Public Comment
March 1, 2004

DR. MCCABE: But now it's time for us to hear from the public. This is something that's extremely important for us. We've just been discussing that this is a public forum for deliberations, and we value the input we receive from the public. We set aside a time during each meeting for public commentary. There will be time today and tomorrow. There are also written comments which appear under Tab 1 of your briefing book, and some additional have been passed out today or are in your table folders.

I'll ask each of our public speakers to please limit your comments to five minutes, and today we'll be hearing first from Dr. Margaret Gulley, Chair, Molecular Pathology Committee, College of American Pathologists.

You can sit or stand, whichever you prefer.

DR. GULLEY: This is what the written comments look like, and there's extra copies out in the hall.

Dr. McCabe and members of the committee, good afternoon. My name is Margaret Gulley and I'm Director of Molecular Pathology in the McLendon Clinical Laboratories at University of North Carolina Hospitals. I'm also a faculty member in the Department of Pathology and Laboratory Medicine of the University of North Carolina at Chapel Hill Medical School. Today I'm here as a representative of the College of American Pathologists, or CAP, where I currently serve as Chair of the Molecular Pathology Resource Committee.

The comments expressed by the CAP reflect a set of fundamental principles regarding genetic testing and quality laboratory medicine. The purpose of these comments is to provide the committee information with regard to genetic testing issues as they relate to the priorities under consideration. Specifically, my comments will describe the College's involvement in the area of CPT coding towards reimbursement for genetic technologies, the CAP's position on patent policy impacting genetic tests and our progress on the College's approach to address genetic test oversight utilizing existing regulatory mechanisms that include laboratory accreditation and proficiency testing programs.

So first, on CPT coding on reimbursement, the College has formed a Genetic Testing Work Group focused on creating appropriate code assignments for molecular genetic testing for recommendation to the AMA CPT Editorial Panel. In an effort to better utilize molecular genetic tests, the work group proposed to the AMA the implementation of a coding system that will offer diagnostic granularity without changing test descriptions and thus be less prone to payment denials.

The work group includes representatives from the American Society for Clinical Pathology, the Association for Molecular Pathology, the American College of Medical Genetics, the American Association for Clinical Chemistry, and the American Clinical Laboratory Association. A consensus was reached that use of code modifiers in the current CPT system would result in widespread acceptance from payers and enable providers to submit specific information to adjudicate claims.

The College led a breakout session during the AMA's November CPT advisory committee

meeting to discuss this approach. The session included representatives from the AMA CPT Editorial Panel, the Centers for Medicare and Medicaid Services, the AAHP/HIAA, and Blue Cross/Blue Shield Association. After careful review of prior options, the breakout session facilitators all agreed that the numeric alpha code modifier system is the most viable solution. Further, the work group concluded that implementation of the modifier option would provide an accurate method of reporting and identifying molecular genetic testing, the ability to track utilization of genetic tests to capture diagnostic granularity, and permit data tracking to determine the genes that are most often targeted for testing, the frequency of the tests, as well as the types of genetic tests that are most utilized.

The work group confirmed that CPT modifiers would result in widespread acceptance from payers. The work group's recommendations were presented to the CPT Editorial Panel and subsequently to the CPT advisory committee members for comment, and placed on the CPT Panel's February 2004 meeting agenda. The College cannot report on the outcome of this proposal because the proposed CPT coding changes are confidential and proprietary until they're finalized. Because code assignments and descriptions can change until just before publication, the American Medical Association, which owns the CPT, asks that participating organizations not publicly release detailed coding information until publication of the CPT volume.

What can be stated is that after two years of intensive work to create a numeric alpha modifier system for molecular genetic test coding, a major milestone was reached this past month when the CPT Editorial Panel favorably considered the proposed system. We're hopeful that the proposed changes will be published this October for the 2005 CPT coding book edition.

