Biomedical and Health Informatics: Impact, Challenges, and Opportunities

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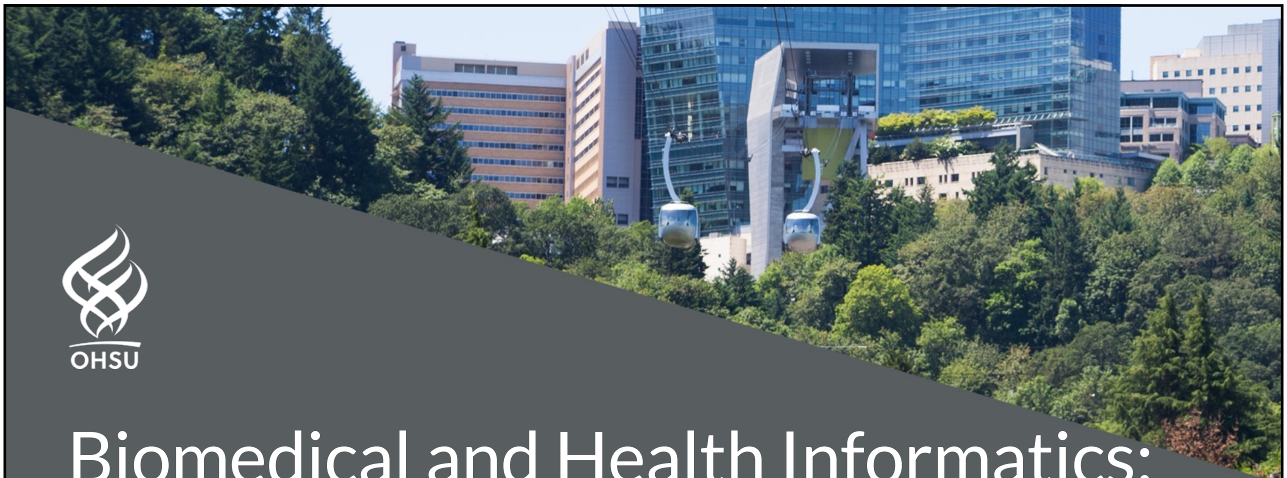

Biomedical and Health Informatics: Impact, Challenges, and Opportunities

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This talk aims to answer the following about biomedical and health informatics (BMHI)

- What is BMHI?
- What has BMHI accomplished? Where has it fallen short?
- How does BMHI help personalized/precision medicine?
- Is BMHI different from data science?
- Who does BMHI?
- Where can I learn more about BMHI?

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What is BMHI?

- I get asked this so often that I keep a Web site
  - http://informatics.health/
- And a blog
  - http://informaticsprofessor.blogspot.com
- Biomedical and health informatics (BMHI) is the field concerned with the optimal use of information, often aided by technology, to improve individual health, healthcare, public health, and biomedical research (Hersh, 2009)

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Other definitions

- AMIA: The "interdisciplinary field that studies and pursues the effective uses of biomedical data, information, and knowledge for scientific inquiry, problem solving and decision making, motivated by efforts to improve human health." (Kulikowski, 2012)
- ACGME (clinical informatics): The field that "transforms health care by analyzing, designing, implementing, and evaluating information and communication systems to improve patient care, enhance access to care, advance individual and population health outcomes, and strengthen the clinician-patient relationship." (https://www.acgme.org/globalassets/PFAssets/ProgramRequirements/381_ClinicalInformatics_2020.pdf)

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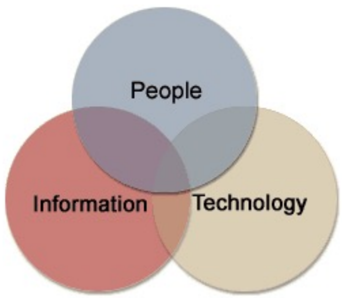
Some additional perspectives on informatics



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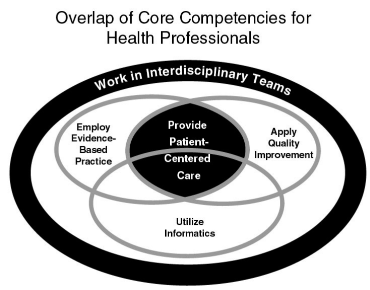
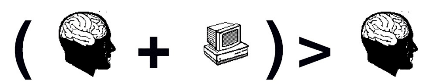


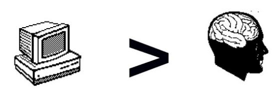
(Greiner, 2003)

Fundamental Theorem (Friedman, 2009)

