Public Perspectives on the Oversight of Genetic Testing

DR. FERREIRA-GONZALEZ: I would like to invite all the public presenters to the front now so we can start moving a little bit faster.

To facilitate the process of hearing from as many stakeholders as possible, we have scheduled an extended public comment period today to focus specifically on oversight. We can move to the front. The groups that are going to be presenting today are Sharon Terry, David Mongillo, Patricia Goldberg, and Dr. Patrick Terry.

We welcome and appreciate the views of the public. We hope that the public comment process will help us collect information and ideas from a wider sphere of stakeholders and members of the public, and that the input that we will receive will help ensure the soundness of the report and the currency, utility, and feasibility of the recommendations.

We are very pleased today to welcome Sharon Terry, president and CEO of the Genetic Alliance, a coalition of over 600 specific advocacy organizations. The Genetic Alliance held a meeting in September on genetic testing, and Ms. Terry is here today to share the key points that emerged from that meeting.

Thank you, Sharon.

MS. TERRY: Thank you. I will try to truncate. Originally, I think Sarah said 30 minutes, and then a 15-minute discussion, but I'm going to try to cram that together. You have covered a lot of the issues that I would cover as well.

So we convened a meeting in September largely because of what we heard from many, many stakeholders across the board, that we really need a place to come together. We brought together a planning committee. I won't go through them all, but they represented the payer community, various industries from biotech and PhRMA, advocacy organizations, policy thinkshops, et cetera, and certainly the provider community as well.

Our starting principles were that we would put our eyes on the prize. The prize was health. So instead of the endpoint of better diagnostics or increased profits or better ability to treat patients, et cetera, we are going all the way to the ultimate outcome, which was health, and really kept that focus throughout the meeting and used it as the lens and the measure by which we made decisions.

"Truth-telling" was a phrase that in fact even scared some people when we put it in the title. I think there was some sense that we thought we were pointing fingers and saying that certain people weren't telling the truth, when in fact we were just inviting an opportunity for all of us to say exactly what we meant and thought based on the concept of health as the ultimate outcome.

That led us to looking at transparency around both IP and conflicts of interest. While many people from usually academia and not-for-profits stand up and say "I have no conflicts of interest because I don't work for a company," we really redefined that there and said that all of us have conflicts of interest and that it is important to talk about those from every aspect.

So people identified themselves by their conflicts, whether it was from a university, a not-for-profit or a for-profit, or the government. Putting those on the table to start with allowed us to move past them.

We also redefined IP there. Instead of saying this means simply that I'm a patent holder or that I have some kind of intellectual property that is by law mine, we talked about in a larger sense in terms of what I carry, what silos I like to protect, et cetera, and we tried to move beyond them as well.

We started with an intermediate starting point in terms of content quality. What I mean by that is we did not allow presentations that defined clinical utility and analytic validity and the baseline stuff. We really started at an intermediate point and moved quickly to a very advanced point.

We didn't allow any PowerPoint, which got people away from exactly what I'm doing right now, which is talking to you from PowerPoint, but allowed people to move beyond the canned presentation that they usually gave into a place that perhaps was new and really required that panelists speak to one another. So we used moderators and/or interviewers who would make people actually address one another and the questions before them.

The attendees of the meeting break down this way. I know all these slides are very hard to see from the back of the room. But basically, a large contingent of advocates, government, and biotechnology companies, small ones, were about half the attendees. The other attendees broke down with a large number of laboratories, a fair number of pharmaceutical, healthcare agencies, and academia. There were a couple policy people and a few media people. So we had a good diversity in the attendees, and we really left a lot of time. Over half the meeting was discussion rather than presentation.

I'm going to go through some recurring themes, and I'm going to go through them very fast at a really high level because this audience is very much advanced in terms of the issues. But these were the kinds of things that resonated with everyone as we went through the meeting.

The issue that personalized health care itself is creating tensions in the system, striving toward personalized therapies and interventions, issues around education, ones that you have heard over and over, that we need better vehicles across the board in terms of the public and clinicians, et cetera.

Resource allocation. How are we going to do everything from rare disease testing to international and developing world issues, looking at genetics and genomics as maybe the great divider or the great convener.

Public-private partnerships were mentioned over and over with very strong support, in order to alleviate some of the pressures on the current system. When we peel back many of the layers, we can go all the way back to the mess that the healthcare system is in and not just diagnostics, for example. So the idea that these solutions are not going to come just from government or just from industry but there has to be some collaborations.

Reimbursement. A lot of focus on reimbursement as the ultimate bar, certainly things that you have mentioned here as well. Do payers understand the value. Is the system right. Is the structure right. Can value-based pricing be sustained throughout the entire system, et cetera.

Biobanks. They are not regulated and they certainly often are silos that are contained by one entity or another and are not shared. They are not in the common, so to speak. How important are they; how should they be maintained; what should we look for going forward.