I will now turn to patent policy impacting genetic technologies. As medical specialists in diagnosis of disease, the College recognizes that genetic testing is an area of growth and change for pathology and for all of medical practice in the decades to come. Pathologists therefore have a keen interest in ensuring that gene patents do not restrict the ability of physicians to provide quality diagnostic services to the patients that they serve. The CAP believes that gene patents pose a serious threat to medical advancement, medical education and patient care.

When patents are granted, subsequent exclusive license agreements and excessive licensing fees prevent researchers, physicians, and laboratories from providing genetic-based diagnostic services. As a consequence, patient access to care is limited, quality is jeopardized, and training of health care providers is restricted.

The field of molecular pathology uses genes and their mutations to predict or diagnose disease. The list of diseases that can now be diagnosed or predicted from gene-based tests is growing rapidly. Physicians and scientists can often easily translate the fundamental information derived from studying the human genome into diagnostic genetic tests and use these tests for patient care. Because information about gene sequences is so fundamental to the understanding of specific diseases, patent holders can gain essential ownership of diseases through patents. Exclusive or restrictive license agreements on gene-based tests have been used to prevent physicians and clinical laboratories from performing these tests as diagnostic medical procedures.

Patients suffer because diagnostic test services are less readily available and affordable. Medical education and clinical research are also threatened. In fact, CAP members have received cease and desist notification letters from patent holders indicating that continued patient testing would be a patent infringement. Examples of diseases where testing has been halted due to physicians receiving such a letter include breast cancer, Canavan's disease, Charcot-Marie-Tooth disease, and Alzheimer's disease.

The recent trend of using patents to monopolize gene-based testing services is a radical departure from historical precedent in clinical laboratories, and it works against the goal of making these procedures widely accessible and affordable to the public.

DR. McCABE: Could you wrap it up fairly soon, please?

DR. GULLEY: Sure.

Especially troubling is the fact that under patent protection, the understanding of the utility of the test, as well as the underlying disease processes, also become proprietary, thereby imposing a profound change in how the profession and the public acquire knowledge about these tests.

In 1996, Congress recognized that medical procedure patents might impede the advancement of medicine, curtail academic access, and place unreasonable limits on the research community, and interfere with medical education and the quality of care provided to patients. As a result, in October of '96 legislation was signed into law, the Frist-Ganske amendment, that permanently precludes the filing of infringement suits against physicians and other medical practitioners for the performance of medical activities that would otherwise violate patents on medical or surgical procedures. A medical activity is broadly defined to include the performance of a medical or surgical procedure on a human body, organ, or cadaver, or on an animal used for research.

However, the act does not explicitly affect enforcement of biotechnology patents or extend to clinical laboratory services. With the advent of new and innovative approaches to gene-based diagnostic testing and the promise of enhanced and expanded diagnostic testing, laboratory services and clinicians should have the same protection from patent infringement as other medical providers and other procedures.

We're facing the unprecedented situation in which a single patent --

DR. McCABE: I'm sorry, but we have your testimony in written form. If you could just move to the conclusion quickly, please.

DR. GULLEY: Okay. We believe that patents set an extraordinary and dangerous precedent, and they affect the availability of diagnostic testing.

The final thing I wanted to touch on was genetic testing oversight. We have presented an approach to the oversight of genetic testing that builds on the existing clinical laboratory improvement amendment, or CLIA, laboratory inspection and accreditation process to provide oversight and approval of genetic testing in lieu of federal regulations.

So we would like to suggest that instead of increasing federal regulations or developing reduplicative federal programs, that we work through the existing programs to improve oversight and federal oversight of genetic testing.

So you have the rest of my comments in the handout, and I would like to conclude with saying that the College of American Pathologists appreciates the opportunity, and we're here to answer questions or work with you as you continue to work on policy issues.

Thank you.

DR. McCABE: Thank you very much.

I think in the interest of time we need to move on to the next presenter. That is Dr. Judith Lewis, who is president of the International Society for Nurses in Genetics, or ISONG, and also a professor in the School of Nursing at Virginia Commonwealth University, and a member of the Secretary's Advisory Committee on Genetic Testing.

Welcome back, Judy.

