I can't find the same pattern of polymorphisms that somebody else has reported in Paper XYZ.

I think that this goes in the category of accountability and the ability to trace this back to where are the levels of competence you have that that really represents a patient with that condition and it is reproducible.

I'm not suggesting that this is a new area that requires a whole lot of attention and focus, but it is something that we have been seeing rising, particularly with a lot of the clinical genomic

characterization studies that are ongoing. A lot of standards efforts have gone into the technology and now into the health IT side, but the biospecimen components themselves still wax that.

Some of the attendant issues related to that are, if you are having a federally funded study for that support, some of that specimen goes back into a common repository where, if for some valid reason one needs to go back and reidentify that particular gene sequence or the associated genes with it, that there is a mechanism or a way to go back and validate or verify that.
To some extent, some of the stem cell work parlays over into this a little bit in the ability to say, do we have the identity of this particular specimen or cell accurately portrayed and it is reproducible.

DR. FERREIRA-GONZALEZ: This is a little bit different focus than the NCI activity because the NCI activity is more how do you qualify the quality of the specimen. Here it is the information. Those are two different issues.

DR. DOWNING: I agree, and they are probably both valid areas to explore. This is, I think, probably in a bit higher domain of being able to provide a credentialing process. If you are going into a large public database and you tell me it validates that that specimen is truly where it comes from and is associated with that particular condition.

So I don't think we are thinking of any huge, elaborate study, but I think there is a fair amount of effort underway and a lot of communities are saying, what are other groups doing around this area that help with standardizing or credentialing the tissues that are being utilized for these very comprehensive studies.

DR. TELFAIR: Thanks. I just have a question. It is two slides up. The other way. Forward. There you go. This is a question I have to get an understanding of the priority in thinking here. I thought that is what we were trying to do.

DR. TEUTSCH: There are a couple of these we haven't gotten to. Let me run through the rest of the things that are on the table, at least that have been put here, and then open it up to everybody else to put them on. Greg, if you can stay, we can benefit from clarification.

DR. DOWNING: I'm going to probably have to leave at a quarter after for a meeting.

DR. TEUTSCH: Then let me ask, are there other questions directed to Greg in terms of clarifying what the issues are?

DR. KHOURY: A question to Greg. The international arena is very important. There is so much stuff going on with genetic clinical research and population research and biobanks around the world. If this group spends a lot of energy dealing with this, would the recommendations that go back to HHS be useful? I guess they are because HHS has recommended for that to be at the table.

But the question to you, Greg, is in what way does HHS want to learn a lot of these informational efforts and what kind of advice can SACGHS give HHS along those lines?

DR. DOWNING: The aspects of what have been done in terms of broad international basic research projects overall I think have been well noted in the annals of science now in terms of how communities came together largely through consortium efforts and basic discovery research activities. I think where we have been seeing some of the gaps are, if there are difficulties in

acquiring specimens or tissues or whatever, that these resources are recognized as another opportunity to go to.

I don't want to say that we have completely identified a specific target in here, but in the focus of translating this into clinical and medical practice the meaning of this information, looking at different populations in different countries, there is, I think, a lot of interest that comes to the Secretary. His overall interest in this for many years now has given him a little bit of a compass in terms of how you put together communities to do this in more and more overarching ways.

So I don't think that there is a particular challenge or a problem here. It is the aspect of being able to share information, recognizing what the challenges are in other communities that may not have the same breadth and depth of the science infrastructure but are still very interested in the whole aspect of taking what is basic science now into solving major clinical challenges that may not necessarily be ones that we deal with on a high priority basis here but may have some relevance in other countries.

It may be, from the standpoint of looking at what are priorities in clinical research areas of applying genomics in other communities and countries, that that may be the opportunity for further collaboration in research. Maybe the challenge is introducing these in regulatory frameworks in other countries and how they accommodate this into their healthcare delivery system and approaches like that.

I know you have taken great care in looking at the regulatory side of this, but in terms of the health care practice side of the applications of genomics, genomic tests and so forth, there are still a lot of questions from many other communities about this.

We have had contingents recently from Japan and China who have been very interested overall in the population-based health approaches to health planning around several of the genetic tests and capabilities around certain types of cancer and cardiovascular disease.

So the emphasis here is really looking at are there ways to share common approaches in clinical research of genomics in broader communities and different contexts than what we would do just here in the U.S.

