Future of Pathology in Era of Personalized Medicine

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Personalized Medicine

*Personalized medicine is an emerging practice of medicine that uses an individual's genetic profile to guide decisions made in regard to the prevention, diagnosis, and treatment of disease*

- Precision Medicine Initiative: $215 million investment in the President’s 2016 Budget
- Accelerate biomedical discoveries, provide clinicians with new therapies
- Next generation sequencing – cornerstone of personalized medicine, provides comprehensive analysis of human genome
Precision Medicine and Pathology

• Pathology serves central role in diagnosis and classification of human diseases

• Pathologists provide interpretation of cellular, biochemical, and molecular markers and guide treating physician in the management of patients

• Critically important that pathologists claim ownership of the genome sequencing and lead on genomic data interpretation
Role of Pathologist in NGS-Based Personalized Medicine

1-Develop and expand expertise in NGS testing

2-Ensure that oversight of genome-based laboratory testing falls under the jurisdiction of pathologists and our national organizations

3-Train “next-generation” of pathologists where knowledge of cellular/tissue diagnosis is deeply integrated with knowledge in genomics and informatics
Pathology expertise in NGS testing
Role of Pathologist in NGS testing

- Patient
- Clinician
- Pathologist

Results Reporting
- Pathologist's workstation

Variant Annotation
- Application server
- Data Storage

Pathology
- Tumor diagnosis
- Specimen selection for NGS analysis (FFPE, fresh tissue, FNA)
- % neoplastic cellularity

NGS
- NGS test development and validation
- Assure technical quality of NGS testing
- Oversight of bioinformatics and informatics
- Developing of NGS pipelines
- Data storage, LIS

NGS data interpretation
- Integration of sequencing results with other clinicopathological data
- Standardization of reporting

Bioinformatics
- Results Reporting
- Variant Annotation

Clinician
- Reporting
- Integration
- Communication

Pathologist
- Communication
- Education

Communicating NGS results to treating physician or patient
- Education
NGS oversight by pathology national organizations
Personalized Health Care Committee

• John Pfeifer, MD, PhD, FCAP, Chair
• Coordinate and facilitate communication between different organizations as it relates to personalized health care

Next Generation Sequencing Project Team

• Karl V. Voelkerding, MD, Chair
• Develop checklist requirements for NGS-based diagnostics for CAP/CLIA laboratory accreditation
• Developing NGS PT materials
Clinical Practice Committee, AMP

- Developing of clinical guidelines

- **Guidelines for Interpretation of Somatic Variants (AMP, CAP, ASCO)**
  - Standardization of somatic mutations reporting in oncology specimens

- **Technical Guidelines for NGS Analysis in Oncology (AMP, CAP)**
  - Establish technical parameters for NGS analysis
  - Standardization across different platforms and sequencing approaches
  - Integration with the CAP NGS checklist
Training “next-generation” of pathologists
Molecular Pathology Training

• Access for pathology residents to NGS testing, including technical and analytical parts

• Molecular and Genetics Pathology Fellowship training

• Participating in Molecular Tumor Boards, close interaction with oncologists

• Join Association for Molecular Pathology (AMP)!