

Association for Molecular Pathology

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

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The Association for Molecular Pathology (AMP) is an international medical professional association representing approximately 1,600 physicians, doctoral scientists, and medical technologists who perform laboratory testing based on knowledge derived from molecular biology, genetics, and genomics. Since the beginning of our organization we have dedicated ourselves to the development and implementation of molecular diagnostic testing, which includes genetic testing in all its definitions, in a manner consistent with the highest standards established by the Clinical Laboratory Improvement Act (CLIA), the College of American Pathologists (CAP), the American College of Medical Genetics (ACMG), and the United States Food and Drug Administration (FDA). Our members lead and work at the majority of clinical molecular diagnostic laboratories in the United States as well as in laboratories in many other countries. We are frequently involved in the development of novel molecular tests, and in the validation of laboratory developed or commercial assays.

Thank you for the opportunity to comment on the White Papers in Phase 1 of NHGRI's Long-range Planning project. Our comments are as follows:

White paper 1:

Applying Genomics to Clinical Problems-Diagnostics, Preventive Medicine, Pharmacogenomics

Q1, additional questions:

- How do we integrate of the vast amount of information becoming available through genomic medicine, into the constraint practical environment of clinical practice?
- How do we sort actionable (vetted) information from early results and when should we consider findings at a genomic level ready for implementation, especially in the absence of an understanding of functional or epigenetic effects?

Q2, additional questions:

- It is important to ask how the cost of testing can be kept low, but also to determine how the cost of interpretation and education can be kept within reasonable limits (and what these are). The technology is expected to allow mass data generation, but QA and reliable interpretation of raw data is lagging behind.

Q3, additional questions:

These are excellent questions. What types of studies will be needed and what is the necessary scope to provide answers?

- Should we consider development of an educational track for a medical subspecialty under molecular pathology ("applied genomics", with many interdisciplinary components)?

Q4, additional questions:

- Where will clinicians find support for their questions about newly discovered information? Should this be centralized?

Q6, additional questions:

- How do we plan for clinician education and translation of identified risk factors into actionable information?

Q7, additional questions:

- How do clinicians ensure that information obtained through recreational genomics/DTC testing is analytically and clinically valid?

White paper 2:

Applying Genomics to Clinical Problems – Therapeutics

(IS: No comments: my comments were addressed in the reviews already posted – promote integration of genetic, genomics, functional pathways, research and clinical disciplines to distill the most powerful data)

Q3. additional questions (Simhan Nagan):

- How do we weigh in practical factors such as the high costs associated with conducting genome-wide association studies, and the elaborate IT-infrastructure support needed to analyze the computationally intensive data generated.
- How can processes that streamline ethics committee approvals and obtaining informed consent documents for genome-wide association studies in the global environment of a typical drug trial be established.

White paper 3:

A Vision for the Future of Genomics: Education and Community Engagement

Q3, additional questions:

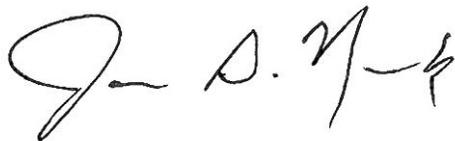
Who is going to provide the infrastructure and resources to educate health care providers. And how are such resources curated?

Q11, additional questions:

How can we best inform the public of the risks associated with freely sharing their genomic information with others (relatives, friends, companies)?

Thank you for the opportunity to comment on this important issue. We look forward to Phase 2; please do not hesitate to contact Iris Schrijver, MD, AMP Clinical Practice Committee Chair at ISchrijver@stanfordmed.org if we can be of further assistance.

Sincerely,



Jan A. Nowak, MD, PhD
President