Goal of informatics is:



Goal is not:



- The science of "sociotechnical systems" (Coiera, 2007)
- What informatics is and is not (Friedman, 2013)

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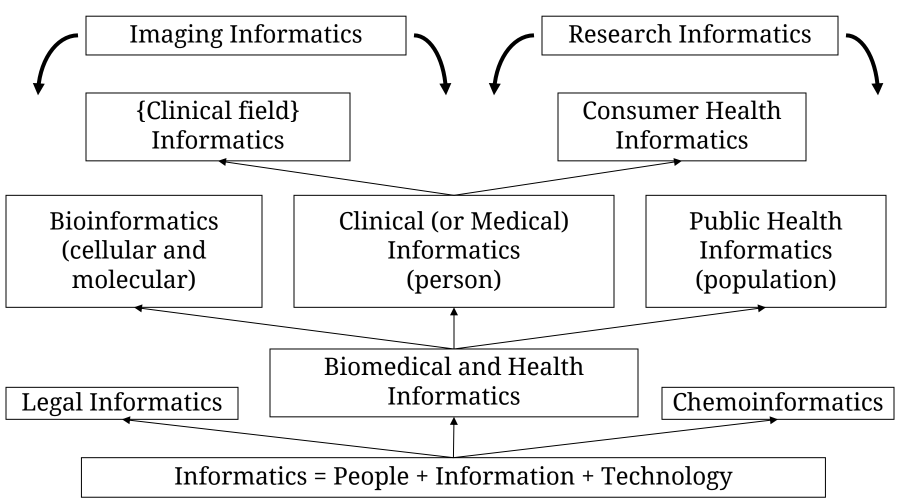
What are the subareas within BMHI? (aka, its "adjective problem")



(Hersh, 2009; adapted from Shortliffe)

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Early work focused on AI

- Focus on hand-crafted "knowledge bases" with algorithms to provide "artificial intelligence"
- Warner (1961) developed mathematical model for diagnosing congenital heart disease
  - System predicted diagnosis with the highest conditional probability given a set of symptoms
- Problem-knowledge couplers aimed to connect patient findings and diagnoses (Weed, 1969)
- Next was emergence of "expert systems" – computer programs mimicking human expertise
  - Early work focused on rule-based expert systems – PhD dissertation of Shortliffe (1975) and subsequent work (Clancey, 1984)

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First era of AI (cont.)

- Another early AI approach developed systems using scoring algorithms
  - INTERNIST-1 (Miller, 1982) and DxPlain (Barnett, 1987) used disease profiles and scoring
- "Demise of the Greek Oracle" led to focus on decision support systems – mimicking human expertise but acting in supportive rather than independent role (Miller, 1990)
  - Led to more focused clinical decision support in 1990s (Greenes, 2014)

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Also focused on early EHR systems

- COSTAR – Massachusetts General Hospital (Barnett, 1979)
  - Built using MUMPS (Greenes, 1969)
- HELP – Utah (Kuperman, 1991)
- TMR – Duke (Stead, 1988)
- Regenstrief – Indiana (McDonald, 1999)
  - Led to development of Gopher (Duke, 2014)
- El Camino – California (Carter, 1987)
- VistA and CPRS – Veteran's Administration (Brown, 2003)

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And early role of the National Library of Medicine (NLM)

- 30-year leadership of Donald Lindberg, MD (Miller, 2021), with torch passed to Patricia Brennan, PhD, RN (Brennan, 2016) in 2016
- Early application was information retrieval from bibliographic databases
  - From Index Medicus to time-sharing systems (e.g., ELHILL; Lindberg, 1986) to PCs (e.g., Grateful Med; Lindberg, 1996) to Web (PubMed)
  - Subsequent connection to full text of scientific literature and other knowledge resources
- Leader in terminology development and standardization (Humphreys, 1998)
- Also funder of primary research as well as training grants and other education

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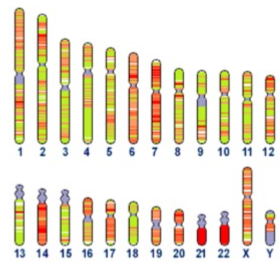
Genomics and bioinformatics

- Human Genome Project to sequence human genome began in 1988
- In 2001, NIH-based project published “first draft” (Lander, 2001) simultaneously with private effort from Craig Venter of Celera Genomics (Venter, 2001)
- Project “completed” in 2003 (Collins, 2003)
- Sequencing of more humans increased understanding of genomic variation and complexity



