Public Comments

MS. CARR: Is Gail Javitt here? Do you mind doing it now, Gail?

MS. JAVITT: I'll speak fast.

DR. TUCKSON: No, no. You don't have to speak faster because we want to hear you. So we'll hear the public comment. We'll take the break, and then we're going to come in and roll up our sleeves and drill deep at the 100th power of the microscope.

Thank you and welcome. Please introduce yourself for the record, and we're glad that you came to speak to us.

MS. JAVITT: Thank you. My name is Gail Javitt, and I am Law and Policy Director with the Genetics and Public Policy Center at Johns Hopkins University. I appreciate the opportunity to appear here today and to speak to you about the center's concerns with respect to engagement testing quality and pharmacogenetics.

As this committee has recognized, pharmacogenetics holds great promise to improve the public's health by improving the safety and effectiveness of pharmaceuticals.

But the success of pharmacogenetics is predicated on a robust pipeline of genetic tests, and these tests need to accurately and reliably detect variations in DNA. This means, in turn, that the laboratories that do the testing need to have the capability to perform these tests accurately and reliably, that the tests themselves provide clinically valid information, and that health care providers, as this committee has discussed, know how to interpret the results.

We are concerned that the current regulation of genetic testing is shaky and is not strong enough to support the foundation on which pharmacogenetics is based. Although the public widely believes that the government regulates the quality of genetic tests, for the most part this is not the case.

First, today there is no specialty area for genetic testing laboratories under the Clinical Laboratory Improvement Amendments of 1988, or CLIA, as it is better known. This situation exists despite the fact that this committee's predecessor, the SACGT, recommended the development of such a specialty, that the CLIAC developed a proposal for such a specialty, and that CMS indicated in 2000 that it intended to create such a specialty.

So we at the center were curious about why there might be such a delay of six years. We wondered whether perhaps the comments that were submitted in response to the notice of intent of 2000 were overwhelmingly negative, leaving the agency to perhaps change its mind. So we undertook a thorough review of those comments, but instead of overwhelming opposition, we actually found significant support for the creation of a specialty, although there were differences of opinion about the details. But on the issue of proficiency testing, which we believe is the central element of ensuring the accuracy and reliability of testing, there actually was consensus that a specialty should be created.

So based on our analysis, in November of 2005, we sent a white paper to CMS Administrator Mark McClellan, along with a letter urging him to issue a proposed regulation for a genetic testing specialty under CLIA, and subsequently the Genetic Alliance sent a similar letter.

So turning, second, to the genetic tests themselves, there are gaps in oversight here as well. Today there are two paths by which a genetic test can come to market and be offered to the public. Genetic testing laboratories, in performing genetic tests, can use a so-called test kit or they can make the test themselves in-house. The vast majority of tests are performing using in-house technologies.

As you've already heard, if a laboratory uses a test kit, the manufacturer of that kit must first obtain approval from FDA, and FDA assesses both the analytical and the clinical validity of that test kit, ensures that the labeling is adequate, that the claims made are supported by the data, and that post-market surveillance is done, and any adverse events associated with the kit are reported.

In contrast, if a laboratory uses an in-house test, there is no pre-market review of that test. Some components of the test may have to conform to FDA labeling and good manufacturing practice requirements, but there's no expert body to review the test and ensure that it actually detects what it purports to detect, and that the mutations detected are clinically relevant.

So not surprisingly, of the more than 900 genetic tests that are available today, a very small handful are actually sold as test kits, and an even smaller handful of the handful relate to pharmacogenetics. To compound the regulatory inequity, even once a test kit is approved for a particular indication, a laboratory can offer its own proprietary test for the same indication and this actually is already happening in the case of pharmacogenetic tests.

As we've heard, FDA has, in the past few years, recognized the potential value of pharmacogenetics, and its guidance documents indicate that the agency intends to regulate pharmacogenetic tests, although the precise framework is still evolving. But these diligent efforts will be undermined unless FDA's requirements apply to all pharmacogenetic tests regardless of where they are produced.

The absence of adequate oversight also means that physicians and the public are hard-pressed to distinguish the good performers, the good tests, from the bad tests, and they have little assurance that the tests they're using to make profound health care decisions are reliable and relevant predictors of disease risk or treatment outcome.