So I don't think there is any particular agenda here, just recognizing that the way health care is practiced in many parts of the world is different than here. The capabilities of doing clinical research in many of these countries to solve some of their health problems is not as well developed as it may be in this country. There are probably some areas that we can benefit from in terms of looking more broadly.

I don't think we are trying to shape a new research agenda here. It is the aspects of understanding how different cultures are perceiving the integration of genomic technologies into health care.

DR. TEUTSCH: Marc.

DR. WILLIAMS: Adding onto that, you mentioned research specifically, but we have also identified in different venues international issues relating to availability of rare genetic tests and the CLIA certification implications of that transfer of biospecimens across international borders and issues like that. Is that something that could be considered under that rubric of international issues?

DR. DOWNING: Yes. This was really meant to be a very broad characterization of one area that we thought transcended one particular agency in the Department and was something that we hear a lot about. The Secretary travels a lot, and this is one of the things he has brought back to us to further investigate and develop some approaches.

It could be as simple as identifying issues and leave it at that, but at the same time there may be some ways to use his leadership in some ways. I know from his meetings at the G8 this summer and at the WHO level there have been detailed discussions around the aspects of where genomics is moving in terms of health care. So I think, to this extent, if there are notions or directions about that, that would be helpful to him in operating either in leadership capacity or partnerships or addressing some of the technical issues that are coming up relevant to advancing research areas. That would be helpful to us.

Robinsue can help me here, but I should mention Bill Steiger's office at Global Health Affairs has been very much supportive of our efforts here in the Personalized Healthcare Initiative overall and frequently asks us to meet with the science authorities from other parts of the world. We have not done a careful analysis overall of what people are doing, but we recognize that this is an area that this body may be able to help out in some ways.

DR. TEUTSCH: Mara.

MS. ASPINALL: Just quickly as I'm cognizant of the time, when you talk about codevelopment of molecular targeted agents and diagnostics with biomarkers, there have also been a number of studies there. Is that in search of, and I put the last two together, personalized health care and how the next generation of drugs can or should be linked to diagnostics?

DR. DOWNING: I just, in the last several months sort of stumbled upon this notion of nomenclature about codevelopment can mean a lot of different things at various stages of drug discovery and development through various clinical phases of development and into basically companion diagnostics and therapeutics. We have not really been able to find a very good characterization of what that means at each of those stages and phases.

I'm not suggesting this gets into the aspects of developing guidance, but FDA has indicated that they are interested in developing guidance in this area.

But the attendant issues of how a particular test is used and the processes of looking at the test and the drug during its life cycle through development and clinical trials areas, what does that look like and what are the characteristics of that. What are the requirements of regulatory submission. If you intend to use that test along with the drug in the clinical practice overall, how do you design studies and so forth to do that.

This was an area that we put in here because we understand there is interest within FDA overall. Part of our report last year dealt with the areas. There is substantial industry interest in this as well. We haven't had a detailed discussion with FDA leadership about this yet, but we haven't found a lot of background information about what that process looks like and how companies think about it. There has been relatively little discussion to which we can really point to say here is what the future might look like in that kind of context.

MS. ASPINALL: I'm wondering in the context of almost combining it with the one ahead of it because many of these tests today may be multiplex tests. Part of the issue is looking at the relative value of the test versus the therapeutic and how they combine. It is a new field.

DR. DOWNING: Right. I think we were looking at the steps of this [being] the extension of a biomarker into a laboratory test that ultimately might be used in clinical practice. So that is one of the aspects of why we wanted to really focus on how do you design your R & D components to this, if you will, if your intention is to ultimately apply that marker as a test in clinical practice.

I'm sure they can be combined, but I was trying to distinguish the two approaches.

DR. TEUTSCH: Greg, we know you have other commitments, so thank you. I don't know if you were here this morning to know that we got approval of the recommendations for the oversight test, so they will be headed your way.

DR. DOWNING: Yes, the lights were flickering down on the fourth floor.

[Laughter.]

DR. DOWNING: I was waiting for the lightning bolt to come. All I can say is we have had some internal discussions about how we would embrace what you have to share with us. We realize what we have asked of you over the last almost year now and have been very impressed with the degree of engagement and the thoughtfulness of the Committee members' comments. We look forward to reviewing those.

