

I had the honor of working in Dr. Pinn's office for a year, and she had the vision to let me work on genetic issues. That gave me the opportunity to work closely with Francis Collins and Richard Klausner and to take a proactive role in setting the agenda for genetics. I would like to place Dr. Collins' presentation into a broader societal context. These are warnings that may dampen some of our enthusiasm about what might happen in genetics in the next few years.

First, I caution you about "genetic myopia." This is a blindness that occurs when we try to explain everything in terms of a genetic solution, a genetic answer, or a genetic cause. This can be very dangerous because it blinds us to other things that are affecting us. Individuals should not think that their health is so heavily determined by their genetic makeup that changing their behavior will not affect their health. Thinking that we can not change our health status, no matter what we do, does not empower us to improve our health.

Secondly, we all need to be aware that genetic testing is not a "quick fix" in and of itself. It is a means toward an end — better or improved health and reduction of morbidity and mortality. It is not an end, and sometimes the commercial market may lead us to think that is the stopping point.

Third, we want to be careful about not perpetuating a genetic underclass. For example: Those who have access to prenatal care have access to prenatal testing. Those who do not have prenatal care do not have access to prenatal testing. Those who are involved in a research protocol may be able to get testing, but then may not be able to benefit from any of the information. If they do not have insurance for a prophylactic mastectomy, or they do not have funding to get mammograms more often, what good is this information? Will we continue to perpetuate even more divisiveness in our country?

The fourth issue that concerns me, particularly with respect to women, is accountability. If this information is available, do we have a duty, particularly with pregnant women, to get all this genetic information? Over the past 20 years, we have focused mainly on pregnant women. We have had genetic information on pregnant women, and my prediction is this will continue because they are the targeted audience that can be marketed commercially.

Fifth, a social construct is developing that I call "genetic identity." It is the Jewish gene for breast cancer, the African-American gene for sickle cell trait, and the Native-American gene for alcoholism, and so on. The irony is that to conduct good research, particularly in genetics, you may have to focus on certain ethnic groups. Subgroup population analysis is very important to good research, but how are you sure that you do not target and stigmatize a group so much that these people say we have had enough. How do we really do good research that is meaningful to those particular groups and at the same time not injure them?

Within this societal context, the major legal, ethical, and social challenges include: informed consent, discrimination, privacy and confidentiality, and family rights and responsibilities.

Many unanswered questions create challenges for the informed consent process. For example, in predictive genetic testing, the value of the predictive information is unknown. We have very little data on its value in terms of the individual's health. We risk misinterpretation, both from the perspective of the consumer and from the perspective of the provider. We have limited clinical data on the effectiveness of prevention and intervention strategies. The impact of predictive testing on changes in health behavior is also unknown. If, in fact, one has a susceptibility for a particular disease, for example, lung disease, is one more or less likely to smoke?

In addition, it is difficult to quantify and qualify the social risks of genetic testing and research. How much discrimination is there? What are the privacy concerns? How does genetic information change family relationships? Do people experience group stigma? All these social questions are beginning to emerge. We must still face the unforeseeable risks and benefits that are yet to be identified. In the 21st century we must continue to address the complexity of the social, legal, and ethical implications of genetics.