Personal Genome Service Providers
Ryan Phelan

MS. AU: We will continue on with the session. Thank you, Dr. Church. I'm sorry we abruptly stopped you.

Our next speaker is Ms. Phelan. I want to thank her for holding her session while the Secretary was able to come visit with us for a while.

Ms. Phelan is the founder and CEO for DNA Direct. She has been a strong consumer health advocate for the past 25 years, having started the first medical library for consumers in 1978. Previously, she was the founding director of Plain Tree, a nonprofit consumer health care organization, and founder of Direct Medical Knowledge, an extensive consumer health website which was acquired by WebMD.

We thank you, Ms. Phelan, for being here.

MS. PHELAN: Thank you. It is an amazing honor to be here today and a pleasure to hear Secretary Leavitt speak as well. Thank you for allowing me to be here today.

I started this company now, DNA Direct, in 2004, and I spoke with many of you here in this room in the year before I actually developed it, trying to think through everything from the ethical issues involved to the clinical and scientific issues to actually the challenges of doing Internet commerce. It is really a pleasure to see over this period of time how much has changed in our industry and really, I believe, how progressive society is in moving in a positive way.

The mission of DNA Direct is clearly to bring the power of personalized medicine to patients, consumers, and providers. As mentioned in my introduction, I have been involved in health care information really for 30 years. I started the first medical library for consumers in 1978. This truly is an extension of a belief and a commitment I have to helping consumers take an active role in their health.

We launched in 2005 by working with a very distinguished and important group of scientific advisors who really looked at how could DNA Direct innovate in this whole field without innovating around the science but innovating around the delivery medium which we were using, which was virtual medical genetic testing, and using standard clinical protocols but Web-enabling them.

That meant that we would have everything done under the medical oversight of our medical director. It meant that with his cooperation we would create very clearly guidelines for our genetic counselors and follow that same procedure from beginning to end, starting with informed consent, all the way through the test facilitation and the interpretation.

DNA Direct now currently works as well with healthcare providers significantly as an extension of their healthcare services.

The physician is at the core of a lot of what we do. We do not always facilitate the test. Much of the time we work with physicians where we are doing purely just the counseling and interpretation services. Of course, DNA Direct is never the lab in any of this. We work with all CLIA labs, depending on the type of test that we are offering.

Our medical genetic testing ranges from everything from standard prenatal testing to carrier testing, to currently drug response testing. I'm sure Herceptin is something that we will be adding to that list with the new FDA approval.

We look for, in everything we do, whether or not this is a test that is going to be clinically actionable. Is there something that a consumer can do with this information. We spent yesterday defining what that really means and how these terms can be used and how they can be confusing. But I think we really all know at the end of the day whether or not something has medical and healthcare significance.

I'm going to just walk you through a few slides very briefly about how our testing works. For any of you who have seen our site, anytime anyone goes through a testing area there is significant pre-test information to really help consumers understand the pros and cons of testing. DNA Direct is clearly committed to making sure people know when a test is actually going to be helpful to them and when it might not.

Our questionnaires actually do two things. One, they determine where testing might be appropriate, but secondly, the information we glean from that regarding personal or family medical history for a condition is used to actually build a personalized Web-enabled report with their test result.

All of our customers' medical charts and questionnaires are reviewed, along with the test result, by a medical geneticist before we release any result to our consumers.

From the patient side, testing is easy and can be facilitated with anonymity with a unique identifier that is sent to a CLIA-certified lab.

All of our tests are provided with clear transparency regarding our pricing. DNA Direct does not market any lab fee. We pass that cost directly on to our customer, then charge for our interpretation and consulting services.

This is just an example of a report for, in this case, a BRCA result that is positive. The test result, as I mentioned, is customized based on their family history. All of our BRCA customers go through an in-depth pre-test phone consult as well with our counselor as well as the online experience. Obviously, we want to identify that the right person is getting the right test and that it is really appropriate, and that phone counseling is going to work for this particular patient.

For many of our customers, one of the things that we are constantly talking about is whether or not they would prefer to see an in-person genetic counselor. We have a network of over 60 genetic counselors that are affiliated with DNA Direct where we can refer somebody if we actually think that a phone consult is not as appropriate as an in-person.

All of our reports include a physician letter that is two- or three pages. We heard that from some of the physicians yesterday. The last thing they want from a patient is a ream of useless information. What they want is really specific, easy-to-read fast information about the clinical guidelines and the significance of the test for that patient.

