Discussion

DR. WISE: We have heard that it is all about money, value, knowledge, power, empowerment, responsibility, the future, but particularly to help guide our discussion, it is important to always remember that it is always about ego.

DR. WILLIAMS: On that note, it is time to go over these cluster issues in greater detail to, number one, make sure that we are reflecting in each cluster description the action steps that were outlined that were reflecting the insights and wishes of the Committee.

Also, we want to look at linkages. We very clearly picked up linkages that cut across. Now, we could turn all these clusters into one cluster, but that wouldn't be of particular utility for our work as we move into the future. But there may be ways to group these clusters in our thinking and particularly in the way we present them to the public and particularly to the new administration.

Steve suggested that we can walk through each cluster and have an opportunity to talk in greater detail. Maybe, Kathi, you can put up Cluster No. 1.

Also, in your handout in your book, at the end of Tab 5 is a summary grid of the major points made in each of the cluster presentations, particularly the outline of the action steps. So you could also use that to help ground your thinking and the conversation regarding each of the clusters.

DR. TEUTSCH: What we have to get by the end of the day is a pretty clear sense of how we want to organize this and what our priorities are. Although we didn't say it explicitly, I think we have a number of things that we can do, and we have captured them here.

There are things that we have already made recommendations about that we can just monitor. We have things that we can do over a fairly short term or in a brief report or some more in-depth reports. We need to figure out what that portfolio of work ought to look like.

So as we march through each of these, I think it would be helpful for us to begin to think about, within each cluster, which of these things we think are most important. Clearly, this body of things that we have laid out far exceeds our capacity.

So as we begin to sort through them and think about how they might be rearranged and so forth, we can begin to get a sense as to which of these things are of the greatest importance. It will help us to sort through and consolidate clusters if that is what we need, and begin to set some clearer priorities and organize our work going forward.

DR. WISE: It is fair to say, though, that we still remain flexible about these next steps given that the administration coming in may outline priorities that would require our being responsive.

DR. TEUTSCH: Absolutely.

DR. WISE: So while, as Steve suggested, it would be very helpful to get some clarity about where the Committee feels we should be going, we recognize that we will likely have to be flexible in responding to new concerns and priorities of the new administration.

So, discussion regarding Cluster No. 1. Please, Marc.

DR. WILLIAMS: The good news, if there is any, regarding Cluster No. 1 is that we are not indicating we need to do any reports, since we just did one. There is a lot of work that is ongoing that is going to continue to happen.

As I was reflecting, though, on this and the discussion from yesterday and today, the thing that probably isn't represented there that does have an impact that we should be intentional about is looking at coding systems.

The current CPT and ICD-9, which is going to be transitioning to ICD-10, how is that impacting. It certainly has an impact on reimbursement, as we talked about yesterday. It has an impact in terms of collecting data, which is going to influence Steve's cluster about utility.

So that may be a cross-cutting issue that should be added to our portfolio for study.

MS. ASPINALL: I would wholeheartedly agree.

DR. FERREIRA-GONZALEZ: I want to continue to echo that, but I think this could be a short-term action that we could engage in already.

Like I said earlier, we know the system is broken. It's not working for us. The professional organizations are already trying to look at these issues, and we need to engage them actively. Maybe we can have them come together in different groups so they can talk to each other or for us to continue to evaluate that. They have very in-depth expertise because that is what they do every day. That might be a way to go about this.

DR. WISE: I have a question. This issue has come up. It has been the subject of considerable deliberation by the Committee and discussion. Yet not a lot has actually happened in response. It is still an issue, still a problem.

What do you see as the primary obstacles to moving forward with this, and how would that elevate some of these steps to ensure that we are really finally going to be addressing the essential barriers to making headway on this?

DR. WILLIAMS: I'll take a crack at it. Some of the barriers relate to a lack of clarity relating to some of the interpretations of the statute and regs.

In the letters that we have been writing, we have asked specifically for some clarification. Much as we were talking about yesterday, understanding exactly what can and can't be done will inform strategies going forward about how to address some of these issues. Lacking that clarification does hold this up.