So if pharmacogenetics is to gain the public's trust and, equally important, the trust of payers, and to deliver on its promise of health care improvement, there must be a sufficient level of confidence that the laboratories offering the tests are performing them correctly and that the tests yield information that is relevant to health care decision-making.

Getting to the system that is worthy of public trust will require the Department of Health and Human Services to give the necessary direction to the agencies that are involved in overseeing both the laboratories and the tests and to implement needed changes.

So we would encourage this committee to make a recommendation to the Secretary that CMS issue a proposed regulation for a genetic testing specialty under CLIA, and we would also encourage this committee to recommend to the Secretary that a regulatory framework be established for genetic tests to ensure that they are clinically valid, regardless of whether they are performed using a test kit or an in-house developed method.

And I appreciate your time. Thank you.

DR. TUCKSON: Well, thank you very much.

Let me just ask. Again, we're pretty bright, but does anybody have any questions they want to ask?

I was trying to allude to the fact that I wanted to revisit some of the strategic planning priorities at the end of the session tomorrow, and I know that there are going to be some of us around the table who feel strongly about maybe not revisiting the oversight question because, Lord knows, it's complex. But this issue generically first is something that is on my mind about do we really need to make sure we're clear about where the holes are between CLIA and FDA and FTC and others. You know how we resolved all the home brew. This has been worrying me in terms of our responsibility to the Secretary in terms of continuing to make sure that we're clear on if there are holes, and if there are, either we accept those holes or don't worry about them.

So, anyway, I'm telegraphing that this is something that I'm concerned about. I don't want to live with the anxiety as the chair about this a lot longer, so I'm going to dissipate my anxiety into each of you.

Here we've got that same issue being raised, as well as the specific issue of the notion of a special discipline that you're advocating for. So I'm going to come back to this tomorrow.

By the way, I'm giving fair warning to the committee to kibitz me at the break or anywhere else if you want to try to tackle me and prohibit me from going down roads that you really don't want me to go down tomorrow. So I'm giving you a chance.

But meanwhile, are there any questions that you want to ask specifically of this report before we get to that larger discussion tomorrow?

DR. LEONARD: I'd just like to lend support. It would be great if CLIAC could come out with their genetic CLIA recommendations or subsection. It's been missing in action for I don't know how long.

DR. TUCKSON: Exactly. Right.

DR. LEONARD: But I ask all the time, and it's coming, it's coming, it's coming is all I hear, but it never appears. So can we put this on the agenda to get an update and explanation as to why it's missing in action for so long?

DR. TUCKSON: This is where I'm coming from. So could you make sure, knowing that I have a memory like a bowl of jello here? This is what we want to revisit. Would you write down that particular recommendation? We'll do that tomorrow. Terrific. That's just what I'm getting at.

Are you guys good. I pulled out of my notes that were just handed to me. CMS says genetic testing specialty under CLIA rules, forthcoming.

PARTICIPANT: What's the date on that?

DR. TUCKSON: Now, first of all, I'm not going to do that to CMS. I'm just letting you know. We like CMS. We're not going to mess with them. Besides, people report on everything we say. It comes back to haunt you. So we'll give them a chance to respond.

Anything else on the recommendation, which is actually an interesting one? Do we have it in writing yet?

MS. JAVITT: I'll be happy to submit it in writing.

DR. TUCKSON: That's a good answer.

One was the CLIA and one was the --

DR. LEONARD: Right, and the other one, CAP has gone to including the review -- there's a specific now review in their checklist. So every two years, any new tests that are brought on-line need to be reviewed during that review process. But that's not part of CLIA. So if that were addressed under CLIA, then it would become more generally applicable for anyone doing inspections of laboratories. So that's one way that that second oversight of non-FDA-approved or reviewed test kits or genetic tests done using non-FDA-approved or reviewed test --

DR. TUCKSON: So we'll bring that up again tomorrow. Did you get that second one? If not, get with Debra. That was perfect.

All right. You're going to submit that in writing so we'll have it?

MS. JAVITT: Yes.

DR. TUCKSON: Thank you.

By the way, is Marilyn Zigmund-Luke around? You might have wanted to testify?

MS. ZIGMUND-LUKE: I'll talk tomorrow.

DR. TUCKSON: Okay. We'll get you when you do come tomorrow.

Thank you. Good job.

A 5-minute break.

(Recess.)