



Introduction

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What is Personalized Medicine?

Hype, Myth or 21st Century Medicine?

Broad Generic Definition –

- Every medical/health care service provided to an individual patient

Narrow Currently Accepted Definition –

- Based on Current Scientific Development and Concept



Definition

Knowing

- * What works
- * Why it works
- * Who it works for

and

Applying that knowledge for each individual patient.

Goal of Personalized Medicine

Align the work in

- Biomedical science
- Health information technology
- Healthcare delivery

To produce the

- Right treatment
- At the right time
- For each individual patient

Next Generation Sequencing (NGS) Milestones in Oncologic Disorders:

- Nobel prize for the discovery of Double Helix -----1962
- 2001 First draft of the Human Genome published
- 2002 Identification of BRAF mutations in melanomas
- 2004 PIK3CA mutation in Colon Cancer
EGFR mutation in Lung Cancer
EGFR mutants responding to gefitinib
- 2005 Common genetic database for human disease created
JAK2V617F & JAK2 exon 12 identified in MDS
T790M mutation in EGFR linked to TK-inhibitor resistance
ETS-ET4 translocation identified in prostate cancer
- 2006 MPL codon 515 mutation in MPD
ABL mutations identified
KRAS mutation linked to resistance to Cetuximab, anti-EGFR antibody
First Cancer Exome, breast & colon, revealed
- 2007 First GWAS for cancer done
EML4-ALK translocation in NSCLC
cMet & Her3 confer resistance to EGFR inhibitors
- 2008 Creation of non-profit organization to facilitate the translation of genomic discoveries
1st cancer whole genome published, AML
Mechanism of Cetuximab resistance identified
- 2009 IDH1 mutations in gliomas, brain tumor

Next Generation Sequencing (NGS) Milestones in Oncologic Disorders (cont)

- 2010 Targeted NGS used for detecting inherited breast & ovarian cancers
1st comprehensive genetic maps for lung cancer & melanoma published
- 2011 Vemurafenib FDA Approval to target V600E mutation of BRAF
Ovarian cancer subtyping by molecular signatures
- 2012 NGS used for comprehensive molecular characterization of colon & rectal cancer
NGS used demonstrating mutational landscape of breast cancer
NGS using liquid biopsy approach for identification & monitoring of cancer mutations
- 2013 Genetic landscape of kidney cancer is mapped
Angelina Jolie Newsbreak: s/p Double mastectomy for hereditary BRCA₁ (+) breast ca.
CALR exon 9 mutation in Myeloproliferative Neoplasms
Targeted NGS to map 100 network associated with MDS
- 2014 Retrospective analysis of phase II ovarian cancer trial reveals BRCA mutation sensitize to Olaparib, a PARP inhibitor
Comprehensive molecular profiling of 230 resected lung adenocarcinoma (KRAS, EGFR, NF1)
Olaparib approved in USA/EU for ovarian cancer with germline BRCA mutation
- 2015 NGS for Liquid biopsy approach identified new EGFR mutation, C797S, mediates EGFR-inhibitor resistance
Targeted NGS analysis of 113-gene panel identified mutational profile of Prostate cancer response to Olaparib

Personalized Medicine by the Numbers

13 prominent examples of personalized medicine drugs, treatments and diagnostics products available in 2006

72 prominent examples of personalized medicine drugs, treatments and diagnostics products available in 2011

\$300,000,000 cost of sequencing a human genome in 2001

\$5,000 cost of sequencing a human genome in 2011

4% U.S. hospitals with fully operational electronic health records in 2008

50% U.S. population that had medical information recorded in electronic health records in some form in 2010

30% of all treatments in late clinical development rely on biomarker data

50% of all treatments in early clinical development rely on biomarker data

60% of all treatments in preclinical development rely on biomarker data

30% of all biopharmaceutical companies surveyed require all compounds in development to have a biomarker

50% of all clinical trials collect DNA from patients to aid in biomarker development

75% increase in personalized medicine investment by industry over the last 5 years

34% reduction in chemotherapy use would occur if women with breast cancer receive a genetic test prior to treatment

17,000 strokes could be prevented each year if a genetic test is used to properly dose the blood thinner warfarin


\$604,000,000 annual cost savings to the health care system if patients with metastatic colorectal cancer receive a genetic test for the *KRAS* gene prior to treatment

20 members of the Personalized Medicine Coalition in 2004

200 members of the Personalized Medicine Coalition in 2011

Some Interested Parties in the PHC Space

- Payers
- Reference laboratories (Large and Niche)
- Test Manufacturers: „On demand“ rapid point-of-care tests
- Non-Pathologist MDs (and other healthcare providers)
- Investment community: Boutique (high volume) Laboratories
- Genetic testing benefits management companies
- Consumer-oriented Testing Services
 - Direct
 - Via Physician/healthcare professional intermediary
- Advisory/Educational services
- New payment models: Accountable Care Organizations (ACOs)



There are many issues to be addressed to achieve the goal of “Personalized Medicine”!



Thank You