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TESTIMONY OF THE GENETICS AND PUBLIC POLICY CENTER BEFORE THE SECRETARY'S ADVISORY COMMITTEE ON GENETICS, HEALTH, AND SOCIETY FEBRUARY 12-13, 2008

The Genetics and Public Policy Center would like to thank the Committee for taking up the important issue of genetic testing oversight. We commend the Committee for working so expeditiously to deliver recommendations to the Secretary and particularly want to acknowledge the incredible work of Sarah Carr and Cathy Fomous.

In this testimony, we want to both highlight key points from our written comments and make some observations from our vantage point as audience members during the Committee's first day of deliberations.

With respect to our written comments, we would first like to clarify our recommendations for proficiency testing (PT). In the draft report, the task force recommended expansion of PT for genetic tests and not the creation of a genetic testing specialty. The framework of specialties is one created by the Centers for Medicare and Medicaid Services (CMS) to implement the Clinical Laboratory Improvement Amendments (CLIA), but in no way is required by the statute. Nor does the creation of a specialty in and of itself ensure that laboratories perform PT; indeed, there are only 83 analytes for which PT is required. Attempting to work within the preexisting framework, prior advisory committees recommended creation of a specialty, and the Center, along with Public Citizen and Genetic Alliance, petitioned CMS to create a genetic testing specialty.

Given CMS's steadfast objection to creation of specialty and the overarching goal of expanding PT, we support the revision to the draft recommendation that now would require that labs performing tests for which a CMS-approved PT program exists must enroll. Implementing this recommendation will require proposed changes to the CLIA regulations, which will of course be subject to public comment. These changes would be quite straightforward. In fact, the Center has drafted a model regulation that would both fulfill the requirements of the report and avoid concerns of genetic exceptionalism expressed in several public comments. We have submitted the model regulation along with this testimony.

Second, we would like to address the recommendation to create a genetic test registry. The draft report included a recommendation for the creation of a voluntary test registry, perhaps as an extension of GeneTests. The Center has carefully reviewed the public comments and we have

submitted our analysis of them to the Committee. The public comments overwhelmingly support a mandatory registry, and the Committee responded by changing this recommendation to a mandatory registry. We applaud the change and strongly support a mandatory registry.

Several commenters urged that the registry be housed and managed by a federal regulatory body. In considering what agency within the Department of Health and Human Services (HHS) should have lead responsibility for the registry, we believe there are three key questions that must be answered.

(1) What functions will be carried out by the agency, e.g., facilitating data submission, managing and analyzing submitted data for quality control purposes, ensuring compliance with data submission requirements?

(2) Does the Secretary have sufficient statutory authority to carry out these functions, including authority to ensure compliance, and will the Secretary need to delegate these functions to different agencies?

(3) Which agency or agencies could best carry out these functions (recognizing that it may be a combination of several entities)?

Several agency homes were suggested in the public comments: the Food and Drug Administration (FDA), CMS, the National Institutes of Health (NIH), and the Centers for Disease Control and Prevention (CDC). All of these agencies have an interest in the data that would be collected. While we do not take a position on which agency would be the best home for the registry, we agree with CMS's own comments that it lacks the capacity to create or operate such a registry. Although the Secretary could build that capacity within CMS, it would be costly and delay implementation.

Thus, we believe that an agency within HHS with experience and expertise in database design, development, and operation would be better equipped to meet the need for publicly-accessible, comprehensive information on genetic tests. FDA maintains a number of easily accessible and useful databases, CDC maintains a registry of all U.S. IVF clinics and extensive annual data, and NIH manages publicly searchable databases that allow timely, easy access to trillions of interlinked pieces of information.

The development of a genetic testing registry will instill public confidence in genetic testing that will be critical in a future where genetic medicine will play an increasingly important role in health care. We are very pleased that this issue has received such serious and thorough consideration by the committee and we are hopeful that the important steps you have recommended to ensure the safety of genetic tests can be quickly and efficiently implemented.

Now, we would like to reflect upon this Committee's first day of deliberations. As yesterday's discussion reflects, getting to a set of recommendations that provide meaningful guidance to the Secretary regarding genetic testing oversight is a grueling, sometimes contentious, process. It requires dedication, tenacity, and perhaps most important, the ability to put aside personal interests in the service of the public, which both believes and expects that the genetic tests they use to make

important life decisions are analytically and clinically valid. Unfortunately right now we cannot provide them this assurance. Joan Q. Public cannot today determine whether the lab that tests her sample is CLIA certified or that being CLIA certified means the lab knows how to get the right answer. Nor can Joan Q. Public today get the information to differentiate between a test for which there are extensive data to support clinical validity and a test where the claims are pulled out of a hat. This is unacceptable. Your recommendations need to ensure adequacy and transparency of the evidence. We need to lift the curtain.