DR. TEUTSCH: Thank you. We know they are going to fall on receptive ears and an enormous amount of effort has been devoted. So we will be interested in following up.

DR. DOWNING: Thank you.

DR. TEUTSCH: Thanks so much for your time.

DR. DOWNING: You're welcome.

DR. TEUTSCH: Before we open the floor, just two items, just to be aware there are other things that have been put on the table as issues. At the end of Tab 5 is an article from Nature Reviews Genetics which actually specifically requests that SACGHS provide guidance and set standards for determining what data from whole genome sequencing should be included in electronic health records.

Second, just to remind you that we had anticipated having a meeting in July to deal with the whole issue of personalized genome services, which has gotten a lot of attention. We heard a little bit about it yesterday from the public comments. As you know, several companies -- 23 and Me, Navigenics, Gnome, and others -- have launched direct-to-consumer genomic screening and analyses, which raises a whole variety of policy issues that we may want to address that are within our charter.

Since they touch on so many critical issues, the staff has actually proposed having a half-day session in July that will help us learn more about these companies and their services. So I would like to think that that will be on our list of things that we may want to address.

I wonder if I could ask Sylvia, would you mind working with staff to help organize that session? That would be very helpful.

MS. AU: Volunteers? No.

[Laughter.]

DR. WILLIAMS: I apologize because I'm going to have to disappear for flight reasons. But if the floor is open for a topic?

DR. TEUTSCH: Right. I know several of you have to leave, so I would love to get the ideas. I know Paul has an early flight. Why don't you get yours on the table and we will try and get them down as best we can.

DR. WILLIAMS: Very quickly, I think that we need to explore the reality of the $1,000 genome and the impact that that will have on what we consider to be a genetic test. It is painful to think about just after going through all the oversight, but the reality is that if the price point comes down for a whole genome then genetic testing becomes an informatics query. It does not become a lab test.

As much as we have already stretched the 1972 regulations on devices, this may well prove to be the camel that broke its back. So I think that would be an interesting thing to do at least a future scan and topic on.

DR. TEUTSCH: Great. Paul, I know you have to [go.]

DR. BILLINGS: I have a couple. One follows directly on from I think what Marc is getting at, which is we have seen already in the last couple of days talking about what is health-related genetics and genetic testing and what is cosmetic, recreational, ancestral, whatever you want to call it. I would think that a paper or an activity from this group at least fleshing out how technology is driving the changes in that and just the whole idea would be a really interesting thing.

A couple of other things occurred to me. One is, it is striking that we see no discussion anymore about gene therapy. When I was first coming up to Washington, there were any number of groups that were meeting on that issue. So it does seem to me that some update, reevaluation, whatever, in that area might be of some interest.

The other issue that was on my list was, again, the translation from our work on pharmacogenetics and genomics to the whole concept of personalized medicine and what is the yin and yang or the pro and con or whatever of personalized medicine.

DR. TEUTSCH: I think these are great issues to put down. I think there are lots of them that actually go on, whether it's genetically modified organisms, whether it is about stem cell research. All of these things could be within our purview. There are other committees that have obviously worked on them. But I think we should get them all on the table now so that we can begin to sort through them and figure out whether we are the right place to house them. So, think broadly.

Let's throw it open widely. Julio, Jim, Mara.

DR. LICINO: This was said before, but I think maybe we [can] make it very specific. I'm just [suggesting], either at the discussion that we are going to have in July or as part of these recommendations, essentially the interface between the informational tool that people are going to be getting more and more, which you described, and your medical records. When should those

things go to your medical records, who decides if they go to your medical records, can you place them in your medical records yourself.

The way I see it, I think that some of that information may end up in people's medical records not always completely voluntarily. If you are a prisoner, if you are in jail, if you are in the military, your DNA is collected. If you are at school, if you are a missing person. No, seriously, because that is how they find missing people now. You start to query and they can be described.

So this interface between law enforcement, informational things, and your medical records. How is that looked at.

DR. TEUTSCH: The privacy and controls.

DR. LICINO: Where does that happen.

DR. FROHBOESE: Directly following on what you just said, the idea of privacy, privacy of genetic information in an age when more and more of it is out there for more and more uses. I'm not sure if privacy as a big concept fits with what you were just saying or with other issues that we have come around, but it is an issue that has been around for a long time and is not going to go away, and I think is only going to get more and more intense as more of our genetic information is out there in more sources.

