Realizing the Potential of Genomics across the Continuum of Precision Health Care

Karen Kaul, M.D., Ph.D.
Chair, Pathology and Lab Medicine
NorthShore University HealthSystem
Clinical Professor of Pathology
University of Chicago Pritzker School of Medicine

Introduction

- Somatic testing of cancer samples has become critical for diagnosis and treatment decisions over the last decade
- Germline testing is crucial for diagnosis of genetic disorders, assessment of risk, pharmacogenomics and more
- Molecular applications have revolutionized microbiology
- Progress has been enormously beneficial to patient care
- Many challenges and barriers remain

Education

- Education of our providers lags behind field
- Medical school and residency curricula lags practice and potential
- Genomics is advancing rapidly
- Solutions:
  - Training in practice needed
  - Training champions in primary and specialty care
  - EMR tools needed to make genomics part of routine care

EMR

- Genomics is complex!
- EMR needs
  - Structured data
  - Family groupings and connections
  - Reflex testing
- Orders are complex
  - Consent
  - Preauthorization
- Display of results
- Availability of results
  - just in time results
  - once in a lifetime tests
- IT/EMR vendors are building tools….

NorthShore University HealthSystem

Reimbursement

- Need payment for clinical sessions to explain testing and results
- Many issues for lab testing:
  - Reimbursement rates often low
  - Tests often classified as “experimental”
  - Consensus guidelines lag behind actual practice
  - Clinical lab fee schedule (CLFS) used: does not cover professional interpretation

Regulatory Issues

- Increasing availability of FDA-approved kits for molecular microbiology
- Most genetics and cancer testing is done using LDTs (LDPs)
- FDA oversight of LDPs has been debated for decades
- Current concerns over the VALID act
  - Would require ALL lab testing to be reviewed by FDA
  - Cost and access concerns
  - Lab testing is a process, not a kit (though it may use a kit)
  - Redundancy with CLIA
    - CLIA assesses quality of entire testing process
    - Includes proficiency testing with national comparisons