Host: This is the podcast from *Clinical Chemistry*. I am Bob Barrett.

Although the majority of all clinical testing is performed in a central laboratory environment within the hospital or reference laboratory, there is a growing trend to provide laboratory results in near-patient or point-of-care testing environments.

For waived, low complexity testing, this approach may be appropriate, however, direct-to-consumer, or DTC, marketing has also made highly complex testing available to the consumer through self collection of a cheek swab and routine mailing to specialized laboratories for genetic testing and risk assessment.

In the July issue of *Clinical Chemistry*, five professionals representing academic clinical laboratories, medical genetic subspecialties and DTC laboratories commented on this growing trend. Our guest in this podcast, Dr. Gregory Tsongalis, Director of Molecular Pathology at Dartmouth Hitchcock Medical Center continues that conversation.

So, tell us Dr. Tsongalis, can you explain what direct-to-consumer genotyping entails?

Dr. Gregory Tsongalis: I think just for a historical perspective, I need to mention a couple of other types of testing for us. Traditionally clinical lab testing has always been done in either a hospital-based lab or national reference lab or so on, and then we moved away from that with point-of-care testing or near-patient testing with indications from the FDA for low-complexity testing or waived-testing. And there are few good examples of that happening, something like the glucose monitor for diabetic care is monitoring this issue, measure their own glucose levels with.

There are other over-the-counter types of tests that may not be clinical tests per se, but the patients or consumers did buy it at the local store, and performed testing, a home pregnancy test, is a good example of that. And what we are seeing now is this whole idea of direct to consumer advertisement and offering of tests, whereby a consumer could send a specimen into a laboratory and not necessarily, and more often than not a clinical-based laboratory.

They would send the sample in and have the test done. When the sample is submitted, there is also an exchange of money and end results gets sent back to

the patient. So this becomes a little bit concerning, because some of the testing now is entering this whole realm of genetic testing and genomic medicine and personalized health. Typically what's happening with a direct-to-consumer type of tests, especially for genetic is that consumers will log into a website for some laboratory, and they will register themselves as a patient in that system, and then they will get notification for payments and they will need to pay by credit card or send a check, and once that happens, it triggers the number of events, including the labs sending a collection kit or device to the patient.

For most instances, it is some type of a swab that you would get a buccal cheek swab specimen from, and send it back to the lab. They would do the testing, and then the results would go back to the patient or the consumer in this case. So, that's where this all has been progressing, and it's progressing very, very quickly to a point where this is now becoming quite a market and profit to revenue generator for a lot of laboratories.

Host: Are these tests typically clinical diagnostic tests?

Dr. Gregory Tsongalis: They truly are not. And a lot of these tests are more behavioral response types of tests. Some of them are risk assessment for certain disease types and categories, but they are not what you would think of as a traditional clinical diagnostic test.

There is really not a good association between some of the assessments and data that comes back with any disease state. And I think part of the problem is just that that these fall under the category of risk assessment versus diagnostic testing, so they tend to get waived or got regulated as tightly as a clinical laboratory would.

Host: Well, with that in mind, is there a demand for these tests—there must be a demand for these tests, and if so, how exactly are they ordered?

Dr. Gregory Tsongalis: Well, I think—you know, you are right. There is a demand for these tests and consumers want to know what's their risk of developing certain neurological disease or cardiovascular disease or cancer and so on and so. There are a lot of curious minds out there, and so I think that there really is a market for this type of testing, and unfortunately I think it's a little bit of a disservice to the consumer, because the

information they get back is really not as healthy as they are made to believe.

Having said that, they usually are ordered over the Internet, where they can log into a website, and register yourself with all your demographic information, and of course, credit card payment information, and then that sets the ball rolling for a collection kit being delivered to your doorstep at home.

Host: I understand the FDA is getting more involved in this area? Would you tell us a bit about that and do you agree with their recent action to increase oversight of these labs?

Dr. Gregory Tsongalis: Well, I think this is really a good thing, because whether these are truly diagnostic tests or not, you are really sending out a result of a laboratory assay, and I think more oversights from the FDA is really a good thing in this case that a lot of these laboratories are operated kind of like research labs, and I don't want to say that they are all bad, but certainly we would like to have their performance, characteristics, and the guidelines on par with the clinical laboratory.

I also think most very recently the FDA made a ruling against the collection device being put in all the Walgreens around the country, and I think one thing that we forget about is that this is still very high complexity testing, just because you can send in a buccal swab or may be a blood spot on a piece of filter paper or a hair sample, doesn't necessarily mean that the testing itself is not high complexity, and I certainly think that it is, even though the collection of the specimen may be of low complexity.

Host: In your opinion, are consumers being best served by this type of laboratory tests?

Dr. Gregory Tsongalis: That's a great question, I am split. Half of me wants to say, no, they are not, and the other half of me says, well, wait a minute, there are certain tests that if people want to know, they should have the right to know, and these laboratory serve that function. Where I have a problem with all of this is in the interpretation of the results, and a lot of these labs if you go to their websites and you actually read the disclaimers that they have attached to their reports, they are very, very significant and you almost have a better chance of flipping a coin to determine whether you are at risk for a disease or not, versus having one of these tests done.

Host: Where do you see this type of testing going in the next few years?

Dr. Gregory Tsongalis: Well, I think certainly these types of tests could be very, very useful in patient managements, in personalized health and so on. I suspect that we are going to see more of this before we see a less of it. I also suspect that the FDA is going to have a really big say in how these laboratories get regulated and what they can and cannot claim as part of their results and the reporting of these results, but I don't think it's going to go away. I think that this is really something as laboratorians, we need to keep a close eye on, because there is nothing to say that clinical laboratory tests really has to be done in the hospital lab.

Host: Dr. Gregory Tsongalis is the Director of Molecular Pathology at Dartmouth Hitchcock Medical Center, and he has been our guest in this podcast from *Clinical Chemistry*. I am Bob Barrett. Thanks for listening.

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