DR. TEUTSCH: Jim.

DR. EVANS: I have been struck in the last three years on the Committee in how often the issues that we address are directly affected by the structure of healthcare delivery and specifically the fragmented structure of healthcare delivery in the country.

The most explicit example of that was in the Large Population Study, in which we stated that one reason that dissuaded us from more firmly embracing that idea was our system of healthcare delivery, to use the word "system" generously, and moreover to achieve the benefits of something like a large population study we needed a less fragmented system.

I think that there are important aspects of genetic medicine that inform us about the type of healthcare delivery and healthcare reform that this nation should be pursuing. I think that is an issue that is very much on the radar screen of the public right now, healthcare reform.

I laid out in an article in JAMA about a month and a half ago at least some of the details about how genetic medicine influences what we should be seeking as we engage in healthcare reform. Since that is likely to be a national topic, and clearly the executive branch has a huge role in that, I think we should consider addressing what aspects of the emergence of genetic medicine can inform the Secretary in the directions we should go.

DR. TEUTSCH: Very good. Mara first and then Muin.

MS. ASPINALL: A couple things. First, let me add to the structure of healthcare delivery. I think we need to add to that the economic incentives that are related to that system and how that drives care or lack of care both in a public health perspective and an individual --

DR. EVANS: Relating back to genetics.

MS. ASPINALL: Exactly. Relating back to genetics and to personalized medicine, which is a lot of overlap but not entirely overlap.
On the one that was mentioned before that in terms of privacy and control of genetic information with the medical record, I guess I go a little bit more broadly. I guess I think that is the secondary bullet. The first bullet is what do we need in electronic medical records that is related to genetics. If somebody came down and said, as there was legislation only two years ago, this is the standardized federal health EMR that exists, what would our answer be? What would we want in that EMR in a rather specific way. At some point somebody is going to ask or somebody is going to create it.

Given that it has been talked about here in a number of different areas that the EMR itself very well may be the key organizing mechanism for all of us as a society and as individuals, I think we should give an opinion for what goes in there when you are putting that together. So, what is needed in the EMR in terms of genetic information.

No. 3 is related to Greg's point about drug and diagnostic interaction. Some studies would say 60 to 70 percent of drugs, particularly in oncology, in phase 2 or phase 3 trials today will require some sort of not inheritable but gene-based information in order to personalize the use of that drug. How do we believe that system should be set up in order to ensure that those tests and companion therapeutics are indeed used appropriately and as necessary.

DR. TEUTSCH: Great. Muin.

DR. KHOURY: I'm going to use sort of a genetic type called The Roadmap Less Traveled.

[Laughter.]

DR. KHOURY: We are into finding genes these days, and lots of energy and resources of the government, with GWAS and others, are geared to finding genes. Test developers want to put them into diagnostic tests and move them through the oversight as quickly as possible. We have discussed the whole morning how we do that or how it is currently done.

In spite of all of this, there is this big gaping hole after you develop these tests, which is sort of that first step on the roadmap. I call it T1. You develop that promising application and what to do with it afterwards. In other words, it is clinical utility and how do you go from there to the development of evidence-based guidelines so that third party payers can pay for it. It is a big gaping hole.

I think Greg mentioned a little bit about that, although it didn't make it on his list. He said, how do we stimulate that kind of research, not the data gathering we are doing as let's say part of EGAPP, where we are looking at the information and saying there is not enough information on clinical utility in the real world to make it worthwhile covering.

That is T2 and beyond and is such a big hole right now. We can oversight to death if we want to. We can develop registries. We can get the FDA to do their thing. But HHS, maybe in collaboration with the private sector, [needs to] begin to invest money in doing the kinds of trials and observational studies that would allow us to evaluate the utility and the validity of a promising application so that we can develop those evidence-based guidelines that we all need so that they can become integrated into health practice.

This is sort of the rubric under which this can go on, The Roadmap Less Traveled, or it could be The Road to Outcomes Research or some other conglomerate depending on what other ideas come in.

DR. RANDHAWA: I would suggest three topics. The first one is, it may be useful to have a white paper on research priorities for pharmacogenomics. The premise of this is there are hundreds of drugs available, hundreds of genes that may potentially predict hundreds of different outcomes. So I don't think it's feasible to do outcomes research on every possible combination of drug, gene, and outcome. Is there a way or are there criteria we can think of that would help prioritize what are the high value, high target pharmacogenomic applications.