World health. How can the transfer of genomic technology to the developing world be implemented.

A great deal of discussion on evidence. How much clinical evidence is enough. What are the pressures in the marketplace to bring something to market before the scientific need is established or the validation process is done. Issues that you have also discussed. Can all the tests be held to a single standard in terms of evidence, and then, what can we do about post-market data collection.

IP models. There are certainly other industries, like the music industry, the publishing industry, et cetera, that have faced various challenges to their IP models. What have they done in a flat-earth, long-tailed kind of mentality, and what can we apply to the genomics industry.

Strong support for the passage of GINA across the board, that it was certainly an essential piece in getting service delivery. The CETT model, which again Stuart threw up here a bit, has been successful for rare diseases. Can it be expanded into the common disease and general kind of populations tests.

The role of patients in the advocacy community, essentially often considered the bridge between the scientific community and the public, but not always so careful about the messages they bring forward. The kind of hype that the advocacy community might do. Earmarks and IP in a not-traditional kind of context need to be really critically looked at.

Study design. We need predictable, well-designed studies, and we need to streamline the process through the pipeline.

Regulatory. A discussion of who has the regulatory authority for genetic testing and what should be the role of the various federal agencies. How would they be coordinated so that there was transparency and clarity.

Tensions between the product and the process. Issues around the technology taking great leaps, but behavior, whether it is the behavior of clinicians to implement these technologies or the behavior of patients to uptake whether or not they should eat brussels sprouts, is important.

Again, intellectual property. Where should the pre-competitive bar be. It has been moved back and back and back in some sectors, such as the information that is pouring into various databases on the federal level from the sequencing of the genome on down, but we haven't seen that mirrored in as many places as perhaps it should be.

An issue about registries. Should they be voluntary or mandatory. What kind of data and who should maintain them.

Medical record aggregation. Would the public support large databases or would there be privacy concerns that might override those benefits.

What should the role of professional organizations be. Should they step up to the plate and do more about bad actors, et cetera.

Costs and values. How to determine the difference between those two.

Risk-based regulation. We should perhaps be looking at tests that pose more risks to society and to patients.

Proficiency testing across the board. People believe the critical role of professional testing was essential to understanding quality control. And, how to increase the proficiency testing without placing undue burden on laboratories.

Direct-to-consumer tests need to distinguish between marketing and testing. How can the public be protected from these fraudulent and exaggerated claims.

Test interpretation. A discussion around clinical utility. Providers of course laying stake to the territory that allows them to do the test interpretation, and then some of the discussions with FDA and others around that.

So you can see there was a real richness and diversity of presenters. We went from the pipeline from research all the way through to delivery of services and looked at all the issues. We came up with a number of conclusions, and I didn't really expect us to because I thought our audience was too diverse. But I think it was helpful that we all did try to keep our eyes on the prize of health and did try to do truth-telling.

The report, by the way, will be about 60 pages long and will probably be out somewhere around January or February by the time we finish editing it, et cetera.

The conclusions were, first, that NIH put more requirements on funding and that there be various standards required along the basic and translational research pipeline so that evidence standards were achieved more effectively. Now, that is a chicken-and-egg because evidence standards are not quite clear, but we would recommend that we would start earlier in the process than later and that the research coming out of the RO1s, et cetera, be able to be evidence that would be useful.

The second one was discourse with and responsiveness from the federal agencies that have jurisdiction over genetic testing. The attendees felt that there wasn't always the kind of responsiveness that would lead to more rapid resolution of some of these issues.

Coordination of jurisdiction and activities of CMS and FDA and other relevant agencies. There didn't seem to be good coordination between those agencies and that is desirable.

Clarity and predictability. The current process is not conducive to a growing or stable marketplace. So, some of the same things that Stuart elucidated in a more deep way.

A risk-based regulatory system is desirable, with a caveat that allowances need to be made for volume. So again, some of the same things that Stuart went over in more detail.

Pretty much unanimous that direct-to-consumer tests need a special kind of oversight. Whether they were actually carved out or whether their risk was considered higher, there should be some way to address that.

Public-private partnerships as a desirable means for ensuring the pipeline of discovery through from research to tests as effective.

Education at all points.

The need for outcomes data collections and clear evidence bars. That was reiterated over and over and would certainly be something that I think focus should be put on.

That the industry itself should have the means to rid itself of bad actors but that regulation should not be based on bad actors, and the balance again between understanding what the industry in general needs in terms of risk to patients, to health, et cetera, versus the kind of various outliers that we have looked at.

A mandatory registry must be established and managed by either a public-private partnership or by a government agency.

This led to a number of action steps for the people assembled. No one entity took control of any one of these steps. Certainly, Genetic Alliance has stepped up on a number of them.

Advocating for enhanced CLIA was important. Promoting a mandatory registry. Convening a summit -- and there were actually several summit ideas -- on reimbursement issues. Another summit on evidence and outcomes data. There was actually another third-party review summit suggestion, and more recently, a suggestion on a summit on models, the models that are emerging lately around genetic testing regulation.

