Reforming Medical Informatics in Germany - Basis for Precision Medicine

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Chair of Medical Informatics
Friedrich-Alexander-Universität Erlangen-Nürnberg
and past board member of the GMDS

11. Oktober 2016
Agenda

• Precision Medicine

• Data Sources for Precision Medicine

• Exemplary Applications

• Challenges for Medical Informatics
  ⇒ Biomedical Informatics
  (Medical Data Science)
http://healthitanalytics.com/features/how-precision-medicine-will-shift-from-research-to-clinical-care
Launched by President Barack Obama, January 20, 2015

Images as a courtesy of Jyothisman Pathak (Weill Cornell Medicine, Healthcare Policy & Research)
The U.S. Precision Medicine Initiative

A research cohort that will engage a million or more Americans who volunteer to contribute their health data and biospecimen over many years to improve health outcomes, fuel the development of new treatments for disease, and catalyze a new era of data-based and more precise preventive care and medical treatment.

Precision Medicine

The path from big data to precision medicine

‘precision medicine’ is promising to provide

‘the right patient

with the right dose

of the right drug,

at the right time’.

The path from big data to precision medicine

Bevan E Huang\textsuperscript{a}, Widya Mulyasasmita\textsuperscript{b} and Gunaretnam Rajagopal\textsuperscript{c}

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\textbf{ABSTRACT}

Precision medicine aims to combine comprehensive data collected over time about an individual's genetics, environment, and lifestyle, to advance disease understanding and interception, aid drug discovery, and ensure delivery of appropriate therapies. Considerable public and private resources have been deployed to harness the potential value of big data derived from electronic health records, 'omics technologies, imaging, and mobile health in advancing these goals. While both technical and sociopolitical challenges in implementation remain, we believe that consolidating these data into comprehensive and coherent bodies will aid in transforming healthcare. Overcoming these challenges will see the effective, efficient, and secure use of big data disrupt the practice of medicine. It will have significant implications for drug discovery and development as well as in the provision, utilization and economics of health care delivery going forward; ultimately, it will enhance the quality of care for the benefit of patients.

“… ‘the tailoring of medical treatment to the individual characteristics, needs, and preferences of a patient during all stages of care, including

- prevention,
- diagnosis,
- treatment, and
- follow-up’.

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**Big data**, as it applies to **precision medicine**, is the generation and repository of large amounts of data from

- bio-specimens,
- health records,
- medical imaging and
- sensors,

from which disease-specific factors, patterns, and associations can be computationally identified and used to customize medical treatments unique to the individual
Data Sources for Precision Medicine

Figure 1. In the future, precision medicine will be enabled by individuals surrounded by a cloud of data. Layers of the cloud represent data varying between those directly affecting/quantifying the individual, and those quantifying the environment and indirectly affecting the individual, i.e. levels of the exposome.

Published in: Bevan E Huang; Widya Mulyasasmita; Gunaretnam Rajagopal; *Expert Review of Precision Medicine and Drug Development* 2016, 1, 129-143.
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Data Sources for Precision Medicine

Bioinformatics
- Transcriptome
- Proteome
- Metabolome
- Sensor-based Wearables
  - Activity
- Clinical Informatics
- Exposome Informatics
  - Phenome
    - Dx, Rx, Hx,
  - Interactome

Imaging Informatics
- Imaging
- Microbiome

Medical Informatics

Published in: Bevan E Huang; Widya Mulyasasmita; Gunaretnam Rajagopal; Expert Review of Precision Medicine and Drug Development 2016, 1, 129-143.
DOI: 10.1080/23808993.2016.1157686
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Exemplary applications

for research

and clinical care
Clinical Care and Translational Research

two (or three ?) separate worlds

The clinician

Communication Server

EHR

DWH

PDV

LIS

RIS

PAS

PDMS

The clinical researcher

Patient Recruitment

Clinical Trial documentation

Pharmacovigilance

The basic researcher

Feasibility Phase / Cohorte Identification

Study Protocol

Registration in Trial Registry

Patient Recruitment

Clinical Trial documentation

Pharmacovigilance

The clinician

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Pharmacovigilance
Challenge: Reuse Clinical Care Data for Translational Research

Medical Informatics & Clin. Research Informatics

Single Source Concept

The clinician

Communication Server

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Medical Informatics & Clin. Research Informatics

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Clinical Trial documentation

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The basic researcher

Clinical Trial documentation

Pharmacovigilance
Biomedical Informatics

*Instrumenting the health care enterprise for discovery research in the genomic era*

Shawn Murphy, Susanne Churchill, Lynn Bry, et al.

