

Policy Perspectives from Members of the Scientific Community
Q & A
Sharon Kardia, Ph.D.

DR. WILLARD: Thank you, Dr. Kardia, for your forthright comments.

We have time for a few questions from the committee, and then there will be a longer panel discussion involving both Dr. Kardia and our next speaker as well. So everyone will get their shot, but I want to get us back on schedule.

Kevin?

DR. FITZGERALD: Thank you very much, Professor Kardia, for those forthright comments. I just have a quick question. How much, roughly, of your comments do you see is specific to the United States, and how much of this would flow over into some of the other large population studies that are being done around the world in different countries?

DR. KARDIA: Well, that's a difficult question to answer because in other countries they have very different systems. I mean, in the U.K., where they have a very different regulatory system around genetic information, they're not going to have the same kind of issues. We could go piece by piece. It's very specific to each one, depending upon how regulatory decisions are made and what their current standards are.

DR. WILLARD: Joseph?

DR. TELFAIR: Dr. Kardia, thank you again. My question has more to do with the practical applications. You alluded to that throughout your discussion. I'm wondering if you can give some specifics to this. I mean, you concluded that it's premature to mount such a study like this, but throughout your discussion you were alluding to specific ways that the process may begin and how things were done. You sort of painted a broad picture of it, but I'm wondering, given your experience, particularly at Michigan, particularly with the group that you work with, the community-based involvement part of it where there are several principles for how you work with communities and how you work with those groups, and also ways of doing professional education, I'm wondering if in your experience and in your efforts, have there been specific activities you've undertaken or specific efforts you've made that have been successful in getting things done? If you could speak to that, I'd appreciate it.

DR. KARDIA: Sure, sure. I'd be glad to, because this is something I work hard on, and I have been amazed at how disparate the solutions are.

At the community level, my understanding of where to start is really in the relationship of genetics to self, to family, and to humanity. What are people interested in? How am I related to my brother and sister? Very basic concepts, things that make them feel good about understanding that I have genome in every single cell. It's very basic, because when you move to here's a mutation, it causes disease, you have a 25 percent risk, all of a sudden they have no context, no personal context with which to use the information. Now, if the doctor says take this pill, they can do that, but they don't retain their genetic information, right?

Now, health professionals are on the opposite end of the spectrum. They want basically the news they can use. I've given many different grand rounds to doctors on cardiovascular disease. The long QT syndrome, sudden cardiac death is a great way to get people excited about genetics, but

then they say how am I going to use that? So there's this gap. Now I've got information, but how does it meld with my current practices? You can see that the needs are very different, and I think that one of the things that this also makes me aware of is that you can see by that big difference why the public would be suspicious. The public doesn't have the basics. The medical practitioners want to use the information, and there's not a connection in the middle, even, where doctors and patients can really talk about genetics in a common language that would help them build that trust so that genetics information doesn't become a liability but an added value.

DR. WILLARD: Okay, we have Francis first.

DR. COLLINS: Thanks, Sharon, for a very thoughtful presentation. You've covered a lot of territory in terms of topics that are at the interface of genetics and society and public policy that this committee has been wrestling with since their founding. Obviously, you have a great deal of experience in the field of epidemiology, so I think your opinion carries a lot of weight.

Let me challenge you, though, on the notion that if we just sort of put this off for five years, that might be a better solution than starting it now, because I think a number of the areas that you have pointed to as being potential barriers are unlikely to improve without some stimulus, and a project of this sort in many ways could provide a useful stimulus.

Having been in Washington now for a dozen years, I can tell you that agencies and regulatory systems, and even public policy decisions that relate to legislation, like genetic nondiscrimination, rarely act unless they perceive a need, and even then it takes a while. A public project with this kind of visibility would, I suspect, be a very valuable additional impetus for taking action to plug some of the many regulatory and legislative issues that you've touched on, and without this kind of project I suspect they will go slower.

Similarly, you point out the issues of public misunderstanding, of scientific communities not necessarily understanding each other and working together. Would not a project of this sort which, if mounted, would be a very visible national enterprise, I suspect more visible than the Genome Project because it would involve lots and lots of people, just regular people, would that not be a wonderful opportunity to try to achieve some of those educational steps for the public, for the media, for public policymakers, and for the scientific community? Because some of the things you said about the inability to work together were said about the Genome Project in 1988 as a reason why it was never going to work, and it probably would not have brought those communities together had there not been a project to provide the glue.

Furthermore, in terms of how this would stimulate the field, you mentioned the concern that maybe this will basically fund a small group of people who will get very rich on the funding from this, and everybody else will suffer. Again, the model would be to have all the data publicly accessible. So having a data set of this sort I would think would be, just as the genome sequence has been, a real stimulus to a field. You mentioned yourself how nice it would be to have the data.

So let me just challenge you in terms of the timing issue, because, of course, this is a long lead time enterprise. You're not going to get anything out of this project until you've set it up, until you've enrolled a lot of people, until you've started to see a lot of incident cases. If we don't start now, we won't really have much useful information five years from now. If we don't start until five years from now, it will be ten years before we have these kinds of data.

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Are those arguments so compelling in your mind that it's better to wait, as opposed to trying to use this, which I'm obviously proposing, as a way of trying to address some of the things that you're most concerned about? I'd like to hear your thoughts on it.

DR. KARDIA: Sure. I guess at baseline my, I'll call it, opinion that we need more time comes from my human experiences, that researchers not being able to work together because of the disciplinary disconnect, and that's a real issue, as well as turf wars. Right now I can tell you from an epidemiologist's point of view that I get funded for collecting data, not analyzing it. The NIH will cut off the fourth year where all the analysis is to be done as long as the recruitment is done. There is not a lot of appeal for genetic epidemiologists who analyze data because we can't get it funded by our peers. Collecting data is what does it.

I think the other thing is that there have been some inroads in terms of these regulatory agencies. I mean, the FDA is really having to struggle with this, even if it's just in the generic case of the BiDil drug. But where in the plan is the resources for the infrastructure? Why aren't we doing a national genetics education? I don't believe education in the mix of research is the way to do it, because it's at different ends of the spectrum. I mean, what you're trying to accomplish is about the genetics of disease and disorders. Where people need to start is way far away from that in terms of their own personal relationship with genetic information.

So it just seems to me that there needs to be some other things in place, and believe me, I understand. Ten years of working on the Family Blood Pressure Program, we're now just getting to the point where we're getting some exciting results and the ability to do things. But there was the natural pressure within the system to show, just like a corporation, quarterly progress that I think actually dismantled much of what would have been, basically, the advances that we needed to make in our complex understanding of genetics rather than going for the single-gene paradigm. There's a huge amount of force right now to do the single-gene paradigm.

PARTICIPANT: That's crazy.

DR. KARDIA: It is crazy. We're suffering. Science has got fashion in it. The HapMap is fashionable. If you don't put a grant in with the HapMap tag SNPs, you're not going to get a good score. From a human perspective, I think we have a lot to get over with this large population study.

DR. WILLARD: Thank you for that. I'm going to let you catch your breath and I'm going to ask others to hold their questions until we come back to the panel discussion, where everyone will get another crack at you.

(Laughter.)