We need to explore the concepts of risk and how we are going to actually divide the lines between various levels of risk. We need to educate Congress, patients, clinicians, and there are probably many more groups we could put in there as well.

We need to work to pass GINA. We need to further examine global perspectives and bring that perspective in more, and we need to report back to the Secretary of HHS or his representatives. We did meet with Greg Downing on October 31st.

In addition, Genetic Alliance has gone on to work with a number of the various entities in the space, like ACLA, 21st Century Medicine Coalition, et cetera, to work together to understand what does this landscape look like and what would be the best solution for us moving forward.

That's the end of my report. Thank you.

[Applause.]

DR. FERREIRA-GONZALEZ: Thank you very much. We didn't mean for you to rush like that.

MS. TERRY: It's okay.

DR. FERREIRA-GONZALEZ: Any questions or comments? Julio.

DR. LICINO: Hi, Sharon. I have a question about is there a difference of opinion. It is really an umbrella group for like 600 organizations, as you were saying. So in terms of direct-to-consumer testing, which you can find susceptibility alleles but you can also find actual genetic disorders, what is the range of perspectives that you get from this very broad group that you have?

MS. TERRY: I should first say that this report does not reflect the 650 advocacy groups. It more reflects the people who attended the meeting. So it is not an advocacy report, really. It is a general report of the people who attended the meeting.

But to answer your question about what do consumers feel about direct-to-consumer testing, my 650 advocacy groups aren't terribly worried about it because it isn't their issue. They are really very much into single-gene disorder testing, and they don't care how they get it, basically.

More broadly, the consumers we represent in terms of public discourse and dialogue have a range of concerns. Most of them are somewhat concerned that what they are getting is what they think they should be getting and so they want to see oversight.

There is, though, certainly a fairly robust subsector who believe they should get any information they want in whatever form they want, and follow more the Amy Harmon article that just came out in The New York Times, having been tested by 23-ME and think that it is important to have this information even if, right on their website they say, tomorrow this could be completely different results. So we do see a huge range.

As a disclaimer or disclosure, I'm on the board of DNA Direct, for example, which really does more direct through health providers, but all of them want to see more clear oversight, more clear guidelines. I'm not sure that they know what they are asking for because I think as we move into that space we will have a better idea of how do we protect the public from fraudulent results and also how do we allow the industries that might result to move health forward, not so much this sort of more recreational stuff.

DR. FERREIRA-GONZALEZ: Kevin.

DR. FITZGERALD: Just picking up on that, Sharon, I noticed you have a couple of points about education. Maybe involved in that was also public engagement, but I'm just wondering if that came up as a specific thing and what sort of methodologies or approaches were recommended.

MS. TERRY: We decided at the meeting not to spend a lot of time on that because there are some good efforts. NCHPEG, whom you heard from this morning, I think is a good effort in that regard. Genetic Alliance itself has a whole wing devoted to quality information and public engagement.

What we got from the meeting, I think, was more a sense of we often silo audiences and we don't understand that these audiences in fact are overlapping. So the public and clinicians and researchers and test developers, et cetera, probably need some common forums where they can have these kinds of discussions. So the idea of more of these summits where people could come together in a cross-talk kind of way resulted.

For example, this report will be written in lay language with lots of glossaries, et cetera, so that the public can read it and not just isolate it to one sector of the stakeholder community.

DR. FERREIRA-GONZALEZ: Joe?

DR. TELFAIR: Thank you, again, for a good report on this. One of the things that struck me, though there is more than one thing. I know I have to be short, so I will.

It seemed that a lot more questions were generated than answered for this. Is that correct? So in your report, if you are going to direct that to consumers, then I'm assuming that there is going to be some degree of summation that is going to go out?

MS. TERRY: Yes.

DR. TELFAIR: All right. The second point, though, is that there seems to be a great deal of overlap with both your conclusions and your action items. For example, you have three in a row that actually overlap significantly one another, the registry, the reimbursement issues, and then evidence and outcomes data. If we pick one, it depends on what the context is.

I'm just wondering, in the discussion, if you can say a little bit about what was the thinking of demarcating those into separate summits as opposed to looking at those issues as they relate to one another. I'm thinking more in terms of level of application and level of use and practicality.

MS. TERRY: Those are excellent questions, Joseph. Basically, the reason that they sort of fell out of this in separate buckets were because of the depth of the summit that was proposed. In other words, certainly you couldn't do any one of these things without the rest, and there would be some presentation on all the rest. But there would be a great deal of depth.

For example, on reimbursement issues, to go very, very deep on that issue, bringing in the other issues around evidence. Evidence has got to be part of that, but then a whole other summit that would be dedicated to just looking at how do we, from the beginning of the research pipeline to the end, post-market data, look at evidence. So they certainly do overlap.