So the hope is that some of the questions that we articulated very clearly about wanting some direction and clarification will be forthcoming in the letter later this month. But if that doesn't happen, I would certainly see as the new administration settles in that we revisit those issues and say we really would like some clarity around these issues.

The second barrier is that different stakeholders hold different pieces of the puzzle. ICD is maintained in one area. CPT is under the control of the AMA. So there are lots of different players in there. As Andrea has pointed out, it is sometimes difficult to get the engagement or to get everybody at the table.

So maybe there is a role for the Secretary to convene, perhaps with SACGHS as the facilitator of that, everybody around the table and say how can we work these things out. But it is a very arcane system to have to navigate and to try and pull all the different pieces together. That does slow progress.

DR. WISE: Jim.

DR. EVANS: Not that I have a solution for it, but just to point out that you are also, in this context, fighting a long entrenched system of procedure-based reimbursement. Genetic services, at least so far, until we invent the genetoscope, is not a procedure-oriented field.

So it is a very broad issue. There are other constituencies in the same fix: general internists, pediatricians, and psychiatrists. So it is a rather pervasive issue and this somewhat irrational system is an entrenched system. That is an obstacle. I'm not sure exactly how you get around it, but maybe enlisting the kind of support of other large and important specialties for which this is an issue.

DR. WILLIAMS: Yes, that is very well taken. Of course, the problem that we have is not only on the clinical side, where there are the issues that you have articulated. Genetic counselors, being a relatively new profession, really don't have any official standing in any of the statutes in terms of being articulated, as opposed to nurses and physician's assistants. That also creates a barrier.

But even on the laboratory side, which is a procedurally-based specialty, it is clear that the reimbursement around those procedures is not adequate for the actual cost of the things that are being done. So that is an issue.

There was another point, but it has lost me.

DR. WISE: Steve.

DR. TEUTSCH: I was just going to ask Jeff a question. I know I'm putting you on the spot a bit, Jeff, but you have been wrestling with these issues. We have had some wonderful dialogues with you and colleagues at CMS, and you have been wrestling with these issues. Can you elucidate some of the things that we might do that would help move this forward?

DR. ROCHE: Hi. Good morning. For those of you who may not know me, I'm Jeff Roche. I'm an alternative for Dr. Straube from CMS.

CMS is indeed an interesting institutional road to navigate. I'm, at the moment, about six weeks past a year, so I'm a relative rookie.

One of the more interesting publications that I found that was useful is something that many of you may already know about which describes how CMS addresses payment and reimbursement issues related to new technologies, which is a larger basket with which genetic and genomic testing issues and genetic service issues in general are in competition. This may be very useful.

Again, it isn't a one-pager. If it were available to anyone to read, it might be very useful.

I welcome the opportunity to continue to work with Sarah and others from the NIH staff who have been trying to get us to consider these questions, which, I think as Marc correctly and

accurately pointed out, are based on an old system designed for a different purpose, which is confronting a rapidly changing field and which has both promise and, in some ways, areas where the promise has not been fully demonstrated.

I call to mind perhaps a recent editorial in the New England Journal of Medicine just last Thursday where the headline was "Payment Now, Possible Benefits Later." The discussion was actually fairly stringent about CMS's perhaps errors in covering certain imaging procedures and the major effect on Medicare costs that that error may have led to.

I think we want to be sensitive to the needs of any community, especially the changing community in health care, but I think we need to be aware that the consequences of our decisions at CMS, as for almost any other government agency of course, have enormous consequences.

So, as I say, we continue to look forward to engaging with both your staff and other Committee members and trying to resolve some of the specific issues, but I think this cluster does point out very nicely.

DR. TEUTSCH: It gets back to Jim's point. As we look towards health reform, it probably also means financing reform in how we pay for services, whether they are going to be episode or bundled or some other ways of reimbursement. Whether we handle this under the coverage and reimbursement set of issues or whether we handle it under health reform, it is clearly one of those things that is going to be tightly linked because it so strongly shapes how care gets delivered in this country.

DR. WISE: Did somebody have a comment?