DR. LEWIS: Thank you, Dr. McCabe. I must say it's really nice to see that some of the work that we did that didn't get finished is also being considered by the current committee, so I thank you for that.

But I'm here this afternoon as the current president of ISONG, the International Society of Nurses in Genetics. Our membership spans six continents and includes nurse clinicians, nurse educators, and nurse researchers. ISONG is a specialty nursing organization dedicated to caring for people's genetic health through excellence in the provision of genetic health care services by fostering the professional and personal growth of nurses in human genetics.

There are over 2.7 million nurses in the United States. Of those, approximately 2.2 million currently are practicing as registered nurses. Approximately 7.3 percent of those, or slightly fewer than 200,000, are advanced practice nurses. Half of these, or about 100,000, are nurse practitioners who are delivering primary health care services. Compared to other primary health care providers, nurse practitioners are more likely to be practicing in sites serving patients who are economically or socially disadvantaged or in medically underserved areas. The average salary for a nurse practitioner in the United States is slightly over $60,000.

Of the 2.2 million practicing nurses, over two-thirds of us work at inpatient hospital settings, with the vast majority of those involved in direct patient care. In the inpatient setting, the nurse is the health care professional who spends the largest amount of time in direct contact with the patient. The nurse is often the health care professional who first notes the dysmorphic features of the newborn, who provides the patient with education about the nature of a newly-diagnosed chronic condition, who answers the patient's questions about the meaning of this illness for themselves and their family members, and who deals with the entire spectrum of the human response to health and illness.

The nursing workforce holds great potential in caring for people's genetic health. ISONG has, in conjunction with the American Nurses Association, developed and promulgated the Scope and Standards of genetics clinical nursing practice. This document, which is currently being revised and expanded, delineates the genetic competencies for nurses practicing at the basic level, as well as enhanced competencies for advanced practice nurses. In addition, the Genetic Credentialing Commission, an affiliate of ISONG, offers the advanced practice nurse in genetics credential to Master's prepared nurses in specialty genetic services, and the genetics clinical nurse credential to the baccalaureate prepared nurse who managed genetic information in a variety of health care settings. These credentials are awarded on the basis of a professional portfolio.

ISONG is committed to working towards ensuring that the nursing workforce is well prepared to serve the patient's and the public's need for genetic information. There are several programs designed to prepare nursing faculty, who may have been educated themselves in the pre-genomic era, with the knowledge and skills that they need to include genetics content in undergraduate and specialty nursing curricula. The National Institute for Nursing Research offers a highly

competitive, intensive fellowship designed for doctorally prepared nurses, doctoral students, and advanced practice nurses. The goal of this summer institute is to prepare nurses to become clinicians and researchers in the area of genetics.

Current programs, while providing a valuable service, do not have the capacity to meet the demand. If we are to continue to prepare an educated workforce, the profession needs resources to enhance its education and outreach efforts.

ISONG is committed to ensuring that all individuals have appropriate access to genetics and genomic health care and has approved a position statement defining the role of the nurse in ensuring access. I have provided you all with copies of this statement that just was published this week. In addition, I have with me a single copy of the Scope and Standards document for your use.

ISONG is eager to work with the Secretary's Committee on Genetics, Health, and Society as you define your priorities and begin your work. We look forward to providing you with information on the genetics nursing workforce, the way genetics nurses practice in the United States and throughout the world, and with knowledge of the resources that will be required to ensure that our over 2 million nurse colleagues have the knowledge and skills they will need to practice effectively.

Thank you.

DR. McCABE: Thank you.

We have time for one or two brief questions, if there are any questions or comments.

(No response.)

DR. McCABE: If not, thank you very much.

Our next speaker is Sharon Terry, president of the Genetic Alliance.

MS. TERRY: My name is Sharon Terry, and I'm president and CEO of the Genetic Alliance. I want to thank you for this opportunity to offer comment and for your work on these issues.

You've 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? I propose articulating the metric, and it is improved human health.