Continued advances in bioinformatics

- Next-generation sequencing technologies and rapidly lowering costs (Goodwin, 2016)
- Other biomolecular technologies (Lesk, 2017)
  - Gene expression
  - Protein structure and function
- Elucidation of other “omes and omics”
  - Proteomics – protein structure and function
  - Transcriptomics – expression of DNA
  - Microbiome – microorganisms
  - Mapping phenotype to genotype – full circle to clinical data to the phenome
- Many data resources from NLM National Center for Biotechnology Information (NCBI) (Sayers, 2022) and others (Rigden, 2022)
- 20-year retrospectives noted accomplishments but also challenges still ahead in science and policy (Gates, 2021; Jones, 2021)

From home-grown to commercial EHRs: ARRA and the HITECH Act

- By mid-2000s, emergence of research supporting value of EHR and CDS for improving quality and safety of healthcare
  - Mentioned in George W. Bush State of Union 2004-2007
- Great Recession of 2008 led to American Recovery & Reinvestment Act (ARRA), which included HITECH Act that allocated $30+B for
  - Incentives for adoption and "meaningful use" of EHR ($30B)
  - $2B investment in health information exchange, regional extension centers, workforce development, and research (Blumenthal, 2011; Blumenthal, 2011; Washington, 2017)
- Maturation of commercial EHR marketplace (Halamka, 2017; Hersh, 2017)

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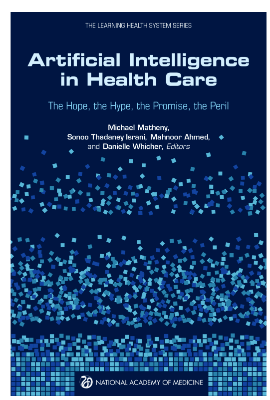
Second era of AI

- Reinvigorated with success of machine learning, driven by increasing availability of data, more powerful computers, and advances in machine and deep learning (Topol, 2019; Rajpurkar, 2022)
- Drawing attention from leading policy bodies, such as National Academy of Medicine (Matheny, 2019)
- Recent system review of clinical trials of predictive AI systems shows modest number of trials, mediocre methodologies, and mixed results (Zhou, 2021)



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Contributions to personalized/precision medicine

- (Denny, 2019; Collins, 2021)
- Approach to diagnosis and treatment of disease based on more precise understanding of genome and other omes along with clinical and personal data
- Contributions of BMHI include
  - Advances in bioinformatics – genotyping
  - Growing amounts of other data from EHRs to personally collected data – phenotyping
  - Developments in machine and deep learning
  - Access to knowledge in literature and other sources

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But there are challenges to personalized/precision medicine as well

- Challenges with large-scale gene sequencing – results impacted by
  - Inaccuracy (Goldfeder, 2016) and DNA damage (Chen, 2017)
  - Gene name errors widespread in literature (Ziemann, 2016)
  - "Annotation" bias (Haynes, 2018)
- So far small number of explicit targets in cancer and even smaller number of treatments for those targets (Prasad, 2016; Joyner, 2019)
- Concerns about reproducibility of basic research
  - https://elifesciences.org/collections/9b1e83d1/reproducibility-project-cancer-biology

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Where has BMHI fallen short?

- EHRs and clinicians – current systems slow work of clinicians, prioritize non-clinical aspects of care, and lead to clinician burnout (Halamka, 2017; Gawande, 2018; NAM, 2019)
  - Clinicians want to read and write the story, which can be at odds with structured data we might want to use for decision support, research, etc. (Vigilante, 2018; Kommer, 2018)
- Standards and interoperability – HITECH led to systems that could not talk to each other (Adler-Milstein, 2017)
- Privacy and security – not limited to healthcare, but growing concern (Gostin, 2018)

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Recalibration: 21st Century Cures Act

- Comprehensive legislation passed in 2016 (Hudson; 2017; Kesselheim, 2017)
- Included some "correcting" aspects of HITECH Act (Anthony, 2020)
  - Data interoperability and "app" framework via SMART on FHIR (Gordon, 2020)
  - Prohibition of "information blocking" (Adler-Milstein, 2017; Black, 2018)
  - https://www.healthit.gov/curesrule/

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Informatics and COVID-19

- As with other areas of healthcare, many impacts from COVID-19
  - Our health information systems, especially public health, were not up to task (Gottlieb, 2021; Topol, 2022)
- Relaxation of US federal rules around (Verma, 2020)
  - Security – allowing communications platforms previously prohibited under HIPAA, e.g., Zoom, Face Time, etc. (HHS, 2020)
  - Telemedicine/telehealth – leading to rapid expansion in use (Mann, 2020)
  - Delay in compliance dates for Cures Rule (HHS, 2020)
- Expansion and problems with "open science" advances, e.g., preprints, open-access publishing, etc. (Lenzer, 2020)
- Attacks on science and scientists (Hotez, 2021)

