

Discussion

DR. TEUTSCH: Thanks to all of our speakers. We have obviously had a tour from the importance of getting measurement accurately to what the future world might look like.

We have just a few minutes, and I think we should take this opportunity to ask questions of any of our speakers who are still here or to have a discussion among ourselves. Let me open the floor for a couple of questions.

Let me ask you, do you have any additional comments that you would like to make from the CDC perspective?

DR. KALMAN: We think that having reference material is really key to assuring the quality of these tests not only for the day-to-day QC of the tests but also for proficiency testing, which is a big deal. It was quite a large part of the Oversight report that this group did a few months back.

We did a count. I think there are about six different diseases for which there are higher-order reference materials either from NIST or FDA or something like that. We count six. On the Gene Test website, there are over 1,300 genetic tests currently available. That is a really small fraction of the current tests that are available.

So the CDC, through the GeTRM program, is trying to address this gap by just simply organizing a volunteer effort among the people in the genetic community. We are just characterizing publicly available cell lines and DNA from the Coriell repository so that we have a larger supply of materials so that we can feel confident in knowing the genotype of these and so labs can use them for quality control and also the proficiency testing needs.

Right now the projects that we are working on are pretty much all being driven by requests from CAP for proficiency testing materials. We are starting a real large project for pharmacogenetic materials. We are going to do over 100 DNA samples for five pharmacogenetic loci. We are going to get other data from other labs as well on other loci. We are going to try to do a project for array CGH.

We were trying to do a project for Duchenne muscular dystrophy, which is something that CAP asked me to work on, but all the labs are stopping their testing because of the patent issue. So I don't know what is going to happen.

DR. TEUTSCH: Coming full circle. Andrea.

DR. FERREIRA-GONZALEZ: I want to thank Lisa for a tremendous effort and the role that she has played at CDC in getting the GeTRM program started and being one of the strongest advocates for this. I think she needs a round of applause from all of us.

[Applause.]

DR. FERREIRA-GONZALEZ: That said, like you said, there is a lot more work that needs to be done. But I think it is interesting that you have already identified through the collaboration with professional organizations or end users of different laboratories what are the current needs of the laboratory not only in proficiency testing but also reference materials that we can use to analytically validate the assays and continue quality control.

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I was wondering, what is the level of cooperation between the GeTRM program and the NIST genomic program. I think a lot of the work that you have done in identifying some of the needs can be translated and the deployment of the work NIST can take over.

DR. KALMAN: I do talk to NIST on a regular basis. Our program has a yearly advisory committee meeting. We always have a few people from NIST at our meeting, so I talk to them. Also, in the area of molecular oncology there are a few people from NIST that I have been talking to.

So, yes, I try to keep the communication lines open. But if you want to talk some more, that would be great.

DR. BUTLER: Margaret Klein went to the meeting that you had last month. We are looking forward to working more with you in the future as we get more into future genetic tests.

DR. TEUTSCH: Marc.

DR. WILLIAMS: I was going to ask Andrea's question. But then as Mike spoke, I said, if that is the vision of where things are going, then in some sense is investing a lot in genomic validated samples really worth it if we are really going there.

I guess the question that I have -- and probably you or Dr. May would be the best ones to address it -- would be, what is your real vision about where you are going to need to invest your limited funds in terms of standards in the biomedical realm? Is it going to focus on genomics? Is it going to focus on proteomics or metabolomics? Are you going to try and do it all?

DR. MAY: I think, in the short term, Mike's vision is 2020. We have a lot of living to do between now and then.

Certainly, in the short term, the focus of the NIST's new activities is going to be on medical imaging and protein measurement science, for sure. Beyond that, we might do some other things.

If you are looking at the near future, I think for the next two to five years the emphasis is going to be on improving our capabilities to support medical imaging and developing more core competencies in protein measurement science.

That would address lots of things. It would address this disease signature issue that Mike talked about, as well as the issue of follow-on biologics.

So we are trying to increase our core competencies and put more tools in the toolkit to address a number of things. Now, in the longer term, we are still going to continue our work in genetics. We are not going to stop those things. But if you look for areas that across all of NIST we are going to expand in, it would be those two.

Now, putting on my director of the Chemical Science and Technology Laboratory hat, certainly in the Biochemical Science Division there is going to be a greater emphasis on genetic testing and DNA-based diagnostics. As John mentioned to you, we have just done some reorganization within our Biochemical Science Division to address just that issue.

DR. WILLIAMS: In follow-up to that, our Oversight report identified, as Andrea pointed out, that this PT issue and having samples is a huge issue. We have 5,000, plus or minus, genetic tests

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that are out there and a small fraction of those actually have PT materials that are available and in use.

From what I'm hearing you say, I think it may be unrealistic to expect that NIST is going to be the savior riding in on the stallion at this point.

DR. MAY: That is true. But certainly, if that is a major issue that your Committee has identified, sending a note to me to that effect, perhaps with a copy to the acting NIST director, would not be a bad idea.

DR. AMOS: Marc, just let me say one thing. It is clear that genomics is going to be an integral part of the disease signature. I think that the discovering technologies of the future are really going to focus on the ability to understand the environmental effect on the genome. So you have to have good genomic data to do that. There are all sorts of issues with the sequencing things that are going forward.

I think my colleagues have decided that genome-wide association studies are something that we don't want to do. We are looking at next-generation sequencing. I will put it that way.

DR. TEUTSCH: Mara, you get the last word.

MS. ASPINALL: I think I also, once again, agree with where Marc is going. So this has truly been a red-letter day.

DR. TEUTSCH: It is a great place to end the meeting.

DR. WILLIAMS: She is going to hit me up for a drink later.

[Laughter.]

MS. ASPINALL: The question really, Steve, was to you. I think this was a great session, with the ability to hear the different perspectives of what is happening today and getting the various approaches to that. What role do you see SACGHS taking? This is great information, but I know that tomorrow we are going to jump into priorities going forward. Where do you see this going?

I love the idea of taking some action and sending some letters to NIST. As Marc said, this is, to me, entirely consistent with the recommendations not just in the last report but in the last two that talk about gaps and the need for essentially standard-setting or ensuring quality across the system. Now we have an opportunity that doesn't require potentially major changes in legislation by Congress or otherwise but just a prioritization. I would vote for taking some action to at least enforce that.