

Cluster 5: Implications of Consumer-Initiated Use of Genomic Services
Sylvia Mann Au, M.S.

DR. WISE: Sylvia.

MS. AU: Cluster No. 5. I agree with Kevin; it is about power. But I think in this cluster it is about empowerment. Now there is pressure, isn't there?

We are all trying to sell our clusters. Unless you have been hiding in a cave without wireless access, you have been inundated by direct-to-consumer genetic testing articles in the media. This cluster came about because, of course, the number of personal genomic services marketed directly to the public has increased in the past few years, which is definitely an understatement.

This is a new model that doesn't have direct involvement of a personal health care provider. So we are wondering whether a comprehensive consumer protection strategy may be needed in this type of medical testing.

Our concerns include the relative value of the information provided, of course going back to Cluster No. 2, clinical utility; the level of consumer understanding, which again is the education cluster; the provider community's ability to understand and translate information for patients, again the education cluster; and the potential risk of misuse of information by consumers or third parties, going back to Kevin's cluster. So you can see that this cluster encompasses every other cluster that we have on the list.

Genome service companies offer a vastly different array of services, ranging from risk assessment to recreational testing such as match-making. As of yesterday, in the U.S. you may now test your children for their sports ability. I believe it is \$149. That was in a New York Times article this weekend.

Past SACGHS activities include letters that the Committee wrote to the Secretary in 2002, 2004, and 2006, expressing concerns about the advertising claims made by companies offering these direct-to-consumer genetic services. We also had an information-gathering session at the July 2008 SACGHS meeting to explore what was going on in the landscape of genomic services.

There has been a definite explosion of U.S. and international activities related to this area: research studies, educational resources, workshops. So there are a lot of activities going on right now in this area.

Not surprisingly, because this area is fraught with so many questions, we have the most policy questions of any cluster. For oversight, of course, we wonder whether these genomic tests will be regulated similarly to other complex laboratory tests.

As to our Oversight report that we did in SACGHS, which we will never forget, we wonder if those recommendations will be sufficient to relate to these direct-to-consumer genomic services.

We have concerns, of course, about clinical validity and utility, which again came up in the Oversight report. What are the best formulas for calculating these risks. What are the criteria to determine whether association between a particular genetic marker and a phenotype is strong enough for that marker to be included in the genetic testing and reported out.

Continuing concerns about clinical validity and utility. Should there be standards for formatting the raw data from the whole-genome scans. How will the clinical validity and utility of such tests be assessed and communicated to consumers. When is it that sufficient data will have been produced to change previously recommended risk calculations.

Issues for consumers and healthcare professionals. Are requirements for public education and informed consent needed before testing. What are the appropriate roles and responsibilities of the healthcare providers, consumers, and public health programs in this non-traditional approach to genetic testing. Do personal genome services actually fill some specific healthcare or public health need. Are providers and consumers adequately prepared for the information provided by these services. What are the benefits and potential drawbacks of direct-to-consumer personal genomic services.

How will the healthcare system and providers be affected by the availability of these personal genome services. What is known about consumer interest in personal genome services and consumer understanding of these services. What are the criteria that should be considered in determining the value of the personal genome service. What are the criteria for determining whether previously tested individuals should be contacted to inform them of modified risk, or should we let individuals fend for themselves and contact the companies for follow-up.

Of course, for advertising, what are the criteria that the companies need to follow before offering these services and marketing them.

As Kevin said, we have privacy and discrimination concerns. What are the privacy concerns. Probably not much with the next generation on Facebook.

[Laughter.]

MS. AU: What cautions and benefits do consumers consider when sharing their genomic information with others, such as their family members, social networks, clinicians, employers. Does GINA apply to this type of personal genome service, and are these companies actually covered by GINA.

Then, for disparities, could personal genome services actually exacerbate health disparities? Most of these are paid out of pocket and not covered by insurance right now.

Our possible action steps are, of course, to monitor the outcome of all these federal and non-federal workshops, work activities, and educational activities that are going on. Short-term actions would include a development of a checklist that patients could look at when they are trying to determine whether or not they want to participate in these direct-to-consumer genomic services. The Personalized Medicine Coalition has come up with a basic checklist that has started already.

We could also do a brief report on selected key issues so we don't have to delve into every policy question. Or, we can do that lovely in-depth report that we love to do on every single issue that we can think of under the sun, and work on this for the next 10 years.

DR. WISE: Comments and suggestions? Marc.

DR. WILLIAMS: At the risk of adding to the list --

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[Laughter.]

DR. WILLIAMS: I'm taking the risk.

MS. AU: That's okay. You're not rolling off.

DR. WILLIAMS: That's right. Although, after dinner last night, I might.

[Laughter.]

DR. WILLIAMS: You can take that any way you want.

I was struck a couple of meetings ago when I think it was a representative from the World Privacy Forum spoke to this group about the concerns about not only the information from the testing but the fact that some companies may in fact be using information that the consumer is providing at the time of testing to sell to others. I don't see that represented there. Since one of the focuses of this relates to potential consumer harm, I think we should fold that in somehow.

DR. FOMOUS: We had a more general question about that, but we can make sure that we capture that.

DR. WISE: Other comments or questions?

[No response.]