DR. WILLIAMS: The other point I was going to make is about the money. The systems we have heard about are not a target-rich environment.

When we are talking about that we should get a bigger piece of the pie, that inevitably means that somebody else is going to get a smaller piece of the pie. That always is problematic when it comes to those types of discussions. That is just the environment that we have.

This ties in with Steve's cluster. We could make a much better case for ourselves if we moved away from being the faith-based specialty that we have been in genetics, which is you have to believe that what I'm doing is really good. In God we trust all of the data. Paul has heard me say that before.

But the bottom line is we do need to develop evidence around the value of what we do because then we are not just whining about the fact that we are not being paid.

DR. WISE: Andrea.

DR. FERREIRA-GONZALEZ: I think that the issue that Jim brings up is a good one. I think there is a lot of momentum right now not only in the laboratory community and the genetics community but across all medicine to start looking at some of these issues.

I think this issue crosses different clusters. Even if we look at Cluster No. 1 where we are going to monitor this, I would strongly recommend that this becomes another point in the clusters of the future. It has a huge impact on how we actually move. Even though it might be in two different areas, we need to connect them.

DR. WILLIAMS: That is a really good point. Mara can speak to this. She and I were the only two Committee members that were at the Summit on Personalized Health Care.

I haven't read the report thoroughly, but in talking to some of you who have actually been through the report already, it probably wasn't conveyed strongly enough in the report about the feeling of the attendees there that none of this is going to happen without substantial reform to the current system. That was really a key element of that summit. I think that that is a really right-on point. I agree that this needs to be carried forward with what Mara has been doing.

MS. ASPINALL: I was just going to add, people may be familiar with it. About six weeks ago, there was a summit in Utah around personalized health care. The report that we have was a key piece of that, really looking at many of the issues that overlap with Cluster No. 7, which says if personalized health care is going to happen naturally, how do we ensure we have enough resources, beds, systems, information, data, to have that happen.

But during this summit, which was relatively small, we were broken up into four areas. One of the key areas had two major recommendations, and I would say the number one recommendation coming out of the business area -- and again, "business" doesn't mean for-profit businesses but commerce in the broadest definition -- was reform of the reimbursement system.

So I think this one has to be a priority. There are a number of groups working on this, as Andrea mentioned, in the professional community, and as Marc knows, I was asked to head up a piece of that from the Utah summit on looking at a fundamentally new system and potentially replacing the CPT code system in order to do that.

I think the good news is, with health reform potentially coming, there was a real groundswell of support to say we need to look at this and how do we do it.

What I'm intrigued with in Cluster No. 7 is, if the reimbursement system changes there is no question utilization is going to change. If utilization is going to change, how can we ensure that we are ready for that and people are not therefore denied access for very different reasons because we don't have the resources there to serve. That is how I see the two working together.

DR. FERREIRA-GONZALEZ: Also, I think we need to work together with the clinical utility piece. As we continue to look at new ways that things are going to be reimbursed, the knowledge is generated to have a very good idea on the clinical utility of the tests over time. So, how do we make sure that as these technologies move forward into practice or not, that we gather more information and make decisions but, at the same time, make sure whatever we communicate or continue to work on within these three different clusters doesn't stifle the movement of personalized medicine.

MS. ASPINALL: That was the second recommendation coming out of Utah, ensuring that the public and professionals have confidence in the products coming out. The clinical utility piece may go a long way toward ensuring that confidence so that people aren't saying, well, does that work or not work. If we don't have confidence in the system, we can't change the reimbursement and then we can't assume adoption.

DR. FERREIRA-GONZALEZ: But to gather that clinical utility information sometimes takes time. We need to see how that process over a gray area of material is now black or white. That is something to keep in the back of our minds.

DR. WISE: Julio.

DR. LICINIO: One of the things, though, that I think is very important here that we are somewhat missing is that the big paradigm shift now is this direct-to-consumer thing. By the time a doctor or a healthcare provider or clinical geneticist or genetic counselor talks to somebody about some genetic finding that may be relevant to health, the person has already paid $400, which is relatively affordable, to 23andMe and has their 500,000 SNPs all there.