*Genome Res.* published online July 14, 2009
Access the most recent version at doi:10.1101/gr.094615.109

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Effective knowledge management in translational medicine

Sándor Szalma¹, Venkata Koka¹, Tatiana Khasanova², Eric D Peraksis³

Translational research platforms integrating clinical and omics data: a review of publicly available solutions

Vincent Canuel*, Bastien Rance*, Paul Avillach, Patrice Degoulet and Anita Burgun

Submitted: 11th November 2013; Received (in revised form): 3rd February 2014

Briefings in Bioinformatics Advance Access published March 7, 2014
http://www.briefings-in-bioinformatics.com/content/9/1/1006

doi:10.1093/bib/bbu006
Discovery Research in the Genomic Era
An exemplary platform

tranSMART: An Open Source and Community-Driven Informatics and Data Sharing Platform for Clinical and Translational Research

Brian D. Athey, PhD¹; Michael Braxenthaler, PhD²; Magali Haas, MD, PhD³; Yike Guo, PhD⁴

¹University of Michigan, Ann Arbor, MI; ²Pistoia Alliance, Wilmington, DE; ³One Mind for Research, Rutherford, CA; ⁴Imperial College London, London, UK

http://transmartfoundation.org/


Discovery Research at Erlangen University

from three separate worlds
to an ecosystem which integrates phenotype data and omics data


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Discovery Research at Erlangen University

from three separate worlds to an ecosystem which integrates phenotype data and omics data


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Integrating phenotype data and omics data for discovery research

Let me „play“ with my data

„This is a great tool to support translational research…“
Linkage to external decision support services

- Pharmacogenomic rule sets
- Decision support for doctors
SMART Genomic Apps for Clinical Decision Support


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Interoperability Standards for Precision Medicine (HL7 FHIR)

How Precision Medicine Will Shift from Research to Clinical Care

Precision medicine is an exciting area of innovation, but there's a long way to go before research meets quality clinical care.

FHIR App Provides Precision Medicine Support at Point of Care

By Jennifer Bresnick on August 08, 2016

FHIR is helping to power a new precision medicine oncology app that brings clinical decision support to the point of care.

Two of the most intriguing trends in healthcare may be able to work together to bring advanced clinical decision support directly to the point of care, suggest researchers who developed a FHIR-based precision medicine application that integrates with electronic health records.

The prototype application, described in a study published in JAMIA, is aimed at oncologists who are increasingly relying on genomic information to diagnose and treat complex patients. Using the Substitutable Medical Applications and Reusable Technology (SMART) and FHIR data standards from HL7, the app presents visualizations of genomic variants to help providers target personalized care to patients.

“The definition of cancer and the care of cancer patients are increasingly being driven by tumor genomics, aka molecular profiling,” writes a team of authors from Vanderbilt University, MIT, Boston Children’s Hospital, the University of Science and Technology in Hefei, China, and Harvard Medical School.

Defining digital medicine

Eric Elenko, Lindsay Underwood & Daphne Zohar

Digital medicine is poised to transform biomedical research, clinical practice and the commercial sector. Here we introduce a monthly column from R&D/venture creation firm PureTech tracking digital medicine's emergence.

From Medical Informatics to Biomedical Informatics

Defining digital medicine

Eric Elenko, Lindsay Underwood & Daphne Zohar

Digital medicine is poised to transform biomedical research, clinical practice and the commercial sector. Here we introduce a monthly column from R&Dventure creation firm PureTech tracking digital medicine’s emergence.


Original Articles

The New Role of Biomedical Informatics in the Age of Digital Medicine

Fernando J. Martin-Sanchez; Guillermo H. Lopez-Campos

1Department of Healthcare Policy and Research Division of Health Informatics, Weill Cornell Medicine, New York, NY, USA;
2Health and Biomedical Informatics Centre, The University of Melbourne, Melbourne, Australia

Martin-Sanchez eet al. Methods Inf Med 2016; 55:
From Medical Informatics to Biomedical Informatics

- Exposome/Expotype
  - Environmental risk factors (pollution, radiation, toxic agents, ...)

- Phenome/Phenotype
  - Anatomy, Physiological, biochemical parameters (cholesterol, temperature, glucose, heart rate...)

- Genome/Genotype
  - Biomarkers (DNA sequence, Epigenetics)

Social media / Personal Health Record / Clinical Health Records

Martin-Sanchez et al. Methods Inf Med 2016; 55:
… the dominant trait among data scientists is an intense curiosity
a desire to go beneath the surface of a problem,
find the questions at its heart, and

distill them into a very clear set of hypotheses that can be tested

AMIA Board white paper: definition of biomedical informatics and specification of core competencies for graduate education in the discipline

Casimir A Kulikowski, Edward H Shortliffe, Lyle Barr, Lawrence E Hunter, Todd R Johnson, Ira J Jacobson, Mark A Musen, Judy G Ozbolt, Jack W Smith, Jeffrey J Williamson
Establishing Data Integration Centres...

The role of the biomedical informatician in a translational medicine team

... requires bringing many people from different disciplines to talk with each other

Sarkar IN. Biomedical informatics and translational medicine. J Transl Med. 2010 Feb 26;8:22
The challenge ahead . . .

Clinical Informatics
Health Informatics
Dental Informatics
Nursing Informatics
Public Health Informatics
Bioinformatics
Consumer Health Informatics
Clinical Research Informatics

Medical Informatics

⇒ Biomedical Informatics (Medical Data Science)

Thank you very much!

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