Introduction

Next-Generation of Pathology: Role of the Pathologist in NGS-Based Personalized Medicine

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Looking Back—NGS at AMP

• At the 2009 AMP meeting there were 2 NGS abstracts
  – Both in technical topics
• At the 2014 AMP meeting there were 126 abstracts related to NGS
  – In all categories
• 63 fold increase over 5 years
Adoption Rate of NGS in Clinical Labs

• The adoption rate of NGS in clinical labs has been rapid
• The first NGS testing offered in clinical labs was ~2011
• According to CAP there are currently over 100 clinical labs that provide or are implementing NGS-based testing in 2015.
• On a percentage basis the adoption of NGS in the clinical molecular lab is on par with the DVD or iPad
We’re Not Done Growing

The Market Is In a Transitional Stage:
It is reaching maturity in select areas of clinical practice (e.g., prenatal testing), and is approaching widespread adoption

Early Stage
Growing market but focus is on science and research. Lack of infrastructure, clinical evidence, and physician education delay integration of genetic services into clinical care.

Moderate Adoption
Clinical genetic standard of care for select targets and therapeutic areas. Bioinformatics increasingly crucial to care delivery, but remains the bottleneck for interpretation at scale.

High Adoption
Greater availability of data around testing, with genetic services becoming standard of care and genetic testing for a majority of patients across therapeutic areas.

Six Growth Factors

1. Regulatory Landscape
   Evolving oversight and legislation

2. Testing Technology
   Availability of new tests

3. Reimbursement
   Shift toward more genetics coverage

4. Physician Adoption
   Incorporation of tests into clinical care

5. Bioinformatics
   Increasing sophistication of data analytics

6. Consumer Demand
   Consumer interest in genetic services

Source: Booz Allen Hamilton
What’s Next for NGS Technology

• Lower cost of sequencing
• More rapid turn around
• Longer reads will allow for phasing of haplotype information and better SV detection
  – PacBio, 10x, Illumina
• High Sensitivity Sequencing will enable MRD
  – Sensitive and Specific Detection of VAF <0.05%
• Point of care sequencing
  – Oxford Nanopore MinION
Role of the Pathologist in NGS-Based Personalized Medicine

• Implement clinical molecular testing
  – Does the assay ultimately improve patient care?
  – Is the assay cost effective?

• Ensure quality results
  – Pre-analytic review of materials
  – Set reasonable limits of detection for various classes of mutations

• Interpret NGS based data
  – What do detected mutations mean in terms of patient care?
  – How best to communicate complicated data to patient treatment teams?
Learning Objectives

• Understand the technical and clinical applications of next-generation sequencing for the solid tumors, hematological malignancies and cancer predisposition

• Describe the role of the pathologist in facilitating NGS testing

• Be familiar with sample requirements for genomic testing

• Understand challenges in sequencing data interpretation and reporting
INTRODUCTION TO SPEAKERS
Charles E. Hill, MD, PhD

Overview of NGS Testing: From Sample Preparation to Data Analysis

• Director of Molecular Diagnostics Laboratory
• Director of Residency Programs, Emory University Hospital
• Co-Director, Molecular Genetic Pathology Fellowship, Emory University School of Medicine
• Emory University Hospital
John D. Pfeifer, MD, PhD
Applications of Next Generation Sequencing in Solid Tumors - Pathologist Prospective

• Vice chairman for clinical affairs
• Washington University in St. Louis
Maria E. Arcila, MD
Utility of NGS and Comprehensive Genomic Profiling in Hematopathology Practice

• Director, Diagnostic Molecular Pathology Laboratory
• Memorial Sloan Kettering Cancer Center
Colin C. Pritchard MD, PhD
NGS for Cancer Predisposition

• Associate Director, Clinical Molecular Genetics Laboratory
• University of Washington School of Medicine
Format

Please hold questions until the end.