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National Institutes of Health
Office of Biotechnology Activities
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

Re: Draft Report to the Secretary of Health and Human Services (HHS), U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of HHS

Dear Dr. Fomous:

The following comments are submitted on behalf of the Personalized Medicine Coalition (PMC). The PMC represents a broad spectrum of academic, industrial, patient, provider, and payer communities that seek to advance the understanding and adoption of personalized medicine concepts and products for the benefit of patients. We thank the Advisory Committee for the opportunity to actively engage in this process, and applaud its work in this arena.

Our members include professional and industry organizations, public policy and consumer advocacy organizations. As a federation of all stakeholders in the field, we strive to guide policy that impacts personalized medicine by developing consensus on issues affecting the community. Because of these efforts, the voice of PMC is unique. Secretary Leavitt has recognized the unique role of PMC in advancing personalized health care, and has used PMC platforms to make important announcements. In March of 2007, he announced his personalized health care initiative during the PMC annual meeting. In September of 2007, he unveiled his vision of the future of personalized health care as well as his catalog of all HHS efforts in personalized health care during our conference entitled *21st Century Medicine: Personalized and Evidence-based*.

We agree with the report's overarching recommendation that new and enhanced collaborations and partnerships between the Federal Government and the private sector are needed.

PMC especially supports creating an interagency coordination committee led by an office of personalized health care at HHS. Such coordination is urgently needed and will hasten the adoption of personalized medicine, which will, in turn, improve the quality of health care while controlling costs.

We congratulate the Advisory Committee for drafting this thoughtful review of important issues regarding the oversight of genetic testing. A few specific comments follow.



System of Genetic Oversight

PMC and member organizations have been actively engaged with FDA, CMS, and the larger HHS community regarding the system that governs the regulation of genetic tests. All agree that federal regulators need to engage the community when considering regulatory paths. By facilitating an open dialog, different stakeholder groups, including regulators, will develop a better understanding of diverse perspectives. *The committee should suggest that FDA convene a workshop to discuss different models of genetic test regulation.* PMC is prepared to be a convening group if such work would be seen as helpful by the community.

Analytical and Clinical Validity and Proficiency Testing

PMC supports a genetic specialty under CLIA. We appreciate the careful analysis outlined in the report and support the recommendation that HHS require proficiency testing for genetic tests regulated under CLIA. Further, we appreciate the care with which the committee has outlined incremental steps for proficiency testing. While we still believe that a genetic specialty under CLIA would be best, we understand that CMS has decided not to have such a specialty.

We support the recommendation that more resources should be invested in developing standards for evaluating clinical validity and utility. HHS is the appropriate agency to oversee those efforts, and we especially endorse public-private partnerships as a means to more efficiently address those evidence deficiencies.

We were pleased that SACGHS recognized that further analysis, deliberation, and consultation are needed to determine the appropriate role for FDA in regulating laboratory developed tests (LDTs). The field needs a clear and predictable regulatory path that protects patients and supports innovative companies and technologies. On line 3919, the report outlines all of the relevant HHS agencies that should have input into the development of a risk-based framework for the regulation of LDTs. *We suggest that you list the stakeholders with as much specificity as possible, including diagnostics companies both publicly traded and privately held, pharmaceutical companies, professional laboratory associations, manufacturers of kits, ASRs, and diagnostic tools, academic medical centers, and consumer groups.*

Evidence Development and Evaluation

Developing evidence of clinical utility is central to the adoption of genetic testing into everyday clinical practice. However, the gold standard of evidence, prospective clinical trials, is expensive and time consuming. Although we support the ideas behind the recommendations outlined in chapter 5 of the draft report, we fear that it does not address the need to develop alternative, yet acceptable levels of evidence. For example, the Centers for Disease Control and Prevention's Evaluation of Genomic Applications in Practice and Prevention (EGAPP), reviews clinical evidence and usually concludes that available evidence is insufficient to recommend that a particular genetic test be part of routine clinical practice. Most find this approach disappointing since more research is invariably called for without reference to the cost of conducting it. *We suggest that the committee outline acceptable alternative evidence levels for consideration regarding decisions about regulation and reimbursement.*



Effective Communication and Decision Support

The PMC actively supports the creation of a national health information network that enables the interoperable exchange of medical information securely among the diverse stakeholders in the healthcare system. Interoperable electronic health records (EHRs) will improve the quality and value of healthcare by facilitating predictive and preventive care and supporting effective approaches to disease management. By recognizing the value of secure, consented clinical outcomes information, HIT will accelerate new personalized medicine breakthroughs into practice, as well as support patient and physician access to new medical technologies and information.

Incorporating genetic information into EHRs is particularly important for improving the quality of health care. While we agree with the committee's statements in lines 5600-5602 that "representations of genetic and genomic test results as scanned images or free text" does not effectively communicate test results, the committee failed to note that standard terminology sets and nomenclatures have not been properly adopted around the attributes of genetic tests such as standard codified names for tests, results, genotypes and phenotypes. Without some common standardized terminology, it will be impossible for EHRs to represent and communicate the results of genetic tests. *We suggest that the committee urge HHS to fund and support activities that promote the adoption of such standards by all stakeholders in the industry.*

Direct to Consumer Genetic Testing

Direct to consumer marketing of genetic tests could pose a great opportunity for or great challenge to personalized medicine. We agree that more regulation might be necessary, although given the international nature of the field and the opportunities to distribute data on the internet, complete regulation may not be possible or desirable. *The committee might suggest that HHS develop a set of voluntary standards for the field. PMC is ready to convene stakeholders who could propose such standards to educate consumers and guide the field.*

Role of Third-party Payers

We agree that while third party payers are not usually considered as having a role in genetic test oversight, they are important stakeholders in the genetic testing arena as they could help develop evidence about the clinical utility of genetic tests. Proof of clinical utility will be required before these important tests are adopted by patients, providers, and payers. In addition, if oversight is to be risk-based, then post-market surveillance of genetic test utility is another important role for third party payers.

Conclusion

Clear, predictable, modest regulation of genetic tests must protect patients and encourage innovation. These issues are tied to others that confront the advancing field of personalized medicine. In the coming year, PMC will engage in conversations about evidence development, evaluation, regulation, and reimbursement. We will share our findings with SACGHS as they become available. Thank you for your consideration of our comments. If you have any questions, please contact me at AMiller@PersonalizedMedicineCoalition.org or 202-589-1770.

Respectfully submitted,

A handwritten signature in cursive script that reads 'Amy M. Miller'.

Amy M. Miller, Ph.D.
Public Policy Director