

Cluster 2: Ensuring the Clinical Utility of Genetic Information
Steven Teutsch, M.D., M.P.H.

DR. WISE: Why don't we move on, then, to Cluster No. 2.

DR. TEUTSCH: Great. Thanks, Paul. I have to disagree a little bit with my colleague, Marc Williams. It is about the value, not just following the money. We are going from money to value.

That is ensuring the clinical utility of genetic information. This is a topic we have already begun to discuss.

Actually, I should say, Paul Billings, I think I heard you get on the phone; is that right?

[No response.]

DR. TEUTSCH: I think Paul has joined us from the West Coast.

DR. BILLINGS: I'm here, yes.

DR. TEUTSCH: Great. I'm sure you will chime in.

DR. BILLINGS: I doubt it, but go ahead. There seem to be enough chimes in the room, thanks very much.

[Laughter.]

DR. TEUTSCH: Clinical utility is an issue that we have also been tackling over the last few years in different ways. There are multiple challenges in establishing the clinical utility of tests.

One is the general paucity of clinical studies that actually do look at the clinical utility. Even when they exist, we don't have a clear set of accepted evidentiary standards against which to judge the studies, particularly for their different applications, everything from screening and prevention on to pharmacogenomics and things of that nature.

At the moment, there is no organization that is actually dedicated to performing utility assessments. There are a number of them that are involved to varying degrees.

There are also a set of concerns about how we are going to deal with the avalanche of information that is going to come out of whole-genome sequencing and how to assure that the information there is going to be applied in a way that actually leads to real benefit for patients. Some organization will be needed to perform some utility assessments on that so that we use the information well.

There are a number of groups that have begun this work. EGAPP is probably the one that is most clearly dedicated to it. That is the group that is out of CDC. It has been working to define standards and has actually been performing some assessments, but it is small and only beginning to tackle a modest number of these at the moment.

There are a variety of commercial and non-commercial entities that actually do technology assessments. You have two of them up here. Hayes is one that provides information largely to

the payer community. BlueCross BlueShield Tech actually makes their evaluations public. There are EPCs and other organizations that do some assessment of genetic tests as part of their larger efforts to assess technologies.

There is also the IOM Roundtable on Translating Genomic-Based Research for Health, which is very much concerned with these issues and then how we get effective technologies actually translated into the healthcare system.

The reports on Pharmacogenomics, Coverage and Reimbursement, and Oversight have all touched on this issue to varying degrees. We have recommended that the Secretary create a public-private partnership and a group that would define the types of underlying studies that are needed for assessments and the standards by which they should be judged, as well as to do those evaluations and help with the dissemination of clinical guidelines based on those assessments.

As many of you, I think, are aware, there are bills in Congress that do that, not specifically for genomics but do that generally within health care. There are other proposals, of course, that would like to do that exclusively in the public sector.

The kinds of issues that we think we could address are which groups would be most effective in defining the evidentiary standards and actually doing the reviews. A second would be how the government can better inform those involved in research and development about those evidentiary needs so that the appropriate studies will be done and we will have the information that will allow us to assess specific technologies and how they can be applied for specific conditions.

Although we have recommended in the past that this be a public-private organization that would be responsible for assessing the clinical utility of genetic tests, as I indicated, that is just one of several options that would be available for how this might get done. We could revisit the issue not so much of what it would do but more how it should be organized and structured, as well as a little bit about the scope.

All of this, of course, ties in more clearly with how to integrate what we know about genetic tests and effective technologies in the healthcare system, and will tie into the topic of the future of the healthcare system, which Mara will be talking about a little later.

These are some of the things that we could do. We could provide a forum for discussion to help define the evidentiary needs and standards for evaluating clinical utility. We could recommend that any governmental organization or group tasked with assessing clinical utility apply different clinical utility assessment methods for different clinical users of genetic tests so that we have that range.

We could develop some brief reports on how the clinical utility assessments, which are usually mostly very scientifically oriented, can also incorporate many of the important contextual issues: cost, cost effectiveness, ethics, legal issues, feasibility, acceptability, et cetera, to better inform the users, particularly patients, regulators, payers, healthcare providers, and performance measurement specialists, about how they can use that information in their decision-making.

This isn't quite worded right. We can look at how to better inform those involved in research and development about the evidentiary standards so that we can provide some better direction to the research community.

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The next one is about government organizations or groups that can establish evidentiary standards for clinical utility assessments, that can create methods for assessing them, and can actually do those utility assessments on a more systematic basis than what we have right now.

So that's it. Jim.

DR. EVANS: One question. Maybe this is better discussed later. But, since a common theme in this is the observation that we don't really have the kinds of studies to assess clinical utility and that that is needed, and getting back to the other theme of the morning that it is about money, is it worthwhile to look at mechanisms for encouraging or obtaining funding specifically for those purposes. There is a finite pie. Sometimes it is more appealing to fund certain things. These are sometimes seen as boring studies, but in the end they are obviously incredibly important.

DR. TEUTSCH: They are fascinating, Jim.

[Laughter.]

DR. TEUTSCH: No, I think it would be part of helping to provide that. I think some of the legislation that is out there today that is not specifically in the genomics area actually carves out a fairly broad list of things. They actually talk about sponsoring or conducting research, all the way from, for those kinds of things, doing clinical trials on through the economic evaluations.

I think we would want to talk about what the scope should be and how that should be used to inform the research committees. Pardon?

DR. EVANS: These are very expensive kinds of studies, but there is no shortcut for a lot of them.

DR. TEUTSCH: I don't want to get into a long discussion here, but there are different ways. This gets to evidentiary standards. When do you actually need an RCT, right? When is a simple decision model going to be adequate, right?

DR. EVANS: Sometimes it is adequate, yes.

DR. TEUTSCH: For the most part, if you ask people in this field about how to do these things, they are talking about clinical epidemiology mostly. We are not talking about, for instance, how do you understand the biological mechanisms of action and use that to inform the likelihood. How do you know when a biological mechanism is likely to be, in our understanding, informative or misleading. We have plenty of examples of both.

There is a whole range of things here that could be done under this general rubric.

DR. EVANS: Sounds great.

DR. TEUTSCH: Other comments?

[No response.]