All of our data is secured from the get-go of the company. We look very clearly at SSL protocols to make sure that we set up the system. Our genetic data regarding a result of a patient is kept totally separate from any Ecommerce transactions. So basically, the shipping, billing, and

personally unique identifiers are kept in one server and another where all genotype and phenotype correlations are.

Just a few minutes about why consumers use our service. First of all, they often come to us because they have had a problem obtaining a genetic test for one reason or another. Sometimes it is purely geography. Everyone in this room knows that there is a dearth of genetic expertise out there, and as consumer awareness increases around this there will continue to be this bottleneck of services.

For some, they referred us to a physician purely because a physician doesn't want to be doing the interpretation. Physicians are not paid in their eight-minute visit to be really taking significant time to do the kind of consulting or counseling around family history and everything else.

Some purely want anonymity. I suspect that may change somewhat with GINA, but the truth is anonymity is not just about genetic discrimination. Sometimes it is about personal privacy.

I thought I would take a second and explain our experience with customers. First of all, 46 percent of our customers have a personal family history for the condition that they are testing. It is a very large number. Eighteen percent have a personal diagnosis with the condition. Twenty-one percent have a known family mutation. A combined 53 percent have both.

A really important take-away here is 34 percent of our customers across the board -- since we started the company it has been anywhere from 35 to 40 percent -- test positive for a mutation.

Now, for those of you who are geneticists in this group, you will probably recognize that in a traditional genetics clinic positive mutation rates probably run much closer to 5 to 10 percent in the general public. By the way, what that means is DNA Direct is testing appropriate people for clinically valid tests.

I would like to end on this one slide, which is something that I showed yesterday. Unfortunately, this Adobe Acrobat Reader is not reading this correctly, but those little square boxes, or rectangles, were meant to be check boxes. What I'm trying to do here, and I'm really trying to help the Committee think through this whole field of genetic testing, [is show] that not all genetic tests are equal.

I started at the bottom of this triangle with serious diagnostic testing, like Huntington's disease. Most of us could agree that that is probably best facilitated in a bricks-and-mortar setting with in-person evaluation by genetic experts and very clear physician oversight.

But as you go up this ladder, you have to go up this ladder and start to think [more broadly] about the implications for genetics. Right up the ladder I say predictive testing for serious health concerns. These may not be 100 percent predictive like Huntington's, but they certainly have very clear clinical indications. BRCA is just one example.

At DNA Direct, we believe, obviously, that this can be done with a genetic consult by phone. It does not have to be done in person. In fact, DNA Direct is under contract with Moffett Cancer Center where we do, for part of their research with black African women, all of their phone consult for pre- and post-test consulting as part of their research trial.

The academic centers already are acknowledging that phone consults can work very well and Web-enabled education is clearly going to be a wave that is moving and integrating into health care.

As we go up this ladder, we look at genetic screening for very common things, carrier testing, risk screening, risk assessment, drug response for pharmacogenetics for Warfarin or Herceptin. That probably doesn't need to be done in a physician's office. If we want to look out of the box at this, we might want to say that a healthcare insurance plan could be looking at how to target particularly select populations and making sure that they could get a genetic test easily facilitated through the Web or through phone as appropriate.

But as we continue go up this ladder to genome-wide arrays and to full genome sequencing, I think that is where we really have to start to think out of the box. Our company doesn't provide those services, but what we do is provide support services for anyone who has had information gleaned from one of these tests who may have a personal or family history where they want to test further.

But what I think is important here as we look at this area is we have to question whether or not there should be a different guideline for people who are engaging in genome-wide arrays or full sequencing for different purposes than clinical genetics.

I think that this is something that we are not just wrestling with here in the U.S. I just came back last week from the United Kingdom's Human Genetics Commission. They too are wrestling with these same issues that every one of you as Committee members are thinking about. They are calling for a voluntary code of practice regarding the delivery of genetic information services.

One of the things they were very clear to say is this is a regulated industry, whether you are in England, Europe, or the U.S. We clearly saw that with the recent California cease and desist letters. There is regulation going on here. The question is, what kinds of further regulation are needed and is there a place, and I believe there is, for creating a best code of practice that I believe industry here, right now, is working very closely together to try to move this field ahead in a really responsible manner.

I appreciate your time. Thank you.