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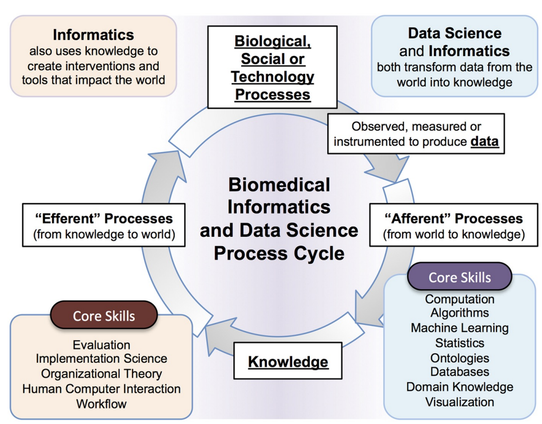
Is BMHI different from data science?

- (Payne, 2018)
- Consider process cycle of going from biological, social, or technical processes with afferent and efferent loops of going to and from knowledge
- Data science and informatics focused on generating knowledge but informatics also engaged in use of that knowledge in real world of health, healthcare, research and other areas



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Who does BMHI?

- Professionals
- Leaders
- Everyone else

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BMHI professionals

- Overlapping but distinctive from
  - Information technology (IT)
  - Health information management (HIM)
  - Data science
- What distinguishes BMHI professionals?
  - Application domain expertise – healthcare, biomedical research, public health, etc.
  - Focus on information more than technology
- Certification for professionals
  - Clinical informatics subspecialty for physicians (Detmer, 2014)
  - AMIA Health Informatics Certification for many of rest (Gadd, 2016)

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Workforce and competencies

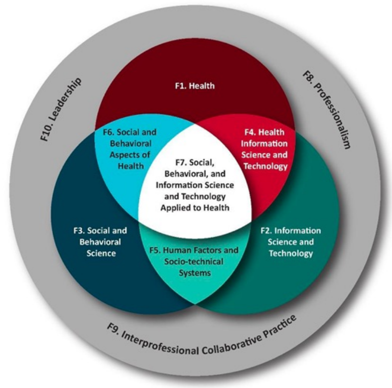
Health Informatics

Domains	Task statements	KS statements
Domain 1. Foundational Knowledge and Skills	NA	31
Domain 2. Enhancing Health Decision-making, Processes, and Outcomes	11	21
Domain 3. Health Information Systems	26	36
Domain 4. Data Governance, Management, and Analytics	17	28
Domain 5. Leadership, Professionalism, Strategy, and Transformation	20	28
Total	74	144

Clinical Informatics Subspecialty (CIS)

Domains	Task statements	KS statements
Domain 1. Foundational Knowledge and Skills	NA	26
Domain 2. Improving Care Delivery and Outcomes	7	28
Domain 3. Enterprise Information Systems	16	33
Domain 4. Data Governance and Analytics	10	27
Domain 5. Leadership and Professionalism	9	28
Total	42	142

(Silverman, 2019; Gadd, 2020; Hersh, 2020)



(Valenta, 2018)

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BMHI leaders

- Chief {X} Information Officer (Kannry, 2016) – where X =
  - Medical (CMIO) (Rydell, 2018)
  - Health (CHIO)
  - Research (CRIO)
  - Nursing (CNIO)
  - Data/Quality/Privacy/etc.
- Growing number of Chief Information Officers (CIOs) from informatics

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BMHI for everyone else

- Physicians and medical students – first addressed by AAMC Medical School Objectives Project (1998)
- Patients – 58% of US adults look online for health information and 35% attempt to diagnose illness in that manner (Fox, 2013)
- Clinical and translational scientists (Valenta, 2016)
- Next-generation research scientists (Moore, 2019)
- Nurses (Forman, 2020)

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Informatics and health professions education

- "Informatics training for clinicians is more important than hardware and software" (Safran, 2009)
- Health informatics is a "required skill for 21st century clinicians" (Fridsma, 2018)
- Competencies (Hersh, 2014; Hersh, 2020), curricula (Hersh, 2017), and challenges (Welcher, 2018)

