Secretary's Advisory Committee on Genetics, Health, and Society

Comments to the Secretary's Advisory Committee on Genetics, Health, and Society

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Thank you for this opportunity and for your work on these issues.

You have identified 12 important issues; some, as you have noted, are subsets of others. Prioritizing them can only be done in the context of a framework: by what metric should these issues be measured and weighted?

In thinking about this, I realized there are some premises by which we in the genetics community operate:

1. Genetics is significant for more than the gee-wiz factor – it has something to do with human health

2. Success for the science of genetics means translating this body of basic knowledge into technologies and treatments that improve human health

3. The government should be involved in facilitating success

4. The translation of this basic science is different from other basic science translations – whether or not genetic exceptionalism is right or wrong – this field has engendered these discussions which set it apart from other basic science to health translations, i.e. basic biology to Avastin.

Against this backdrop I'd like to comment on the issues – but first a disclosure: I have a huge conflict of interest and an overarching "agenda".

I speak, mindful of the millions of individuals affected by genetic conditions:

- I know what I know because I, as one among them, have worked along side them for almost 10 years.

- I know what I know because my colleagues, other lay advocacy group leaders, face the loss of their child, face the enormous impact of disabilities, face the inadequacies of the health care system

- I know what I know because my two children face blindness and a host of other difficulties as a result of causal mutations in a gene

I live with the issues you have laid out – have discussed them in numerous federal advisory committees, have analyzed them over drinks, have written papers about them – what I think after all the collective work of the community, and after all your work, is that the issues you defined are only symptoms, symptoms of a disease that needs to be described, and we, like so much of medicine, are more comfortable dealing with the phenotype rather than the etiology of the disease. I believe it is the challenge of this committee, if you want to make a difference, to uncover the basic roadblocks, and not continue to describe these 'symptoms'.

In fact, as several people have noted, both here and in written comments, many of the issues are a subset of access – coverage and reimbursement, genetic discrimination, genetics education and training, oversight, Direct to Consumer advertising, patents and public awareness. Many of these issues are examined in a kind of isolation that doesn't reveal the underlying cause. And many of them are examined in a political, agenda-driven, light. I don't think this committee, or any Federal advisory committee, has the resources to recommend solutions to these problems.

Two of the other issues are a step closer to considering the major priority – if the reason we care about genetics is because it will lead to improved health (I already disclosed that I have an 'agenda') – then we must put the symptoms together for a diagnosis. The questions that pharmacogenomics and large population studies raise are related and are closer to the root of the problem. Neither can be done well in the current regulatory climate – both are impeded by important protections that are misguided in implementation – thus thwarting the very research we need to move an enormous body of basic science toward translational research. The steps along the way: meaningful epidemiology, natural history studies, longitudinal studies, environmental studies, gene-environment studies are all thwarted, cumbersome and de-incentivised. This committee has a bully pulpit that can have an impact on policy recommendations that could facilitate the climate necessary for these studies.

The ultimate questions are ones of integration – how will genetics be integrated into medicine, how will scientific evidence be integrated into policy making, payor decision making, agenda setting for research priorities? Right now the system in place has no incentive for physicians to be early adoptors of proven genomics technologies, for payors to pay for new technologies and treatments, for researchers to strive for health outcomes as an endpoint, for industry to take risks that will benefit marginalized communities – be they racial, ethnic or rare disease communities.

So the answer to genetic exceptionalism is an easy one – genetics should be integrated, and the path to integration probably involves both segregation and affirmative action.

The question before this committee is whether you are ready to be bold, to look at these issues without the lens through which you normally look. Are you willing to go beyond the symptoms to understand the etiology of the disease? Are you committed to discovering the real roadblocks in the system that create all these other issues and grapple with the system, not the symptoms?

This leaves you with the issue of a vision statement – an issue which could be considered without substance. I contend that if you cast aside your usual 'imaging tools' and look at the whole 'patient', in the context of community, you have the brain power on this committee to formulate a vision of genetics integrated, of a pathway to translation, of a future where genetics and genomics improve human health. You have the ability to recommend systems whereby politics no longer set the scientific agenda, and basic science no longer holds policy hostage.

I strongly suspect the answer will include universal health care, and while you may feel that this is beyond the scope of this committee, I suggest that to not name the disease increases morbidity.

We, the people who live with genetic conditions everyday, who watch our children die, who care for sick siblings and parents, who are limited by disease ourselves – know well what the rest of the world will come to know – that science will never step up to the plate and set a health outcomes agenda on its own; and that politics will never understand the complexity of the system without the evidence that science offers. It is time for the two to be integrated, to formulate, based on hard evidence, from all the sciences, a vision for the future: identify the roadblocks, recommend the treatments. It is time to engage us in the future for which we hope. We are ready, we hope you are too. Thank you for your service, your thoughtfulness and your dedication to genetics, health and society.

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