For the other two topics I will take Steve's comment literally about thinking outside the box. One would be genetic modification labeling of food, and the second would be genetics and cloning.

DR. TEUTSCH: Genetics and clothing. Oh, cloning or clothing?

DR. RANDHAWA: Cloning.

DR. TEUTSCH: Here we have genetics and clothing.

DR. RANDHAWA: I meant cloning.

DR. TEUTSCH: Paul, you had some things I know you wanted to put on the table. Do you want to raise them now and then we will get to Scott?

LT COL McLEAN: I have seen a paper or two recently about genetic screening for populations beyond the newborn period, either mid childhood, adolescence, or adulthood. I think Beth Hutcher wrote a paper about that, which is an interesting topic. I mean, there is an entire advisory committee for just newborns. If you move that into other populations, it would be very interesting and raise a different set of problems.

DR. TEUTSCH: Paul.

DR. WISE: I have two. One that I would put as a subconcern under the issue that Jim raised about health care reform and that is the implication of new genetics for relationships over the life course. What we are seeing is new genetic insights creating new precursor conditions that never existed before.

Basically, any genetic predisposition for an adult-onset disease is automatically or perhaps a pediatric concern. In other words, The New York Times article about 23 and Me ended with the reporter saying "I'm going to get this for my six-year-old daughter."

There are a lot of issues that are raised. However, the structure of health care in the United States is in no way capable of facilitating appropriate response to those kinds of insights. In other words, Medicare doesn't talk to Medicaid for Kids. Insurance programs that have 30 percent turnover per year have no incentive to look long-term at preventive or precursor management. FDA will have new issues about extending the use of medications into precursor areas that really have not been examined. You begin to look at the whole structure of healthcare delivery and assessment in the United States over the life course and things begin to break down very quickly.

But I like the framing of this as a subset of the broader issue of the implications of the new genetics for health care reform.

The other issue that I would like to get on the table, and I know that it has come up here earlier, is just the implications of the new genetics for minority health. There are equity issues that are of special status that are captured by the excess uber-concern that the Committee has because of the special history genetics has played in these communities, the special requirements for the provision of genetic services in these communities, but also because public discourse around minority health is heavily influenced increasingly by genetic insights.

It is not sufficient to talk about this merely as a reimbursement issue or an access issue. It has special requirements because many would feel that the justice requirements would insist that any technical capacity to improve health, policy should respond in ways to preferentially address the needs of the neediest communities. It may require a special concern being voiced and examined by this Committee in this regard.

DR. TEUTSCH: Barbara.

DR. McGRATH: Exactly. A follow-up on that, or ditto that. Two little issues with that. One is that in most of our reports we make some statement that the new genetics decreases health disparities. We kind of use it as a phrase, and I'm not exactly sure how we should address that, but I think we should look at it a little more deeply and not use it as a throw-away.

The other phrase we use a lot is "genetic exceptionalism." We all agree there is something to it. You mentioned probabilistic risk. But by making it an overriding theme, we somehow aren't addressing it. It is very ephemeral. I'm not sure what a taskgroup for us would look at that, but I think we have things like stigma discrimination, probabilistic risk. We have enough features on it that it would be useful, I think, for us to figure out a way to address some of those.

My third little thing is a question. When I came on the Committee, the issue that was most in the press that I saw was stem cell research. I was kind of surprised that that wasn't talked about at all here. This is my question: is it not under HHS? It is less of a topic now, but it certainly is in the press a lot but not much in this room much. That is a question.

DR. LICINO: I think the stem cell discussion is very important. I'm going to skip that, but just going back to --

[Laughter.]

DR. LICINO: I think if we had something like on, let's say, genetics health diversity and health disparities, it would be very nice. I think it may impact on different groups very differently.

DR. TEUTSCH: I wasn't here to tell you exactly why stem cell was or was not on the agenda, but it seems to me that is something that we need to put down here for at least consideration going forward.

MS. CARR: Can you be more specific about that, though? What would you put on that? What is the issue?

DR. McGRATH: What is HHS's policy on promoting stem cell research. That is why I'm not sure if it is under HHS or not.