Our sense with things like this is that if we keep having very broad discussions we don't seem to go down in the weeds as much as we need to. We also don't seem to capture the right audience. Very often the reimbursement community doesn't come to these kinds of meetings. So I think we have to start marking some out and going in depth, but not ignoring the fact that there is a breadth of stakeholders needed at each of these meetings.

DR. FERREIRA-GONZALEZ: Mara and then Francis.

DR. COLLINS: Thanks, Sharon, for a very nice summary. I was fortunate to attend a chunk of the meeting. It really was a very useful exchange because of the way the format was set up. People were not talking past each other, they were really interacting. I think you got much deeper, therefore, into the sense of what needs to be done than often happens.

I want to ask you again about this mandatory registry, which came up in the previous discussion as well. Again, I think the sense of the Truth-Telling meeting was that this was so important that it ought not to be left to chance.

Do you want to say anything about the general sense of the group, if there was any worth reporting, about exactly who should run that registry or what combination of organizations should run that registry? Because, obviously, that becomes a critical question.

MS. TERRY: So while that wasn't exactly nailed at this meeting, and we had a couple of times we could go through that we had a panel discussion about issues like that. We also had a wonderful debate dinner, sort of in the European style, that in fact Stuart moderated. We needed a guy with a British accent to do that for us. Or, not British, sorry.

The sense we got there was that it probably had to be a federal agency, it probably had to be FDA, although again it wasn't nailed down, so I can't say that. If it was a public-private partnership, it might be a professional society or a coalition of laboratories, et cetera, that could be involved in that.

It didn't seem to the assembled masses there that a completely voluntary registry would result in the kind of data that we needed, and that some of that data is already available, although hard to get, and just needs to be expanded on.

So [it could] be married with something like GeneTest, that has begun a Mendelian disorder registry, and then moved over to FDA with some input from NIH. There was a lot of discussion around how those partnerships could be put together. But the sense was, if this is completely voluntary, it won't get done, which is what we know about all our kids, right?

[Laughter.]

DR. TUCKSON: By the way, as we get to the next point, I just want to make sure to highlight some of these key issues for the Committee, especially for new folk. This is why we have public comment. We had a discussion in our first draft of this and we said, in our initial outreach, the establishment of a voluntary system of genetic test registration through a public-private partnership. Now we are hearing that some of this feedback is perhaps that there should be a mandatory one.

I want to make sure that you ask the right questions to feel good that you understand why they do that. You may well decide based on this kind of feedback to change your original draft report to reflect this kind of input. So I'm highlighting this as one of the key issues that we are looking for as we go forward.

DR. FERREIRA-GONZALEZ: Sharon, let me further comment on these action items that you have and where you promote a mandatory registry. Was that a sense of most of the members? Were there any dissenting members? What was the sense of the group? I'm sure there were different views. What are the other views from the other groups that do not actually endorse a mandatory registration or a system at all?

MS. TERRY: Right. This was the sense of the meeting, at the end of the meeting. Now, certainly, some people had gone home. They didn't get to give their sense. I read these items out at the end and said, "Does anyone object to these items?" No one there objected.

Now, the report will be published, and people, the same way it is happening here, can comment on the report.

Would I say when the whole 200 were there would everybody have raised their hand for a mandatory registry? No. But the vast majority did think that a mandatory registry was probably the only way a registry truly would work.

The other thing I should say here is because we were truth-telling, it wasn't like "Do you want a mandatory registry?" Probably most people don't want one. They don't want a registry at all. They just want to get a good test and go home and know what they have.

But would a mandatory registry give us the result of leading to better health because of A, B, C, D, and all the way to Z? Most people said yes. But again, not unanimously, and like Francis' question, not nailing who exactly is going to run this thing.

DR. FERREIRA-GONZALEZ: Were there discussions on the value of that kind of registry?

MS. TERRY: I'm sorry. Say that again?

DR. FERREIRA-GONZALEZ: Were there discussions on the value of having a registry?

MS. TERRY: The discussion was on the value, whether or not it should be mandatory or voluntary, and who would run it.

DR. FERREIRA-GONZALEZ: Were there any discussions or any comments about genetic exceptionalism?

MS. TERRY: Yes, there was. The discussion pretty much went like what you just had before in terms of there are some extraordinary things these tests are doing in the sense of moving into a space we haven't regulated before in a particular way. There are also issues around risk, but there are also issues around the fact that these tests are very much like other tests. So the real intrinsic need here is to look at the value of the test, the risk of the test, et cetera, and not so much just chop something off because it is genetics.

MS. ASPINALL: That was part of my question, which is the issue that you talked about in terms of, on the action items, exploring the concepts of risk, because that is something that in various different ways has been part of many of the proposals and much of the discussion, as well as the issue of relative harm. In the EU, there is different regulation. Is there a different amount of harm. Maybe we will get to that from an international point of view.

