

AMP Commends SACGHS Whole Genome Sequencing Focus, Raises Ethical Questions and Forms Working Group on Whole Genome Analysis

WASHINGTON, DC (June 15, 2010) – Today, the Association for Molecular Pathology (AMP) presented public comments to the Secretary’s Advisory Committee on Genetics, Health and Society (SACGHS) meeting focused on whole genome sequencing. In anticipation of advances in sequencing and its incorporation into clinical practice, AMP raised ethical and laboratory practice concerns for the Committee’s consideration.

“In the next five years, the application of whole genome sequencing techniques in the lab and subsequently the clinic is a very real possibility,” said Dr. Karen Mann, President of AMP. “There is a coming shift from using targeted molecular tests to using whole genome sequencing techniques and AMP commends the Committee for focusing its attention on the many challenges to the adoption of whole genome sequencing into clinical care.”

As the technology advances, AMP’s concerns focus on the clinical applications of whole genome sequencing. The advent or adoption of the technology itself is not controversial, but how clinical laboratories apply the technology and physicians utilize the information to inform clinical decision-making can generate many ethical challenges and laboratory practice questions.

AMP believes that the cornerstone of integrating this technology into laboratory practice will be the assessment of its clinical utility. Dr. Mann posed the question, “How can three billion base pairs of sequence and identification of the sequence of ~20,000 genes be coupled to clinical utility?” She added that “it will be difficult for molecular pathologists to associate meaning with the data generated by these tests and define a normal genome.” AMP further believes that determining normal variation will be a major challenge when interpreting sequence results from tumor samples.

In its comments, AMP highlighted the following challenges and encouraged SACGHS to discuss these areas of concern:

- **Clinical utility:** AMP believes an effective approach to establishing clinical utility will depend upon a multi-disciplinary research agenda and there should be a central repository to submit clinical and analytical data of these analyses to further inform the interpretation and clinical utility of results.
- **Bioinformatics:** The vast amounts of data will require investments in bioinformatics technology not only to analyze, manage and store the data, but also to enable secure access to the data in a useful manner. AMP believes measures to standardize the data for entry into interoperable electronic medical records and to simplify the reporting of sequencing results will be required to ensure responsible adoption and implementation of this technology.

- **Duty to report:** AMP encourages the Committee to consider the complicated ethical issues associated with the duty to report all data, update interpretations as new evidence emerges, and the appropriateness of masking data irrelevant to the test prescriber's indication.
- **Reimbursement & Coverage:** AMP believes that reimbursement policies should be modernized to appropriately represent the value of the information obtained from laboratory tests that utilize whole genome analysis (or next-generation sequencing) techniques.
- **DNA patents:** As whole genome sequencing becomes more widely used in the clinical setting, DNA patents on specific sequences may restrict the reporting and interpreting of the full results of such testing. AMP fears laboratories performing whole genome sequencing will face infringement liability or risk incomplete reporting of clinically significant data.

AMP strives to educate health professionals and the public on molecular pathology and applauds the SACGHS for releasing its draft report on Genetics Education and Training of Health Care Professionals, Public Health Providers, and Consumers. AMP intends to submit comments later this month. With the proliferation of whole genome sequencing, AMP anticipates communication gaps among laboratories, physicians, and the patients. Dr. Mann explained, "Communication and education will have to be a top-down process starting with the molecular pathologists. They will be responsible for educating the healthcare providers who will, in-turn, inform and educate the patients."

To address these many issues among its membership, AMP announced today the creation of a Working Group on Whole Genome Analysis. The Working Group will meet regularly and develop position statements and practice guidelines for AMP members and the other health professionals using whole genome sequencing within their laboratories.