1. Find, search, and apply knowledge-based information to patient care and other clinical tasks.
    a. Information retrieval/search—choose correct sources for specific task, search using advanced features, apply results.
    b. Evaluate information resources (literature, databases, etc.) for their quality, funding sources, biases.
    c. Identify tools to assess patient safety (e.g., medication interactions).
    d. Utilize knowledge-based tools to answer clinical questions at the point of care (e.g., text resources, calculators).
    e. Formulate an answerable clinical question.
    f. Determine the costs/charges of medications and tests.
    g. Identify deviations from normal (labs/x-rays/results) and develop a list of causes of the deviation.
2. Effectively read from, and write to, the electronic health record for patient care and other clinical activities.
    a. Graph, display, and trend vital signs and laboratory values over time.
    b. Adopt a uniform method of reviewing a patient record.
    c. Create and maintain an accurate problem list.
    d. Recognize medical safety issues related to poor chart maintenance.
    e. Identify a normal range of results for a specific patient.
    f. Access and compare radiographs over time.
    g. Identify inaccuracies in the problem list/history/medications list/allergies.
    h. Create useable notes.
    i. Write orders and prescriptions.
    j. List common errors with data entry (drop-down lists, copy and paste, etc.).
3. Use and guide implementation of clinical decision support (CDS).
    a. Recognize different types of CDS.
    b. Be able to use different types of CDS.
    c. Work with clinical and informatics colleagues to guide CDS use in clinical settings.
4. Provide care using population health management approaches.
    a. Utilize patient record (data collection and data entry) to assist with disease management.
    b. Create reports for populations in different health care delivery systems.
    c. Use and apply data in accountable care, care coordination, and the primary care medical home settings.
5. Protect patient privacy and security.
    a. Use security features of information systems.
    b. Adhere to Health Insurance Portability and Accountability Act (HIPAA) privacy and security regulations.
    c. Describe and manage ethical issues in privacy and security.
6. Use information technology to improve patient safety.
    a. Perform a root cause analysis to uncover patient safety problems.
    b. Maintain familiarity with safety issues.
    c. Use resources to solve safety issues.
7. Engage in quality measurement selection and improvement.
    a. Recognize the types and limitations of different types of quality measures.
    b. Determine the pros and cons of a quality measure, how to measure it, and how to use it to change care.
8. Use health information exchange (HIE) to identify and access patient information across clinical settings.
    a. Recognize issues of dispersed patient information across clinical locations.
    b. Participate in the use of HIE to improve clinical care.
9. Engage patients to improve their health care delivery though personal health records (PHRs) and patient portals.
    a. Instruct patients in proper use of a PHR.
    b. Write an e-message to a patient using a patient portal.
    c. Demonstrate appropriate written communication with all members of the health care team.
    d. Integrate technology into patient education (e.g., decision-making tools, diagrams, patient education).
    e. Educate patients to discern quality of online medical resources (websites, apps, patient support groups, social media, etc.).
    f. Maintain patient engagement while using an electronic health record (EHR) (eye contact, body language, etc.).
10. Maintain professionalism through use of information technology tools.
    a. Describe and manage ethics of media use (cloud storage issues, texting, cell phones, social media professionalism).
11. Provide clinical care via telemedicine and refer patients as indicated.
    a. Be able to function clinically in telemedicine/telehealth environments.
12. Apply personalized/precision medicine.
    a. Recognize growing role of genomics and personalized medicine in care.
    b. Identify resources enabling access to actionable information related to precision medicine.
13. Participate in practice-based clinical and translational research.
    a. Use EHR alerts and other tools to identify patients and populations eligible for participation in clinical trials.
    b. Participate in practice-based research to advance medical knowledge.
14. Apply machine learning applications in clinical care.
    a. Discuss the applications of artificial/augmented intelligence in clinical settings.
    b. Describe the limitations and potential biases of data and algorithms.

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Where can I learn more about BMHI?

- Self-learning
  - Can find much on the Web
    - e.g., http://informatics.health
- Continuing education
  - Many opportunities in many fields
    - e.g., AMIA 10x10 ("ten by ten") – https://amia.org/education-events/10x10-virtual-courses/10x10-oregon-health-science-university
- Formal training
  - Mostly at graduate level
    - e.g., OHSU – http://www.ohsu.edu/informatics-education
  - Largest funder for PhD and postdoc trainees is NLM
    - e.g., https://www.nlm.nih.gov/ep/GrantTrainInstitute.html

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Key challenges ahead for BMHI

- Improving usability of systems in clinical care, especially EHR
- Integrating omics and other sources of data
- Learning from data while protecting privacy and security
- Integrating new AI into healthcare professions and activities
- Achieving the goals of personalized/precision medicine

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Thank you!

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