Can you talk a little bit more about what the concept was about risk, risk profile, and how even at the most basic level now you would think about apportioning risk and potential harm or potential opportunity based on your report?

MS. TERRY: This was another area, and the reason one of the action items is to explore risk is we didn't have enough time there, nor did we have the right experts I don't think, to actually look at risk and to look at it not just in the field of genetic tests but across medicine, understanding it both from the clinician's side, the patient's side, the test developer's side, or the device developer's side.

I think what we saw there in terms of understanding risk was every opinion, from "There is nothing here, there is no risk, and so why has this been pulled out and treated specially?" in the case of, for example, IVDMIAs, to people saying there is a great deal of risk, including some of the advocates who aren't terribly much proponents of new technologies, et cetera.

So what we felt like is we really need to peel back people's complicated and confounded ideas about technology, that technology doesn't inherently mean more risk; about deliverables in terms of decision-making and in terms of life-altering matrices that you might have to go through; in terms of the complexity of algorithms and formularies that bring us to another point; in terms of what a clinician does or doesn't understand. Again, does a clinician understand what it means that my body mass index is too high versus a really complicated test that is done with multiple genes and an algorithm.

So there was no clarity, in my mind, at this particular meeting around this issue, which is why we pulled it out and said we need to really talk about how we are going to assess risk going forward.

DR. FERREIRA-GONZALEZ: Gurvaneet's turn.

DR. RANDHAWA: Thanks. I was going to ask this of Stuart and David, but I think, since you raised the issue here, we can discuss it now. I want to explore further the concept of registries and mandatory data submission.

I'm not quite clear yet as to if you are thinking the registries are there to ultimately improve health. That means we are thinking of linking clinical data and outcomes data with the lab data. There are several issues entangled here. One is of course the data submission by the lab developers prior to approval, getting that in there. Whether or not there are clinical outcomes dealing with that, we already know that is not the case. That wouldn't solve that problem.

Then, after the tests have undergone approval, they will be used in the real clinical world, where the clinical conditions are in different databases and not really under the control of lab developers.

Then there is a third issue of where do you draw the line between a genomic test or a biomarker and another diagnostic test.

So I'm going to think this through and see how are we going to get a registry that will be having pre-market data, post-market data, health outcomes data, and be mandatory. Can you shed some light on that?

MS. TERRY: No.

[Laughter.]

MS. TERRY: Gurvaneet, those were also issues that were discussed at fairly long and lengthy discussions that didn't lead to a lot of clarity in my mind, either. There are going to have to be some dividing lines, and there is going to have to be an attempt made to get some clarity around this.

The very simplest things could be just that the molecular tests are in a registry like gene tests and that is blown out or expanded on in terms of what labs are doing those tests, whether or not they are CLIA-approved, and whether there has been proficiency testing, all the way to what probably in terms of health would be much more valuable, and that is an aggregation of all the data that you just mentioned.

I think there are lots of other issues inherent in the problems around that, including the fragmentation of our healthcare system, and ones that we probably can't overcome in terms of dividing strict lines between the various kinds of tests and evidence. So I don't have an answer to that, except to say that I think we should wrestle with the question. I think as we do try to integrate genetics into medicine and also improve our healthcare system, we shouldn't just accept that it is fragmented and broken and so we are going to stay broken here, too, but maybe have this field blaze a pathway into the brave new world.

DR. FERREIRA-GONZALEZ: Thank you, Sharon, for sharing these important points. I'm pleased to see that many of them echo some of the findings of our Committee.

Our next speaker is a friend to our Committee whom we have seen before, Mr. David Mongillo, who is the vice president for policy and medical affairs at the American Clinical Laboratory Association.

DR. TUCKSON: As David comes up, I want to mention Amy Harmon's name came up from The New York Times. I do want you to know two articles ago I did call Amy Harmon up and invited her to come, just to be able to meet someone who was doing such terrific, incredible work. She was very eager to do it, but checking with her editors, they wanted her to finish her series before she entertained such a thought because they wanted to be very careful about separating those things out.

So she is going to eventually, I think, accept our invitation to come and meet her and just hear a little more from her about the stories and the folks that she has met and the impressions that they have left. But it will just be whenever she completes this little series of stories. Anyway, I just wanted to make sure that you knew that we had reached out to her.

Remarks by David Mongillo

MR. MONGILLO: Thank you, Andrea, and thanks to the Committee for allowing us the opportunity for public comment. My comments will be brief, partly because we are still digesting the draft report and partly because you have had a very full agenda with a lot of complex topics. It is not long before everybody gets a chance to go home here.

We think this is an important report. We believe it is critically important and it is really going to serve as a roadmap -- which is, I think, a term that Dr. Tuckson had used when he identified the importance of this report -- a roadmap, really, for the future of genetic testing oversight, which has so many implications for so many components of 21st century medicine.