There are some premises by which we in the genetics community operate. First, genetics is significant for more than just the "gee whiz" factor. It has something to do with human health. Second, success for the science of genetics means translating this body of basic knowledge into technologies and treatments that improve human health. Third, the government should be involved in facilitating success. Fourth, genetics has engendered these discussions, which set it apart from other basic science to health translations. So whether genetic exceptionalism is right or wrong, it exists. Against this background, I'd like to comment on these 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 alongside them for over 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. I have discussed them in numerous federal advisory committees, analyzed them over drinks, written papers about them. I think that after the collective work of this community and all of your work, I think that the issues you define are only symptoms, symptoms of a disease that needs to be described, and we, like so much of medicine, are much more comfortable dealing with the phenotype rather than the etiology of the disease. I believe that it is the challenge of this committee, if you want to make a difference, to uncover the basic roadblocks and not continue to just describe the 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 are all subsets of access. Many of these issues are examined in a kind of isolation that does not 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 human health -- I've 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 towards translational research.

The steps along the way, meaningful epidemiology, natural history studies, longitudinal studies, environmental studies, gene/environment studies, are all thwarted, cumbersome, and deincentivized. 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 policymaking, payer decisionmaking, agenda-setting for research priorities? Right now, the system in place has no incentive for physicians to be early adopters of proven genomics technologies, for payers to pay for new technologies and treatments, for researchers to strive for health outcomes as the 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 creates all these other issues and grapple with the system, not just 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 a 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 might include universal health care, and while you may feel this is beyond the scope of the committee, I suggest that not to name the disease increases morbidity. We, the people who live with genetic conditions every day, who watch our children die, who care for our sick siblings and parents, and 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. You must identify the roadblocks and recommend the treatments. It is time for us to engage 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.

DR. McCABE: Thank you.

I think, unfortunately, it's necessary for us to move on.

Our last speaker is Dr. Andrea Ferreira-Gonzalez, who is director of the Molecular Diagnostics Lab and Associate Professor of Pathology, Virginia Commonwealth University.

DR. FERREIRA-GONZALEZ: Dr. McCabe, members of the committee, good afternoon. My name is Andrea Ferreira-Gonzalez. I'm a director of the Molecular Diagnostics Laboratory and Associate Professor of Pathology at Virginia Commonwealth University. I'm currently past chair of the professional relations committee for the Association for Molecular Pathology, and I speak to you today as a representative of AMP.

The purpose of these comments is to provide information to the committee on issues that are affecting the ability of laboratories to provide genetic testing services. There are three major issues that I will address today. First is the inadequacy of the CPT coding and reimbursement for genetic tests. Second is the negative effect of gene patents on molecular diagnostic laboratories. And the last issue is an update on advances in the oversight of genetic testing laboratories.

With regards to the first issue, CPT coding and reimbursement for genetic tests, a widely held view which AMP upholds is that molecular genetic tests will influence all aspects of medical practice in the future as we unravel the variations in the human genome that correlate with disease and disease risk. Molecular genetic testing must be financially viable so that physicians and patients may realize all the diagnostic benefits of our understanding of the human genome.

Currently this is not the case, and several factors prevent the appropriate cost recovery for genetic testing laboratories. These factors are the inadequacy of the CPT coding system available for the billing of molecular genetic tests and the low reimbursement levels set for the CPT codes that are available and currently in use. The Association for Molecular Pathology has been working closely with a Genetic Testing Work Group over the last two years, with the goal of developing and implementing an appropriate billing code system for molecular genetic tests.

The College of American Pathology chairs the work group with members of other associations. The work group has proposed to the AMA implementing a coding system that will provide payers with more specific information about genetic tests performed, and thus be less prone to denials. The genetic working group's recommendations were presented to the CPT Editorial Panel and subsequently given to the CPT Advisory Committee members for comment. The proposal was discussed at the CPT Panel's 2004 meeting, and AMP understands that the recommendations were received favorably.

AMP asks that the SACGHS remains cognizant of the progress in implementing the proposed changes in molecular genetic test coding and the impact on payment for molecular genetic testing in the future.

