National Alliance

Understanding the Spectrum from Genomics to Precision Medicine
A Primer for Benefit Managers

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Speakers

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Genetics in Healthcare: A Primer for Benefits Managers

Confusion

Fear
Basic Categories of Genomics in Medicine
“Curiosity”

“Screening”

“Genomics & Proteomics”

“Targeted Therapies”
First Question to Ask....

“Whose genome are we testing?”

Person or the tumor?
“Curiosity”
“Curiosity”
To my utter shock, the results showed that I have a mutation in a gene called BRCA1, which puts me at a huge risk of developing breast and ovarian cancer. I broke into tears.
“Screening”: Family History

- Breast Cancer (BRCA)
- 291 genes (1%) associated with Cancer
- Developmental Disorders
- Muscles/Skeletal
- Cystic Fibrosis
- “Polygenic Risk Score”
“Screening”: Family History
“Screening”: Family History

Genetic Counseling

https://www.cdc.gov/genomics/gtesting/genetic_counseling.htm
Genomics & Proteomics

- Specific genetic mutations and proteins within a tumor that can predict the aggressiveness of a tumor and affect prognosis
- Directly affect how patients are treated.

- IDH
- 1p/q19
- H3K27
• Majority of tumor genetic testing should be part of routine tumor analysis
• May require “special stains”
• “Personalized Medicine” for Cancer Treatment
Historically, there are three treatments for cancer:

- Surgery
- Radiation Therapy
- Chemotherapy
• Standard chemotherapy affects all cells
• Effective because prevents growth of the most rapidly growing cancers
• Very effective in many types of cancer but does have side effects
• Balance between killing cancer cells and sparing normal cells
“Targeted Therapies”

Chemotherapy

Simponi®
golimumab

IMMUNOTHERAPY VS. CHEMOTHERAPY

TRADITIONAL TREATMENTS

CANCER IMMUNOTHERAPIES

KEYTRUDA
(pemtrolizumab) injection 100mg

Chemotherapy
Healthy Cell

Normal Growth

Cancer Cell

Immunotherapy

“Targeted Therapies”

- “Precision Medicine”
- “Precision Treatment”
- “Targeted Therapies”
- “Pharmacogenetics”
- “Biomarkers”
- “Proteomics”

National Comprehensive Cancer Network®
“Curiosity”

“Screening”

“Genomics & Proteomics”

“Targeted Therapies”
K. Andrew Crighton, MD
Crighton Consulting Group
Potential Benefits

- Improved medical decision-making
- Delivery of appropriate therapies that are tailored to a patient's sequence variants or genotype rather than the general population
- Optimized disease prevention strategies, including lifestyle and behavioral modification, as well as pharmaco-prevention
- Avoidance of medications of lower efficacy
- Reduced exposure to medications that have the potential for greater toxicity, with resulting lower incidence of treatment-related side-effects and complications
Limitations of Widespread Use

- Limitation for clinical use
  - Cost for those at low or no risk
  - Variability in genetic test clinical relevancy between labs
    - Exception is single gene conditions such as Hemophilia B and Cystic Fibrosis
- Lack of clinical expertise and knowledge in interpreting actual risk of results
- Lack of diversity in genomic databases
  - Primarily populated with genetic data of European origin

The Field Continues to Evolve and Provide Great Insight for Clinicians
Barriers to Genetic Testing Implementation

- Limited predictive value of most tests
- Lack of physician knowledge
- Inadequate informatics infrastructure
- Information provenance and patient privacy issues
- Inconsistent standardization and oversight of testing
- Reimbursement issues
- Genetic discrimination
- Societal issues and misconceptions
Takeaways

- Critically evaluate the relevance of generalized testing for your population
- Evaluate proposals that target high risk populations with screenings
- Have candid conversations with medical carrier on process to evaluate and update new biomarkers and genetic tests directing medical treatment
- Ensure genetic counselors are used to evaluate the need for any genetic testing and interpreting results
- How will your plan design handle follow up testing for a probable positive genetic screening test?
  - Will the confirmation test be considered screening or diagnostic?
- Have a trusted medical resource to assist you in evaluating, on-boarding and monitoring personalized medicine offerings for you population (through broker, consultant, medical carrier or second opinion partner)

No such thing as a harmless test
Questions?

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Upcoming Webinars and Meetings

August

- Fiduciary Check In National Webinar
  August 19, 2021, 1 - 1:30 pm
- Mental Health Index National Webinar
  August 20, 2021, 12 pm – 12:30 pm
- COVID-19 Townhall
  August 23, 2021, 4 - 5 pm

All times are Eastern Time

- Registration is open
- Early Bird Rates:
  - Non-Member - $650.00
  - Member Rate - $450.00
- Hotel Discount Rate - $229/per night until October 15, 2021

COVID-19 ANNOUNCEMENT: Based on the current environment and for the health and safety of all our attendees, we will require all attendees to be vaccinated