We want to comment on three what we think are key areas of the report. The first has to do with the oversight role of the federal agencies. We certainly share the Committee's goal to bring the full promise of genetic and molecular medicine to the healthcare system while incorporating the highest quality diagnostic tests. As such, we agree and can work with the Committee to gain consensus on many of the report recommendations.

However, to ensure continued innovation in laboratory medicine and to provide continued patient access to laboratory services, it is critically imperative that CMS, as the agency responsible for CLIA, the Clinical Laboratory Improvement Amendments, continues as the lead agency responsible for the oversight of laboratory-developed genetic test services.

That is in no way suggesting that there isn't a very critical, clear role and definitive role for FDA in this process. In fact, FDA should have a significant role. They should be involved with CLIA in reviewing clinical validity claims and promotion of claims for certain high-risk laboratory-developed genetic tests.

We heard earlier that there were some recommendations made about some of the issues and concerns that people have identified in this area. We think there can be models, and we are posing models, that would deal with the information disclosure, independent validation, enhanced QA and QC, and enforcement if the claims are not met.

As the report stressed, interagency coordination is key and fundamental to ensure that this oversight is least burdensome and does not place unnecessary or duplicative regulation on clinical laboratories providing these genetic test services.

ACLA supports the report's additional recommendations for HHS to convene a workshop with the relevant agencies, as well as stakeholders, to provide input into the development of a risk-

based framework for the regulation of genetic laboratory-developed tests, and encourages and supports the development of new and transparent models for private sector or public-private partnerships.

I have heard ACLA's position characterized as sort of the JUNC, "Just Use Normal CLIA" approach, and I want folks to realize that we really are not saying that. We really are proposing models that really incorporate some really, we think, innovative interagency coordination and some, as I said, opportunities for full disclosure, full transparency, third-party reimbursement, enhanced quality assurance and quality control, and enforcement.

The second area that we want to mention briefly is the implementation and timing of the report recommendations on interagency coordination. To allow for a well reasoned and orderly regulatory process, ACLA urges the Committee to include one critically important stipulation in the draft report, namely that the report recommendations should be implemented and understood before the FDA's IVDMIA guidance is finalized or its ASR guidance is enforced.

The benefit and information to be derived from these well thought out recommendations will inform and therefore should precede further guidance and regulatory action.

Finally, the section on effective communication and decision support is particularly noteworthy. We face a critical dilemma for healthcare delivery in the 21st century. Genetic and molecular medicine will revolutionize the ability to capitalize on preventive medicine and target therapeutics but will also become increasingly complex in nature and more available through electronic communication directly to the consumer.

Clinical labs play a critical role in healthcare delivery by allowing for the rapid and timely utilization of health information by providers. The reach of laboratories into physician offices and hospitals by means of health information technology is unparalleled.

ACLA pledges its support in working with the Committee and other interested entities to ensure that clinical decision support systems effectively communicate the appropriate information to providers and consumers in a timely manner and with the necessary level of information to make informed decisions about effective health care.

We are reviewing the full draft report. We plan to provide written comments by December 21st. We thank you for the opportunity to comment and look forward to working with you and the agencies to finalize this process.

DR. FERREIRA-GONZALEZ: Thank you, David. Any questions for David, or comments?

[No response.]

DR. FERREIRA-GONZALEZ: Thank you.

[Applause.]

DR. FERREIRA-GONZALEZ: Our next speaker is Ms. Patricia Goldberg, who is here today representing the International Society of Nurses in Genetics.

MS. GOLDBERG: Good afternoon. I'm Patricia Goldberg, speaking for ISONG today, the International Society of Nurses in Genetics. Our membership spans six continents and represents

nurse clinicians, nurse educators, and nurse researchers. ISONG is a specialty nursing organization dedicated to caring for people's genetic health through excellence in the provision of genetic healthcare services by fostering the professional and personal growth of nurses in human genetics.

My brief remarks are part of a longer statement that will be submitted in December responding to the Committee's draft report on the U.S. System of Oversight for Genetic Testing, A Response to the Charge of the Secretary of HHS.

ISONG is enthusiastic when there are advances in genetic testing that our membership can use to improve health care for our patients and the public in general. We consider the work of the SACGHS as a serious public health endeavor and the draft on the genetic testing as a valuable contribution that represents a work in progress.

We take seriously our commitment to our patients and the public's right to the highest quality genetic health care. Patients expect us to ensure that the information they receive is accurate, valid, reliable, and truly useful as they struggle to make informed decisions. We hold this public trust as the highest measure of our success as nurses in genetics.

It is this responsibility that drives our concern regarding the lack of evidence and clinical validity and clinical utility of the genetic tests that are being advanced as useful for common disorders such as diabetes and hypertension.

We suggest that more attention be given by SACGHS to the interpretation and application of the genetic and genomic results obtained by direct-to-consumer tests for these common disorders. Even though the draft report on the U.S. system of oversight of genetic testing notes that counseling will be given, we believe that there is still too little data on the accuracy, reliability, and true usefulness of the results.