Yesterday's discussion, as you may have surmised from the murmurings in the audience, was troubling in several respects. First, we heard a constant refrain that increased oversight will stifle innovation, in the absence of a shred of evidence that such stifling has ever or will ever occur. Today, manufacturers of IVD test kits are subject to FDA regulation; if they were being stifled one would expect to hear it in their public comments. To the contrary, however, the comments of AdvaMed, Roche, and others argue that more, not less, oversight is needed.

Interestingly, there was no discussion of the deleterious impact of the status quo on innovation. IVD manufacturers face significant disincentives to producing validated assays. The problem of course is that for any given test, a manufacturer may develop the evidence to take the test to FDA and receive approval, only to face competition in the marketplace from a laboratory making identical claims in the absence of any premarket review. This committee will not fulfill its mandate if it does not make recommendations that substantially level the playing field for businesses that can and are innovating in this space while at the same time doing the work required to get FDA approval.

The discussion of the role of FDA also was troubling yesterday. Dr. Gutman eloquently laid out the choice the Committee had to make: Is the public health best served by FDA oversight of laboratory-developed genetic tests, which would ensure that the investigational design is rigorous, that data collected is carefully reviewed by disinterested experts, that claims made about the test's benefits are truthful, and that any adverse events resulting from the test are promptly reported? If the public health is not best served by this model, which FDA currently applies to drugs, biological products, and medical devices, including IVD test kits, what alternative does the Committee want to recommend?

Dr. Gutman thus laid down this gauntlet, but the Committee did not pick it up. Instead, the suggestion was made that FDA be precluded from reviewing all LDTs for an extended period of time. Such a suggestion is unsupportable at a time when a veritable tsunami of genetic tests are emerging from the laboratory and are poised to make their way into the clinic. This Committee should not suggest that FDA be handcuffed pending numerous meetings of interagency committees and the drafting of yet additional reports on the issue.

Finally, we want to comment on yesterday's discussion of direct-to-consumer (DTC) genetic testing, and make five points.

(1) There were a number of inaccuracies in the statements made about the regulatory status of DTC tests. The definition of a clinical laboratory is one that examines samples "derived from the human body" to provide "information for the diagnosis, prevention, or treatment of any disease or

impairments, or" for "the assessment of the health of human beings." This definition and all the CLIA regulations cover labs whether or not the tests are sold DTC.

(2) Based on the above definition, the vast majority of DTC genetic tests are today subject to CLIA because they explicitly or implicitly provide "health assessment." The Center has developed a comprehensive review of current DTC websites and the tests offered, which we have submitted to this Committee. Our review reveals nearly 30 DTC companies offering health or health-related tests. Most of these companies are (or claim to be) CLIA certified. Of course, it is not possible to verify their CLIA status unless you happen to have Judy Yost's phone number. We think this information should be made publicly accessible by CMS. Nevertheless, DTC should not be made the scapegoat here. Failures in oversight apply across the board. Whether a test is offered DTC or through a provider we need to lift the curtain so we can know what data support the claims being made.

(3) Also in light of this definition, the road map that the Lewin Group prepared is incorrect, because it shows a non-CLIA regulatory pathway for DTC genetic tests. With the exception of those DTC tests where it is unclear that the tests are for "health assessment," selling a genetic test without CLIA certification is against the law. We would argue that the road map should not include all the possible ways people can break the law.

(4) Concerns were raised that a DTC company could skirt regulation by claiming its tests were for "research purposes" only. CLIA does provide an exemption for research labs but the exemption is limited to research in which individual test results are NOT returned to subjects. While 23andme, according to their comments to the Committee yesterday, may consider what they are doing to be research, the claims they make for their tests are for prediction of disease risk and/or health assessment, and, by returning them to customers, the laboratory doing the testing is subject to CLIA.

(5) In yesterday's discussion, Federal Trade Commission (FTC) representative Matthew Daynard stated that the interagency collaboration between FTC and FDA on DTC is "working." We frankly don't know what that means. Since the issuance of the consumer alert, which perhaps not coincidentally appeared the same day as GAO released its damning report on DTC genetic tests in a Senate hearing, what progress has been made? FTC has taken no enforcement actions against DTC companies, despite numerous consumer complaints, a class action law suit, and numerous clearly false or misleading statements on DTC websites. Perhaps, the Committee could recommend that the Secretary "check in" on the progress from this collaboration.

In closing, we would ask that at the end of your deliberations you read carefully over your recommendations. Will they, if implemented, afford meaningful protections to the American public that increasingly relies on genetic tests to make profound health care decisions? If you cannot answer yes, then you will have wasted your time. If you can, then you will have done a great service to the American public.

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