They actually have a service that they call, I think, a Genome Browser. If a paper comes out today in the New York Times, they can immediately go and check do I have that SNP or not, even before the scientific community is aware of the finding.

I don't think we are doing it in a deliberate way, but I think we are being very traditional and paternalistic. I think we are trying to find guidelines or policies to make recommendations to the field. But, is that what the public really wants.

I think that one of the recommendations should be a broad survey or community engagement process. I have done this before. It becomes very sticky: who speaks for whom? Who represents what? You cannot engage the whole country. So, which group do you engage; how representative are they. That is why before the word used was "consultation." That held with some people but not with others, so that has been changed to "engagement."

But I think that some element of community engagement should get an impression from the public. Or, recommend some kind of broad survey as to what the public really wants or needs. We may be preaching one thing, but if the public is willing to pay and get the information anyway in spite of the risks, they will do it.

DR. EVANS: Does that really belong in this cluster or does that fit better in Sylvia's cluster?

DR. LICINIO: Probably in Sylvia's.

DR. EVANS: I think your points are well taken, but I don't think they are necessarily part and parcel of reimbursement by third-party payers because of exactly the issue that many of these people will pay out of pocket for such information.

DR. WILLIAMS: Yes. I think the overarching theme that I'm hearing that really reflects all the comments is that clearly we as a relatively small group are not somehow going to reform the system. Yes, I know we all would like to.

I think that, as I see this, we don't necessarily have a good idea about where things are going to go, whether it is reform of the traditional healthcare system, whether it is a consumer-driven system that has radical alterations in reimbursement, or how evidence is going to play into this.

It seems to me that the role of Cluster No. 1 is to be able to assess where things are going, to hopefully be engaged as best as possible with the movers and shakers that seem to be making a difference in terms of the reform effort, and then being nimble to say what are the strategies that would work best given whatever system we end up with so that we can basically be ready to provide what is needed at the time that new things are rolling out.

It seems to me less a task of pulling everything together and creating something that is a static document in some ways and is more of an ongoing, nimble process to try and respond to a rapidly

changing environment, and just basically trying to develop as best we can the information connections that allow us to really have a good sense of what is happening.

DR. TEUTSCH: To build on what you just said, it seems to me that what I'm hearing here is that this particular cluster is dealing with a whole set of tactical issues that we are really facing right now. They deal with the system as it currently is and how to optimize those issues.

There is a whole set of linked strategic issues about what the future is going to look like which probably belongs more in the health reform future issues.

So it might be a way to keep our eyes on, yes, we have a whole bunch of these acute, short-term issues that we need to address, but we understand that probably over the longer term we need to deal with the broader issues. If we are going to move to a personalized healthcare system that adds real value, what do we need for that system.

DR. WISE: Other comments or suggestions before we move on?

[No response.]

DR. WISE: Why don't we talk formally about Cluster No. 2. Oh, I'm sorry.

DR. KIRCHNER: Peter Kirchner from DOE. I wanted to bring up the relationship between clusters again. That relates in particular to the reimbursement and how you are going to address the importance of reimbursement for any given area to the information coming out of Cluster No. 2 regarding the strength of the associations that have been created.

I'm just wondering whether there is some kind of a plan as to what will be a minimum amount or a necessary amount of evidence before you can address that.

Some time ago, the internal medicine organizations used to put out data of high correlation versus medium versus low, depending on the amount of evidence that was available for a given treatment or a given association. I don't know if anything like that might be useful in trying to address this difficult issue of when do you cross that threshold so that it is justifiable to spend public money on reimbursement.

DR. WILLIAMS: I think that is an excellent point. That is clearly something that is at the center of the healthcare reform debate in this country. The issue is that the reimbursement system, right now, essentially, is disconnected from the system of evidence and quality. It is basically based on work units and how much you do, and an evaluation system that, as Jim pointed out, tends to favor procedures over non-procedural activities.

I won't use any denigrating characterizations about thinking and not thinking, as some have chosen to do.

I think you are absolutely right. We are going to have to take the step in this country to move in that direction where reimbursement is really tied to best practice, to quality outcomes, and to evidence-based medicine.