Our responsibility to our patients and the public to provide the highest level of counseling warrants our concern in this area.

Also, as nurses, we are very concerned about the genetic testing being marketed to the consumers outside of the patient-health provider relationship. For example, over the Internet. We urge the Committee to recommend oversight of false marketing of testing aimed to identify made-to-order weight loss plans based on genetic makeup.

Another potentially dangerous area of inappropriate marketing relates to testing to identify the individual's rate of metabolism of drugs so the individual can inform his or her healthcare provider what drugs and dosages to prescribe for them.

We also find the offer of testing for ethnic background to be especially misleading and potentially divisive for families and communities. This medium of exchange requires unique and greater protection for consumers.

ISONG is committed to working towards ensuring that nurses and other clinicians are well prepared to serve responsibly their patients and the public's need for genetic information. ISONG is committed to ensuring that all individuals have appropriate access to genetics and genomic health care, and part of that responsibility includes access to accurate, valid, reliable, and useful information.

Thank you. Any questions?

[Applause.]

DR. FERREIRA-GONZALEZ: Reed, do you want to make a comment?

DR. TUCKSON: I just want to, again, remind folks that was very helpful because, again, in our current iteration of the draft we expressed concern about certain types of health-related genetic tests that are marketed directly to consumers and appear to fall outside of the scope of CLIA. Some nutrigenomic tests, e.g., for caffeine metabolism, and tests to determine the gender of a fetus are examples of health-related genetics tests that are skirting the boundaries of CLIA's authority. There is insufficient oversight of laboratories offering such tests and their potential impact on the public health is increasing concern.

So I think this direction, again, just reminds you that what Patricia has done is to speak specifically to one of the issues that we have highlighted in the report.

DR. FERREIRA-GONZALEZ: Kevin?

DR. FITZGERALD: Yes, I too would like to thank you for the letter and the comments. I would like to dig a little deeper into the specificity, if you are prepared, or to ask ISONG, if they would, to perhaps respond at a later date, if that makes it easier.

But the two areas of specificity which I think we would find particularly helpful would be what sort of oversight regulation did you have in mind specifically, and secondly, even at the end, the very last words you used, you talk about "useful information." Who gets to decide what is useful information? What if the public decides that finding out their genealogy they consider to be useful information. How do we engage in that process of determining what "useful" is and who gets to have input into that?

MS. GOLDBERG: I know they are developing their opinion on that. That is part of what they will be talking about in December.

DR. FITZGERALD: Thank you.

DR. TELFAIR: I just want to piggyback. That was actually part of a question that I had as well, the specificity, particularly in relationship to the issue of marketing, to whom, and then who makes that decision as to what information actually goes.

One of the concerns is not only just the test and the type of test but also assumptions about the population itself in terms of receptiveness. Then, also, I know that behind all this is this question of duplicity to the public itself and duplicity to these groups. I'm just wondering, given who you all are, could you also cover that?

I respect a lot of what you all do because several of the groups I work with are disease-specific international nursing groups. I know that that is a real concern that they have as well. So I was wondering, piggy-backing on what Kevin suggested, could you also, or will you also be able to address that issue as well?

MS. GOLDBERG: You also want us to address -- I'm not sure what it was.

DR. TELFAIR: There is a question, and I'm actually trying to just be diplomatic about it, because --

[Laughter.]

DR. TELFAIR: What I'm saying in another way is that certain groups are targeted for certain types of drugs in terms of what they believe about that and also in terms of who they are. I was just wondering if in your deliberations related to duplicity, dealing with the issue of duplicity, which seems to be underlying some of the marketing, will you be addressing duplicity as an issue and suggestions on how that could be part of the regulatory process and could be addressed as it relates to specific subpopulations and other groups.

MS. GOLDBERG: I'm not sure, but I will have to ask that of one of our representatives of ISONG that is sitting back in the back, if it is okay. Or she was.

DR. TELFAIR: Knowing who you all are, I have been told that is a legitimate question to ask.

MS. GOLDBERG: Right. We will have to address that with the leadership in time for the December meeting, when they will be submitting another report.

DR. FERREIRA-GONZALEZ: No more questions? Thank you so much again.

Our last speaker, but not the least, is Dr. Patrick Terry, who is here today representing the Coalition for 21st Century Medicine.

DR. TERRY: Thank you. Hello, everyone. Thanks for this opportunity. The Coalition of 21st Century Medicine also wants to thank the overall Committee and the ad hoc working group for putting this report together. I agree with David Mongillo's assessment of the importance and the timeliness of this particular report.

My name is Patrick Terry. I'm one of the cofounders of Genomic Health. Genomic Health's product, OncoType DX, would squarely be in the target area of IVDMIA, so full disclosure on that activity. We are a California-based diagnostic company.