MS. CARR: The other thing that I would point out [to] this Committee is that the focus of SACGT's work was more in the healthcare arena and public health than in research. Large population studies was certainly a research topic and certainly research topics are part of your charter, but I think because of the many, many opportunities and needs related to the integration of genetics into health care, it seems to me that that is why that has been such a focus.

I will say on that I think there are other mechanisms to receive advice about that very important issue. Not to say you shouldn't [think about] this because that is what this is, throwing things at the list.

DR. TEUTSCH: Denise, did you have something earlier on consent or something related to privacy that you wanted to raise?

DR. GEOLOT: As we talk about privacy and population studies and the use of genetics, you have the whole issue of informed consent. Even though there is another advisory committee that focuses just on informed consent, it might be a good idea to explore meeting with them to see what their views are in terms of informed consent with children and with regard to genetics and broader issues.

DR. TEUTSCH: Sylvia.

MS. AU: I want to add to that minority health issue. For minorities it is not only health issues, there is identity. There is a lot of movement towards genetic testing for ethnic background. So there are a lot of other issues besides health-related issues for minorities and genetic testing. I would like to make sure that that is included under that topic, too.

DR. TEUTSCH: Mara.

MS. ASPINALL: I apologize. I have to head out. Maybe it is implied in a number of these, and it reminded about stem cells, but to deal with new and evolving technologies. We talk about a number of things in the context of today, but how to integrate new technologies, whether it is proteomics or the genome piece. There are some specific ones, but there is going to be a series of new technologies. Each one can be evaluated in and of itself, or is there an overarching mechanism that says how to recognize a new technology and how to integrate it into a system.

DR. TEUTSCH: Do you have a comment on this issue?

DR. FITZGERALD: Yes, just following up on what Mara said. Again, if you are going to go into that and even some of the other things that have been mentioned, this is the Committee for Genetics, Health, and Society, but as we all know, what is genetics, as we have just seen in our last thing on genetic tests, is being blurred. Even to go back and review the vision statement and the terrain of this Committee, you are going to say, what if it is an epigenetic issue? Does that fall under SACGHS or is that something else?

That is something I think the Committee is going to have to wrestle with at least a little bit because we are going to keep coming up against this genetic issue.

DR. TEUTSCH: What is it, right?

DR. FITZGERALD: What is it.

DR. TEUTSCH: Yes?

MR. DAYNARD: Just to build on something that Paul the Wise had mentioned, perhaps the idea that the aged are a separate community, a minority community, whose special needs as far as health care and the impact of genetic testing and genetic techniques on that population might be worth considering as a separate population sub-issue.

DR. TEUTSCH: Are there others? I see we are gradually losing our critical mass. I want to run one thing by you. One thing I think I would like to do, and I just want to get your concurrence, is to actually direct solicit the ex officios. Within their agencies I think there probably are some specific issues that they are facing that we should elicit. I just wanted to see if that was consistent with where you are thinking.

DR. EVANS: Should we ask for public comment about important issues that the Secretary's Committee should take up? It would seem logical.

DR. TEUTSCH: Do we have a process for such?

MS. CARR: We can, sure.

DR. TEUTSCH: Do we have a listserv, at least?

DR. EVANS: Some would say, whoa, we are going to start being logical now?

DR. TEUTSCH: A foolish consistency. Other thoughts? We have a really rich list of topics. Paul the Wise, the table remains open. We have talked about at least through the 22nd, and if we are going to have more of an ongoing process it could be longer than that. So if there are other topics that people want to get on the table, let's do it. It would be good to start with a rich count.

LT COL McLEAN: One of the minor themes that ran throughout was identifying subpopulations within our society which may not be getting as much of the benefit as possible. Maybe we should look at the population more systematically to find the other subpopulations that are not receiving the benefit of the genetic advances we have.

DR. KHOURY: One more.

DR. TEUTSCH: Muin, don't disappoint us.

DR. KHOURY: I'm not. Over the years I have seen an interesting dialogue and sometimes clash between the two worlds, the world of genetic and genomic medicine and the world of evidence-based medicine. I would like to see whether or not this Committee can weigh in on this dialogue.

DR. TEUTSCH: That really raises some different issues. There are some really interesting issues on evidence-based medicine about the individual versus population issues that come up in many guises. It would be an interesting thing for us to address.

Let me wrap up. I'm sure most of you won't mind leaving a few minutes early.