How that is ultimately going to play out, and how that is going to look is not clear, but it is incumbent, I think, on the field, and in particular, under Steve's Cluster No. 2, that we are able to

deliver on the evidentiary standards that are going to emerge and then use those to tie into how the reimbursement reform is going to work.

DR. KIRCHNER: The other thing I wanted to mention is that, in addition to reimbursement, should there be consideration in your cluster regarding payment for additional research needed to bring the level of evidence up higher.

Now, CMS has actually been doing that for the last several years. I remember when it announced that it would begin to reimburse certain types of research as long as it met very rigid protocol standards.

That, I thought, was very important and might be applicable to the kinds of things that you want to do.

DR. WILLIAMS: I think that's a good point. The idea of coverage with evidence development has had some visibility. Jeffrey could certainly comment on this. I think that, as with anything, there have been some good things and some bad things that have come out of it.

The problem, of course, is that we sometimes decry the idea of how long it takes for physicians to adopt something. What we forget is that it takes them just as long to unadopt something.

I think one of the problems that has been looked at with coverage with evidence development is the idea that we allow a procedure and develop the evidence. Then we find out this really doesn't work, so we decide we are just not going to do it. But now everybody is in the practice of doing it and it is hard to extract that out. I think that has been reflected in some of the issues around imaging and, in particular, some of the more expensive treatments in the oncology arena.

Of course, the other thing that gets involved in that that we haven't even talked about, the real elephant in the room, is how a single liability case can dramatically impact the reimbursement landscape.

If we look at the bone marrow transplantation in breast cancer situation, one lawsuit against a payer for tens of millions of dollars essentially overnight changed the reimbursement policy in this country and caused payers to cover a procedure which, as evidence was developed through coverage, we realized really was not worthwhile. There are estimates that it cost this country in the range of $500 billion. I think that is the right number of zeroes associated with that. But it was a heck a lot of money, not to mention the morbidity and mortality associated with a procedure that was essentially in the vast majority of cases futile.

While I think that we are also going to see how this can play out, we should realize that that is also a double-edged sword. But it certainly is something that we would want to consider as part of our toolkit.

DR. WISE: This discussion of Cluster No. 1 has identified that there are clearly linkages across different clusters and that coverage and reimbursement issues will depend heavily on the success of Cluster No. 2 for sure and that will be an important contributor to Cluster No. 7, healthcare reform.

But as much as Cluster No. 1 puts a demand on Cluster No. 2, Cluster No. 2 also must respond very much to the immediacy of Cluster No. 1, reimbursement and coverage, and be intensely

practical and relevant in how it goes about its business in order to truly be engaged into the issues raised by Cluster Nos. 1 and 7.

Why don't we move to Cluster No. 2 and discuss it more formally, even though we just moved into it to an extent. Any specific comments regarding Cluster No. 2, which is ensuring the clinical utility of genetic information? Marc.

DR. WILLIAMS: I think the biggest thing that this group can do is to really highlight the funding disparity across the translational research arena.

As we heard from Muin when he presented at a previous meeting looking at the T1 through T4 translation, about 97 percent of the current research dollars are residing in the T1 basic science arena, which leaves very little for even establishing what might be considered rudimentary clinical validity, not to mention evidence around utility.

I think that one of the strategies that we should definitely look at and promote as members of SACGHS is some redistribution, if you will, of the research funding to move more monies into the T2 and T3 areas. That will allow development of evidence around clinical validity and clinical utility, which we all recognize is going to be exceedingly important if we are really going to move this down the road.

DR. GUTTMACHER: You are asking for redistribution, not an increase in spending, to allow that?

DR. WILLIAMS: That is what I said.

DR. GUTTMACHER: So you are taking that money away from other areas of research. I just want to make that clear.

DR. WILLIAMS: Yes.

DR. GUTTMACHER: I think some of us might have difficulty with that.

DR. WISE: Alan, tell us what the difficulties might be.

DR. GUTTMACHER: Well, exactly what research are we taking the money away from?

[Laughter.]