But I'm also one of the founding members of the Coalition for 21st Century Medicine, which is a group that is self-organized around this issue of oversight and regulation and includes industry groups, venture capitalists, academic groups, as well as disease-specific patient organizations, with the concern of balancing oversight and regulation with access and innovation.

Specifically, I wanted to share with you, as a direct result of the Genetic Alliance Summit, the Coalition has tackled a lot of what was identified as the challenges and the opportunities here for what was, I think, very well described by Stuart's presentation. We fully embrace a lot of Stuart's concepts and proposed solutions. We are crafting a private sector regulatory initiative to present to HHS and to FDA in the near future.

Just quickly to go through it, the proposed framework is based on the following concepts: the importance of advanced diagnostics and their continued development, the importance of reimbursement, the rationale for a revised regulatory framework, which I will speak a little bit more about, and the detailed regulatory and subregulatory approaches that HHS and FDA and CMS can apply and consider moving forward.

The focus of the framework is to identify IVDMIAs as well as multiplexed ASRs that do indeed need enhanced oversight and to clearly define a risk-based approach that includes mandatory pre- and post-market requirements which include a mandatory registry.

The goal of the proposed regulatory framework is to offer specific and detailed regulatory approaches for IVDMIAs and multiplexed ASRs that provide a clear and defined role for CLIA and FDA in a joint framework and that also provide a predictable pathway and a set of expectations for test developers, industry, and the investment community.

The regulatory framework attempts to balance and to achieve a balance between the following principles: innovation, timeliness, transparency, truthfulness, and risk-based regulation.

Finally, in conclusion, the 21st Century Medicine Coalition will formally submit this proposal of a private sector regulatory initiative and will outline alternative model approaches as well as a phased implementation strategy to HHS and to FDA in the near future. The Coalition is more than willing to share that document with the ad hoc working group and to review and discuss further this private sector initiative as you move forward with the finalization of your report.

Again, thank you for your attention.

[Applause.]

DR. TUCKSON: Let me just make sure I got that. That is great. That is terrific. You have a response to this. It sounds like what you are saying is that the private sector is saying it is trying to step up to the plate and diminish the need for more regulation and oversight in this area, that you are responding to the needs.

DR. TERRY: Right.

DR. TUCKSON: So you are going to send that report in.

DR. TERRY: We will submit written comments to the draft report that you are crafting as well.

DR. TUCKSON: All right. That is what I wanted to key [on], that we would get that in time to consider those things that you are doing.

DR. TERRY: Yes.

DR. TUCKSON: Thank you. That was key.

DR. FERREIRA-GONZALEZ: That was the question that I had, are you actually going to respond to the Committee. But as you come up with some final draft or any version of a document that you can share with our steering committee, we will welcome that as part of this open process that we are trying to establish.

Kevin.

DR. FITZGERALD: Thanks again, Patrick, for that and the work that you are going to be doing. That is great. To follow up on what we have been asking the other panelists, can you give more details about the process you are going to use to ascertain your risk-based platform, your approach, how you are going to identify and delineate risk?

DR. TERRY: There is a variety of model solutions that are being proposed, and there are pluses and minuses to a variety of solutions. I think the context in which we want to present these solutions is not that they are canned fixes but ultimately that these are fodder for further dialogue and debate with the regulators and with HHS.

Part of the solution would be an expert third party review. Another suggestion is return to the post-1976 medical device regulation. During that implementation, there were risk classification panels. [We could] return specifically to a formal mechanism such as that.

So there is a variety of past activities that the agency has implemented in the past to deal with risk and also to add this issue of having important clinical relevance on the table when risk is being assessed for a particular test. So in the absence of the reality of clinical care, you can have a risk-based assessment. But with the reality of benchmarking against trial and error or the standard of care for a particular disease state, the risk-benefit calculation could dramatically change.

So part of the third party mechanism would allow the agency to convene experts between particular disease categories or technical expertise around a test or a technology to participate in a risk-based classification.

DR. FITZGERALD: Great. Thanks very much.

DR. FERREIRA-GONZALEZ: Thank you very much. Any other member of the audience that would like to do public comments at this time?

[No response.]

DR. FERREIRA-GONZALEZ: Before closing the session, I would also like to extend an invitation to other members of the SACGHS Committee that want to be part of the steering committee. You are welcome. Again, we are currently a village, so the more the merrier.

[Laughter.]

DR. FERREIRA-GONZALEZ: So I formally invite you. Just make sure you let Sarah know if you want to be part of the steering committee so you get added to the different rigorous teleconferences that we are going to be having once a week.

MS. CARR: One a week in January. There are five weeks in January.

DR. FERREIRA-GONZALEZ: Oh, I just realized that. Okay.

[Laughter.]

DR. FERREIRA-GONZALEZ: Anyway, I want to thank everybody for very thoughtful, good presentations. We look forward to all your written comments.

[Applause.]

DR. FERREIRA-GONZALEZ: Reed.