Statement of the Genetics and Public Policy Center to the
Secretary's Advisory Committee on Genetics Health and Society

March 26, 2007

Good afternoon and thank you for the opportunity to provide comments.

Almost exactly a year ago today the Genetics and Public Policy Center told this committee of our strong concern about genetic testing oversight. Specifically, we highlighted the absence of a specialty area for genetic tests under CLIA to ensure the analytical validity of genetic tests, and the absence of oversight by FDA to ensure the clinical validity of the vast majority of genetic tests.

Sadly, little has changed over the last year and we still have a grossly inadequate system of oversight to ensure genetic test quality.

One thing that has changed is that this committee is focusing on the issue of oversight. We are pleased that you are paying attention. I would like to use these few minutes to review what has occurred with respect to CMS oversight specifically and to respond to some of the inaccurate statements that have been made to you and in other forums by CMS officials. I would also like to address the need to ensure clinical validity of tests and to enhance transparency of test information for health care providers and consumers.

Last June, a CMS official told you that a proposed rule for a genetic testing specialty was on its way, possibly by early 2007. Fewer than three months later, the agency stated that no specialty area would be developed.

CMS has given many explanations for its reversal. CMS officials told you last November that there was "no evidence" that there was a problem.

With respect, I must differ.

On the issue of evidence, the Center fielded a survey of genetic testing laboratory directors to assess laboratory practices. Directors were asked whether their laboratory enrolled in all available formal proficiency-testing programs. Only two-thirds of those surveyed said they participate in all available programs. Directors were further asked whether, in the absence of a formal proficiency-testing program, they performed proficiency testing using some other mechanism. Almost a quarter of respondents said they did not always perform proficiency testing using some other mechanism when no formal program was available.

CMS officials have asserted that a specialty area would not fix this, but this is simply not the case. When Congress passed CLIA in 1988, it was gravely concerned about the failure of laboratories to perform proficiency testing and its consequences for patient health. For this reason Congress directed the Secretary to require laboratories to participate in proficiency testing unless the Secretary determined that an appropriate

proficiency testing program could not reasonably be implemented. Congress stated that analytes could not be exempt by the Secretary simply because proficiency testing programs were not available yet or because it was difficult to determine the best method for performing proficiency testing.

Bluntly stated, CMS is following neither the letter nor the intent of the law. Because of the way in which CMS has implemented CLIA, in order to require proficiency testing, CMS first needs to create a specialty. If there is no specialty, there is no practical means to require labs to enroll in PT programs. So, reducing the equation to its barest essentials: no specialty equals no proficiency-testing requirement.

CMS says the lack of a mandate for proficiency testing has no practical effect since there are so few formal programs. While there are many more tests than proficiency-testing programs, to the extent that formal proficiency testing programs do exist, CMS should require laboratories to enroll. Moreover, it is reasonable to predict that if laboratories were required to enroll in proficiency testing programs, there would be an added incentive for the development of more programs.

With regard to informal proficiency testing, here too a specialty would be beneficial. While it is true that current CLIA regulations require labs to check the accuracy of their testing twice a year, genetic testing laboratories are given no guidance about acceptable means for doing this, and, as our survey shows, many simply do not. A specialty could provide guidance about how labs should carry out informal proficiency testing in the absence of a formal program.

CMS also characterized the Center's survey as finding errors mostly at the pre- and post-analytic phases of testing. This too is inaccurate. Thirty percent of most common errors identified by labs were analytical errors. Moreover, a strong predictor of whether a lab's most common error was analytic was the level of proficiency testing performed by the lab. The take home message from our survey: proficiency testing matters, and many labs are not doing it.

In enacting CLIA, Congress also directed the Secretary to make the results of proficiency testing performance available to the public. CMS has not done this, making it impossible for any external body to assess the quality of laboratories. There must be more transparency on the issue of laboratory quality.

Finally, CMS has asserted that "only a few organizations" want the agency to issue a genetic test specialty. In fact, about 100 individuals and organizations representing industry, laboratories, patient groups, and others have called on CMS to issue a genetic testing specialty. In September we, along with Genetic Alliance and Public Citizen, filed a petition for rulemaking with CMS requesting that a specialty be created. We have received no response to this petition.

Turning to the issue of clinical validity, CMS correctly points out that the CLIA program is not well suited to address this. However, there is a critical need for leadership on this

issue. If personalized medicine is going to deliver on its promise of providing safer, more effective treatments; then the public must have confidence that genetic tests actually provide accurate and useful information.

FDA has taken some initial steps to address clinical validity for a small subset of genetic tests with its IVDMIA draft guidance, but this approach leaves out the vast majority of genetic tests.

Finally, there is a need for translation of genetic information into concrete recommendations to guide physician practices. Last week Dr. Janet Woodcock shared a telling anecdote that reveals how far we are from the promise of personalized medicine. A person walked into a pharmacy with the results of genetic tests for Cyp450 in hand, and asked the pharmacy what dose of his medication he should take. The pharmacist could not advise him. Nor, apparently, could his physician. No doubt the patient went home confused, and much more skeptical about the real benefit of personalized medicine.

In today's session on oversight, I hope you will focus on the following four issues.

1. The need for CMS to implement CLIA appropriately with respect to genetic testing by creating a genetic testing specialty and requiring proficiency testing for all genetic tests.

2. The need for transparency so the public can have confidence that laboratories are performing adequately on proficiency testing and have the expertise to ensure accurate testing.

3. The need for a coherent regulatory framework to ensure that all genetic tests are clinically valid before they are provided to patients.

4. The need for translation of new knowledge about genetics into concrete information so providers can make informed treatment decisions.

Thank you for the opportunity to appear today.