Personal Genome Service Providers
Linda Avey

MS. AU: Our next speaker is co-founder of 23andMe, Linda Avey. Prior to 23andMe, she developed translational research collaborations with academic and pharmaceutical partners for Affymetrix and Perlagen Sciences. She also spent time at Spotfire, helping scientists understand the power of data, visualization, and applied biosystems during the early days of the Human Genome Project.

Welcome, Ms. Avey.

I would like to remind the Committee members that there is information about the companies in Tab 4 of your briefing book.

MS. AVEY: Thank you, everyone. It is really great to be here in person. I think last time I was on the phone. It is always better to be in person. I think we are going to have some problems here with Adobe Acrobat, but we will try to get through this.

As you all heard, especially from David's wonderful presentation -- that was really fantastic -- we are really on the very forefront of a journey that I think a lot of us are going to take together. That is why I'm so encouraged to see all of you here today, willing to really have a conversation with us as we embark on this experiment, really. That is really the way we look at this.

23andMe is really a new way to do what we hope will be a very effective tool in research: the ability to really engage with consumers for the first time in a very large-scale and Web-based way to conduct genetics research.

What we embarked upon with 23andMe last November was to enable individuals to get access to their genomes really for the first time in a very broad way. What we see ourselves as a company doing is providing an interface to that genome. What we like to do is tell people that this is really the early stages. We have been doing these genome studies now just for a very brief time really, when you look at the history of research and the discovery of the DNA molecule, all the way through the great work that Francis and others did in sequencing that wonderful molecule, now being able to give people access to it and give them dynamic tools and ways to really interact with that information we think will really move and change the field of how we are going to conduct research.

What we have done is we have this system now where you can order our service through sending in a saliva sample. We extract the DNA out of that. We work with a CLIA facility. Then we generate this set of about 600,000 data points for you.

As Teri Manolio so beautifully pointed out -- I would love to have that slide where she has all the chromosomes and all the papers -- 2008 really has been a watershed year in GWAS. Having come from Affymetrix and Perlagen and seeing all of these wonderful tools that companies like they and Illumina have created, we are really now see the fruits of the labors of a lot of the researchers who have been doing these studies and have been publishing all these great results.

What we have done is we have created this interface where, especially through what we call our gene journal, our scientists really go through and comb through that literature, read through those papers, and then they come up with a white paper that goes through the criteria that we use of

how do we present this information to consumers who have very little understanding of genetics to begin with.

So we have this transparent way of coming up with what we call established research versus preliminary research. The reason we did this is that we first started out with 14 of what then was the ER category, and we left it at that because we felt like we really wanted to stay with these studies that we thought were going to pass the test of time.

But right after we launched we found everybody, including our scientific advisory board members, one of whom is here, really wanted more information. The minute you get access to this, the next thing you want to know is what more can I learn about myself.

We took that into account, and we decided that we would open up the category a bit more, but we wanted to make sure that we put the caveats around that. There are more studies coming out, and we don't pick these people. The whole thing about learning from your mistakes; I don't know who did this study but it did get published, I think, maybe even in Science. So we just are a reflection of what is going on in the research community, and we give that information out to consumers. We don't judge based on what the phenotype is or necessarily what the category was. We want to be really honest and open about these studies.

The star system that we employ now with our gene journals really gives people, hopefully, the idea that if it only has one star you really have to take it with multiple grains of salt. At one point we were going to have salt shakers and have multiple salt shakers, but you have to run this in front of people and when they see more of something they think that means better. So that didn't work.

So it is really this idea that we are taking all of this information out to consumers and hopefully putting it in the right light, that this is all new and there is a lot more to learn.

Unfortunately, you can't see it, but when you look at one of our gene journals, and we have maybe three genes associated with something, we leave the line open because we think there could be a lot more genes filling in those slots. Obviously, there is still a lot of research that is going on, a lot of it funded by the NIH, and we want to be a reflection of that and say let's continue to fill in these gaps. Let's get a broader picture and a better understanding of the genetics that we are discovering, and then be able to incorporate that eventually, hopefully, into our overall care.

We do think that in discussions, especially like these, that we do need other definitions of things. Would this be considered a genetic test per se. When you are getting your whole genome, do you call that a test or is it just information. If we have 580,000 data points and we only know about maybe 40,000 of those, or whatever the number is, what do you consider those other data points. We think of them as information about you that runs around in your cells and now you just have access to it through your computer, which is a different way of looking at it.

Then, the other idea is what is prognostic. What are all these definitions, and how can we work together as a community to come up with a better regulatory means of reviewing it.

Going back to the slide that Teri showed, now that we have our gene journal filled up with all of those great studies, we try to keep that very current. We have people who go through the literature, as I said, and we now have about 78 of these gene journals. I think about 25 of them at

this point now are of the established research category, and then the bulk of them are what we consider the preliminary research.

As new studies come out on SNPs that might be in this preliminary category and they look like they have moved into the ER, or the established research, category we do make those changes. That could explain why David saw a bit of a shift in his risk profile, because these things do change.

He brought up a very good point. I point at us and I point at the other people that are in this industry together, this very nascent industry, that we really need to do our job to work with other organizations to create standards. There are things that we can do, we think, on assumptions that we make. David noted that in his slides, that we do make different assumptions when we come up with these risk profiles, but we should just come together as an industry and say we are going to just settle on the same assumptions. That is a pretty straightforward thing for us to do.

