



Association for Molecular Pathology
Promoting Clinical Practice, Basic Research, and Education in Molecular Pathology

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October 5, 2010

Steven Teutsch, MD, MPH
SACGHS Chair
National Institutes of Health
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

Re: Comments of the Association for Molecular Pathology to the Secretary's Advisory Committee on Genetics, Health and Society, October 5-6, 2010

Dear Dr. Teutsch:

The Association for Molecular Pathology (AMP) commends the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) for continuing their consideration of challenges and promises of whole genome sequencing. AMP recognizes that this is the last public meeting, as the Committee's charter was not renewed. As such, we would like to take the opportunity to also express our gratitude and appreciation for you and your colleagues' great work on exploring complex policy issues emerging from advances in genomics. While we are saddened to lose this valuable public forum and regret that AMP and other stakeholders will not have the opportunity to work with SACGHS on the drafting of a full report on whole genome sequencing, we thank you for your dedication and partnership over the past decade.

As we stated last June, AMP's concerns focus on the clinical applications of whole genome sequencing, and not on the advent or adoption of the technology. The wealth of data revealed by whole genome sequencing creates new practice questions that molecular pathologists will have to address. Sharing data among laboratories will promote faster interpretation and scientific understanding of advances and as such, AMP recommends the creation of a central repository for all sequencing data and corresponding phenotypic information. The submission of clinical and analytical validity information to such a repository would further inform interpretations and the clinical utility of the results.

AMP also views whole genome sequencing to be at times analogous to a fishing expedition and dissimilar to conventional targeted genetic testing. Next generation sequencing can also be used to sequence the entire genome and to perform gene panels for a specific disease. The latter is more in line with the type of testing clinical laboratories have done in the past. However, even with the gene panels, using this new technology will require a significant amount of work from the molecular laboratory professional. Whole genome sequencing will have a significant professional component to the test interpretation and reporting. Understanding the clinical significance of the data generated by these tests will require more cognitive work than usual. The molecular pathologist will be even more instrumental

in reporting results than with targeted genetic testing, and will take on new challenges such as being a gate keeper and deciding which information to report and when to update the interpretation as our understanding advances. AMP believes that many of these issues and challenges will be best addressed through professional practice guidelines developed by thought leaders in the profession.

While molecular pathologists evolve their practices to best implement whole genome sequencing into their clinical laboratories, ordering physicians will also need training and education in genomics to understand and act on the results. AMP believes that medical school curriculum and residency training programs will need to devote more time to applications in genomics and integrating complex genetic testing into the clinic.

As hospitals adopt electronic medical record systems, their health information technology infrastructure will need to be upgraded to handle the large volume of data generated from whole genome sequencing. A major factor in the rate of adoption of this technology into the clinic will be an institution's bioinformatics capabilities. AMP encourages advisory committees, agencies and stakeholders working on health information technology to consider the challenges of whole genome sequence data. As we mentioned in our June comments, AMP has formed a Working Group on Whole Genome Analysis that will address these issues in an ongoing fashion.

Thank you very much for your attention and consideration of our remarks on whole genome sequencing and best wishes as you conclude your work over the next few months.

Sincerely,

A handwritten signature in black ink, appearing to read 'Karen P. Mann', with a long horizontal flourish extending to the right.

Karen P. Mann, MD, PhD
President