DR. WILLIAMS: You have heard me say this before, but I will put it into a public context. In some ways I think we have a hammer-and-nail problem. The Human Genome Project is, arguably, one of the greatest scientific achievements of the past millennium, if you will. I don't think that is overstating it.

One of the consequences of the funding for the Genome Project was the creation of a tremendous amount of sequencing capacity. In some ways, that has created a huge hammer so that every problem that then appears from a research perspective is to say, can we solve this with sequencing.

So we go from genome to HAPMAP, to genome-wide association studies, to sequencing organisms and all this stuff. There is a lot of that infrastructure. There tends to be a bit of tunnel

vision looking at, can we continue to solve the problems that we are dealing with by using this incredible technology.

My contention would be that while there is still value and knowledge to be gained from doing that, we can't continue to focus the vast majority of the research efforts into that. We have to take some of that and use different techniques to be able to say how we can do something with this information that is actually going to provide direct benefit to health care in this country. That requires a different model than we currently have been having.

That may be a simplistic view of it, but it does seem to me to be part of the problem.

DR. GUTTMACHER: I would suggest that might be a form of genetic exceptionalism. What you are really arguing, I think, is that the federal research dollars in general should be spent less on basic research and more on applied research, health services research, et cetera.

If that is what you are arguing, you might want to put it in that context rather than saying genomics per se. I think that is a larger discussion. This Committee, of course, does advise the Secretary of Health and Human Services, so that may be within the agenda of the Committee.

But I would think it is a zero-sum gain to some degree. Or one could ask for increased funding. Those are two different options. But I think one needs to be aware that of course good applied research needs to be based upon good basic research, which I know you wouldn't argue with.

I certainly would not argue against the idea that we ought to know more about what we are doing in health and health care, but we shouldn't just be in favor of apple pie without thinking about exactly what else we are not going to be able to cook and whether the apple pie is going to taste good.

DR. WILLIAMS: I certainly wouldn't disagree with expanding the discussion. I took the G in SACGHS seriously there. But you are absolutely right. I don't think that this is a problem that is unique to genetics and genomics. I think it is a problem that we are struggling with in terms of that balance of basic science research, which is absolutely critical, and then moving that into the translational realm.

I think there has been a relatively strong argument made that there is, and there has been, an imbalance between those two areas that probably does need to be addressed. Yes, it does mean that it is unlikely that there is going to be additional money injected into the system. It is not impossible, but unlikely. That means that there may be some redistribution that will be necessary.

But looking at what our task here is relating to the charge that we have been given by the Secretary, which is to address the needs of the American people relating to these new opportunities to improve health and reduce disparities, I think it is fair to say that we do need some additional evidence of utility to be able to move that agenda forward. That does require some commitment to research.

DR. GUTTMACHER: I would certainly agree that evidence of utility is necessary and research should be done in that area, but I would also argue, of course, that until we understand the basic science of genetic factors in health and disease there is much less point in applying that to health care.

We need to understand where it is along that pipeline of base translation. One could certainly discuss it, but we really need to understand those factors before we run out and study their use in health. We need to understand them and, as we understand them, incorporate them. That is an area of research that I think would be worthwhile.

DR. WISE: On that note, I have been impressed that the conversation between how much money should go for basic research versus applied research has been overwhelmed by the fact that applied research or comparative effectiveness or clinical utility research is going to be demanded by cost containment and by restructuring healthcare services. It won't have much to do with the conversation we just have had, and we need to be cautious that we don't allow this to become a tension that could be unhelpful.

But, to what extent should the clinical utility arguments be framed within healthcare reform and cost containment arguments? In other words, it is a shift in the frame of how we think about clinical utility and it also puts a different set of demands on the clinical utility community to provide products that directly and quickly can address issues of comparative effectiveness and cost reduction.

DR. TEUTSCH: On that note, why don't I suggest, since we have come to 10 o'clock, that we go ahead and take a break. At 10:15 we can come back and get public comments. Hopefully, we have folks here who do have some words for us. Then we will return to more on clinical utility, for which I have already seen some hands emerge. We will get back to those.

Go ahead and take a 15-minute break and meet back at 10:15.