So we definitely plan to do that. We are going to actually be working with the Personalized Medicine Coalition, Ed Abrams and his group. They will take the charge on this as a neutral body to help organize all the thoughts and the desires of these companies and bring this together and hopefully come up with a set of standards. What they are planning I think is by the end of the year, so you can stay tuned. We are really excited about where that will go.

I had planned to go through one of our gene journals, but you don't have to pay any money if you come to 23andMe and just set up a demo account. We have the ability to show all of the data and all of the tools that we have at your disposal when you sign up as a customer, and we have done this through the family called the Mendels. It is a family of eight people over three generations: grandparents, parents, and then three children.

One of the things we really felt was important from the beginning is allowing people to come together as families and even friends and compare certain things about your genome. We have seen a remarkable uptake in the amount of sharing that people are doing, both at the basic level, which is more just sharing things like your maternal group assignment, your Y chromosome markers, or how much you compare to someone overall holistically across your genome. But it has been phenomenal to see that really going on and how many people are really doing that.

You can look through the slides that we have on data security. That is something we take very seriously for our customers. We just feel like we wouldn't have a successful company if we didn't stress the privacy of the information that we are generating for our customers. That is a very important aspect of what we are doing.

What I want to touch on is something new that we have just introduced. Every month, practically, we have a new release of our software and our website where we have new tools and new features that are coming out. Within the last month or two what we did release is the ability now to do surveys with our customers.

We started out with fairly straightforward and simple ones: are you left- or right-handed. Even with that one we took a survey that has been used and validated by epidemiologists. It has been really fascinating to see this because not only do we say do you write with your right or left hand or do you throw a ball, but it is how do you sweep with a broom, how do you open a jar.

I found out myself that I'm right-handed but with a moderate preference for left-handedness. I would have had no idea that I did that. When you sweep, if you have your right hand on the top

of the broom, or if you open a jar with your left hand, I'm sort of guessing but those are the things. I have talked to other people who ended up being right-handed that they had different answers on those surveys.

So we are trying to take these validated surveys and move them onto the Web. This is something that we are looking out to the research community and working with epidemiologists to develop these and see what kind of results can we get. We think starting out with things like eye color, handedness, can you roll your tongue, simple things like that that will sort of help us prove this model of can we do research in a Web-based format like this. We don't know. It is really a new experiment that we want to embark upon.

So far we are really happy with the number of people who are responding, and it is partly because these surveys appeal to pretty much anyone because we all, or most of us anyway, can write and we sweep, although it shocked me how many people don't even know how to hold a broom.

So it is really, then, the next step that we are excited about. We just had a study funded by the Michael J. Fox Foundation to do a Parkinson's study. This is going to be far more in-depth, obviously. The Parkinson's Institute in Sunnydale has been studying Parkinson's disease for many years. They have developed a lot of their own tools for diagnosis.

They are very interested in looking at how can we first move these validated instruments online but then, beyond that, what can we do to develop some tools that might be using Web 2.0 and also using new technologies that would enable any kind of movement disorders people are having that we could measure over the Web.

This is really out there, but this is the kind of thing that Michael J. Fox really is interested in funding, the things that are beyond what we see in the more typical research paradigm. We have had meetings now with SRI, the Stanford Research Institute, where they are working on the Wii game for measuring people's motor skills. We have met with Qualcom. They are getting very interested in health and how different mobile technologies can be used for understanding, measuring, and monitoring disease.

So we are at the early stages, but we are really quite excited about this study. Since then we have had a lot of people coming forward who are interested in submitting grants and doing work with us.

The whole point is that once we get a number of people in our database and they do show that they are interested in volunteering information about themselves, we think this could be a very interesting mechanism for conducting that type of research.

One of the other things we think is really important when you look at people who do surveys online is they want immediate feedback. That is one of the things we do. When you fill out a survey at 23andMe, you find out how do you stack up to the other people in our database. So it is this instant feedback that we find that people get a lot of satisfaction out of.

So that is an aspect that we always plan to have. You will find out are you of the 12 percent that are right-handed with a moderate preference for left or are you more just all completely right-handed. Those are the types of things we are interested in sharing.

Just to finish, really what this goes back to is the interest in translation. How do we translate all of these wonderful studies that are going on with all of this great information. How do we get

that into the clinic. It does seem to be a bit of a gap that we have in our current system of making this mechanism happen.

The NIH has been great to come up with the CTSA, this new funding mechanism that is forcing major universities to come together with their clinical side and say "You have to work together or you are not going to get the funding at this level anymore."

I think on the consumer side we can help match that and meet that and work together but let consumers be more of an active participant. They get their data and they get access to this information because they want it. Actually, there, I guess, are some studies that [show that] some people, if they are asked as part of a research study, think they should get their data. I think it has been pretty obvious that that is going to be the case going forward.

If that is the interest of consumers, then we want to be here to give them some kind of an access and some sort of an interface to that information. The overall goal is to improve health care and to work together and work with you. We are really excited about continuing this conversation. So, thanks again.

MS. AU: Thank you, Ms. Avey.