The second factor affecting the financial viability of molecular genetic laboratories is the current Medicare reimbursement levels set for existing CPT codes. These reimbursement levels, which will be presented later on today, are far less than the cost of performing molecular genetic tests. The reimbursement levels were set more than 10 years ago and were inadequate even then. They do not reflect the current cost of genetic tests, they are technically complex and frequently require highly detailed analysis and interpretation. So AMP asks SACGHS to consider steps that could provide reasonable cost recovery for genetic tests.

The effect of gene patents on genetic testing services. The granting, licensing and enforcement of gene patents is having a broad negative impact on the ability of clinical laboratories to perform genetic tests. While licensing fees may be financially devastating to a molecular genetic laboratory already facing inadequate cost recovery for genetic tests, even more egregious is exclusive licensing or enforcement resulting in a sole provider of a medical service.

AMP strongly holds that a sole provider of a medical service is not in the best interest of the public health. Examples of diseases where testing has been halted due to patent or licensing enforcement includes breast cancer, Alzheimer's disease, Canavan disease, and Charcot-Marie-Tooth disease. The growing trend of using patents to monopolize genetic testing services severely compromises the accessibility and affordability of genetic tests for the public. Particularly troubling is that under patent protection, the growth in understanding of the utility of a genetic test, and even the underlying disease processes, also become proprietary.

Congress already recognized that medical procedure patents impede the advancement of medicine, curtail medical access, places unreasonable limits on the research community, and interferes with medical education and the quality of care provided to patients. As a result, in October 1996, legislation was signed into law that permanently precludes the filing of infringement suits against physicians and other medical practitioners for the performance of medical activities that would otherwise violate patents on medical and surgical procedures. However, the law does not cover biotechnology patents and does not extend to clinical laboratory services.

In 2003, Representative Lynn Rivers, who was not re-elected, introduced legislation that would protect physicians and other providers of clinical laboratory services against enforcements of gene patents and against liability for infringement of patents on genes. The logic of the bill was that genetic test services are part of medical practice and should be widely available to promote optimal patient care, medical training, and medical research.

We anticipate that a new bill will be introduced in Congress to address this concern. AMP

encourages the Secretary's committee to examine the negative impact on medicine of current practices in the patenting and licensing of genetic sequencing and work to eliminate restrictions on the medical use of genetic information.

The last topic I would like to point out is genetic testing oversight. The Association for Molecular Pathology has worked closely with the Food and Drug Administration's in vitro diagnostic division to provide a laboratory perspective on the previous committee's proposal for FDA oversight of laboratory-developed genetic tests. AMP supports the existing clinical laboratory improvement amendment, laboratory inspection and accreditation process to provide oversight and approval of genetic tests in lieu of new federal regulations, and even FDA oversight.

A major concern of the previous committee in the area of oversight was the review of laboratory-developed tests prior to their coming into clinical use. In response to these concerns, the College of American Pathologists has developed and added test validation questions to the molecular pathology checklist that will be used for inspections in the future.

AMP asks the Secretary's committee to review the changes implemented by the College of American Pathology addressing test validation oversight concerns to determine if these changes address the concerns raised by your predecessor.

The laboratory inspection process is only as good as the inspectors performing the reviews of the laboratories. AMP has hosted training sessions for molecular pathology laboratory inspectors at its previous three annual meetings, since AMP members are among the experts in molecular testing. AMP supports the training and use of qualifying inspectors to strengthen the review provided to molecular laboratories during the inspection and accreditation process.

Furthermore, AMP's clinical practice committee is addressing clinical practice issues such as developing a consensus on information to be included in a test report for common genetic disease.

On behalf of AMP, I thank you for the opportunity to speak to you today. AMP remains available to you to assist you with or provide information for your thoughtful deliberations and important work.

Thank you.

DR. McCABE: Thank you.

I think we need to move on because of the pressure of time. We thank all of our members of the public for presenting to us today. We very much appreciate your input.