



June 9, 2010

NIH GTR RFI Comments
National Institutes of Health
Office of Science Policy
6705 Rockledge Drive, Room 750
Bethesda, MD 20892

To Whom It May Concern:

I write to voice my concern about replacing the current GeneTests registry with a new one. As a clinical geneticist and also as an officer of the American Board of Medical Genetics (ABMG) which accredits individuals who train in clinical genetics and the clinical genetic laboratory specialties in the United States, I cannot emphasize enough how important it is to keep the information that is currently offered through GeneTests available.

One of the challenges we face when we see patients with known or suspected genetic disorders is to find out if a clinical laboratory test for the condition is available, what labs are offering the tests, what type of test it is, what is the clinical diagnostic yield of a test, and how to arrange for a patient to be tested. We use the information in GeneTests on a daily basis in our clinic. It saves us many hours of finding this information by other means, or not finding it at all. I know Dr. Collins is aware of the critical shortage of clinical geneticists in this country. At my institution we already have a three month waiting time for patients to get in for an appointment; it is even longer at other institutions in my state. Anything that would hinder our ability to provide care to our patients or to further slow this process will be disastrous.

It is very important that NIH acknowledge the difference between a clinical laboratory test ordered by a physician and done by a CLIA-approved lab that participates in CAP surveys, has rigorous quality control and is run by clinical laboratory geneticists who are intimately aware of the implications of their tests and results with companies such as 23andMe that offer direct to consumer testing with little oversight. I fear that this distinction may be overlooked.

We also utilize GeneTests to locate researchers who are studying conditions for which no clinical test is available. This also benefits our patients who may be interested in participating in such studies and advances insight to their conditions. It also benefits researchers. Our Primary Ciliary Dyskinesia Center at the University of North Carolina at Chapel Hill has increased the number of subjects enrolled in their studies after having information about their research posted on the GeneTests website.

Another critical role of the GeneTests website is in providing GeneReviews. As I am currently responsible for developing the next ABMG exam in Clinical Genetics (the “Book Chief”) and was closely involved with the development of the last exam, I have knowledge of the entire pool of questions we use to write the exam. To ensure accuracy and timeliness of a question, we require at least one reference for every question. More than 50% of our questions cite a GeneReview article. The reason for this is that they are comprehensive, timely and accurate. With rapid developments in the field textbooks cannot keep up. There are far too few review articles to cover the topics we need to include on the exam. We already have great difficulty finding people to write exam questions. If we have to throw out half of our test questions because the reference is no longer available, this would also be a disaster to the medical genetics community. Thanks to Dr. Pagon, GeneReviews and its contributors, we have a wonderful resource that we cannot do without and which needs to be maintained.

Sincerely,



Cynthia M. Powell, M.D., FACMG, FAAP
Associate Professor of Pediatrics and Genetics
Chief, Division of Genetics and Metabolism
Director, Medical Genetics Residency Program
The University of North Carolina at Chapel Hill