THE BIG PICTURE OF GENOMICS

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CLINICAL GENOMICS
the use of genomic data to inform patient diagnosis & care

• characterizing and diagnosing genetic disease
• categorizing tumors for targeted cancer treatment
• informing likely response to drug metabolism
• identifying pre-symptomatic disease risk
CLINICAL GENOMICS - ALSO KNOWN AS

• molecular diagnostics
• genomically-informed medicine
• personalized medicine
• precision medicine
• data-driven medicine
THE VISION

clinical genomics is a routine analysis tool with high clinical utility

• the likelihood that an intervention will lead to an improved health outcome

• the ability to provide patients and their families a diagnosis, regardless of the outcome
CLINICAL GENOMICS - TYPES OF TESTS

• single gene (exons only or the entire gene)
• panel of common mutations or genes
• exome
• whole genome
CLINICAL GENOMICS - TYPES OF TESTS

69,104 Genetic Testing Units (GTUs) available in the US

59,531 single-gene tests
- 959 exome/genome
- 79 NIPT

8,535 panels

13,676 Categories of comparable tests

on average, 10 new tests enter the market each day
CLINICAL GENOMICS - TYPES OF TESTS

Number of GTUs on the Market

- TP53
- KRAS
- PTEN
- BRAF
- CFTR

Panels | Singles

March 1st, 2017
Concert Genetics
THE COMPONENTS
THE COMPONENTS

identify patients

- scientific landscape
- clinical significance
Which subspecialties are included in your organization’s precision medicine initiative?

- Oncology
- Cardiovascular Disease
- Neurology
- Pediatrics
- Prenatal/Carrier Screening with Genetic Counseling
- Infectious Disease
- Other
- Allergy/Immunology
- Endocrinology/Diabetes
THE COMPONENTS

perform testing

• rapid
• economical
• accurate
• comprehensive
THE COMPONENTS

Cost per Genome

Moore's Law

https://www.genome.gov/images/content/costpergenome_2017.jpg
THE COMPONENTS
interpret variants

• evaluate pathogenicity
  - computational & manual filtering
  - compare existing datasets & review functional annotations

• an integrated, reliable & scalable process
THE COMPONENTS integrate into patient care

- clinical context of molecular results
- impact of variant on risk
- match mutations & therapy
- effectively communicate results
- provider (and patient) knowledge
THE COMPONENTS
inform tomorrow

• influence of secondary findings
• re-interpretation of results
• privacy & data safekeeping
• discrimination & protections
• avoid “one-size-fits all”
THE COMPONENTS

integrate into the system

• electronic health records
  - what to store; how to use & protect
• reimbursement
• regulation and oversight
• ELSI issues
  - consent, autonomy, privacy, equity
What are the prime obstacles to achieving the goals of your precision medicine initiative? (1 = this is not an obstacle at all, 5 = this is a major obstacle)

- Limited reimbursement for Precision Medicine testing: 4
- Clinician education, understanding of genomics: 3
- Information Technology: integrating different interfaces and systems: 3
- Limited time for clinicians to interpret the results: 2
- Providing access to the data for patients to share with other clinicians: 1
- Turnaround time for results: 2.5
- Information Technology: providing reliable results and storing genomic data from testing: 3

Trends in Precision Medicine Adoption: Research Report 2018, Oracle Health Sciences and Genome Web
What am I going to get back?

Genomics in specific disease areas

How does this technology work?

The genomic landscape for personalized medicine
Nancy Cox, Vanderbilt

The future of genomics
Vandana Shashi, Duke

Genomics and patient concerns

Value of genomics

Long-term patient care using genomics

How to bring genomics into your clinic

GM Conference
March 26-28, 2018