Biomedical and Health Informatics: An Essential Discipline for 21st Century Medicine

University of Cape Town Department of Medicine Meet the Global Expert Webinar

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Biomedical and Health Informatics: An Essential Discipline for 21st Century Medicine

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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?
- Who are the people of BMHI?
- How can I learn more about BMHI, including in Africa?

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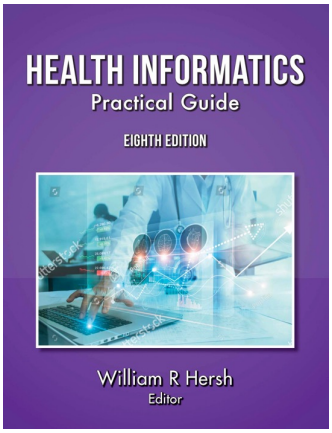
What is BMHI?

- I get asked this so often that I keep a Web site
  - http://informatics.health/
- And a blog
  - https://informaticsprofessor.blogspot.com/
- And a textbook
  - http://www.informaticsbook.info
- 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, 2020; Hersh, 2022)



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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." (ACGME, 2022)

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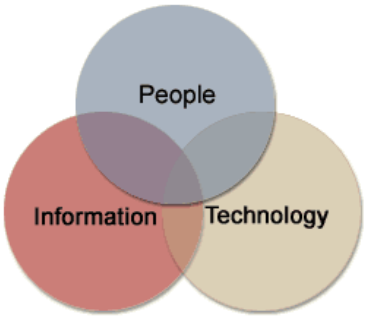
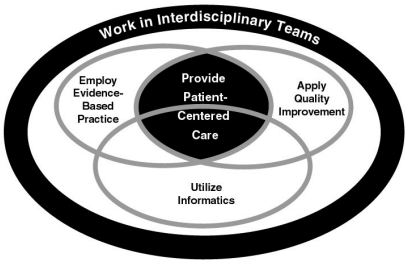
Some additional perspectives on BMHI



SUNY Buffalo

Overlap of Core Competencies for Health Professionals



(Greiner, IOM, 2003)

Fundamental Theorem (Friedman, 2009)

Goal of informatics is:

Goal is not:

Some subareas within BMHI (aka, its "adjective problem")

- Bioinformatics – focus on cellular and molecular biology, e.g., the "omes and omics"
- Clinical informatics – focus on healthcare and name of physician subspecialty in US
- Public health informatics – focus on public health
- Clinical research informatics – focus on managing research data and processes

Early work of BMHI focused on medical decision-making and artificial intelligence (AI)

- Earliest paper attributed to Ledley and Lusted (1959) aiming to model physician reasoning through symbolic logic and probability
- Warner (1961) developed mathematical model for diagnosing congenital heart disease
  - System predicted diagnosis with the highest conditional probability given a set of symptoms
- Next was emergence of AI and "expert systems" – computer programs mimicking human expertise
  - Early work focused on manually-constructed rule-based 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 disease profiles and scoring algorithms
  - INTERNIST-1 (Miller, 1982) and DxPlain (Barnett, 1987) largest and most well-known
- "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)
- Manual construction and maintenance of knowledge bases not scalable or sustainable
  - Led to "AI winter" between 1990-2010
  - More focused clinical decision support emerged in 1990s and used at present (Greenes, 2021)

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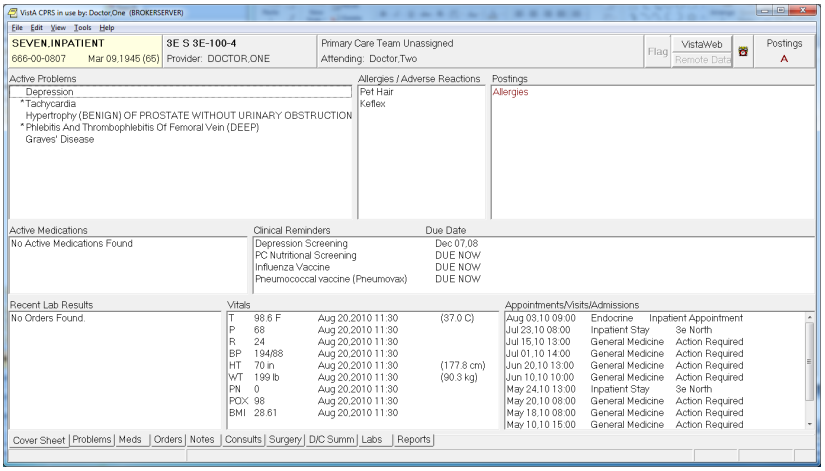
Another early focus of BMHI was electronic health record (EHR)

- With exception of VistA from Veteran's Administration, most "home-grown" EHRs have been replaced by commercial systems, mainly Epic and Cerner



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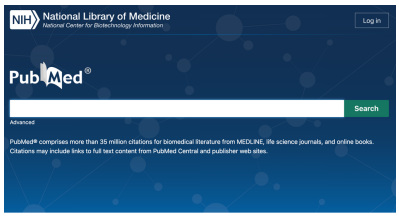
Early role of National Library of Medicine (NLM)

- 30-year leadership of Donald Lindberg, MD (Miller, 2021), passed to Patricia Brennan, PhD, RN (Brennan, 2016) in 2016
- Early application was information retrieval from bibliographic databases
  - From *Index Medicus* books 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, omics databases, and other knowledge resources
- Leader in terminology development and standardization (Humphreys, 1998)
- Funder of primary research as well as training grants and other educational activities (Humphreys, 2022)



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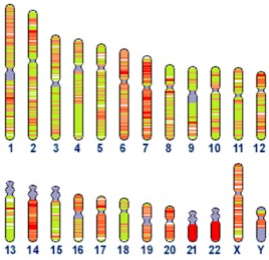
Genomics and bioinformatics

- 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)
  - Although last 8% not fully sequenced until 2022 (Nurk, 2022)
- Cost and time of sequencing has dropped dramatically, leading to increased understanding of genomic variation and complexity as well as emergence of precision medicine paradigm (Denny, 2019; Collins, 2021)
- Many data resources from NLM National Center for Biotechnology Information (NCBI) (Sayers, 2023) and others (Rigden, 2023)



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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)

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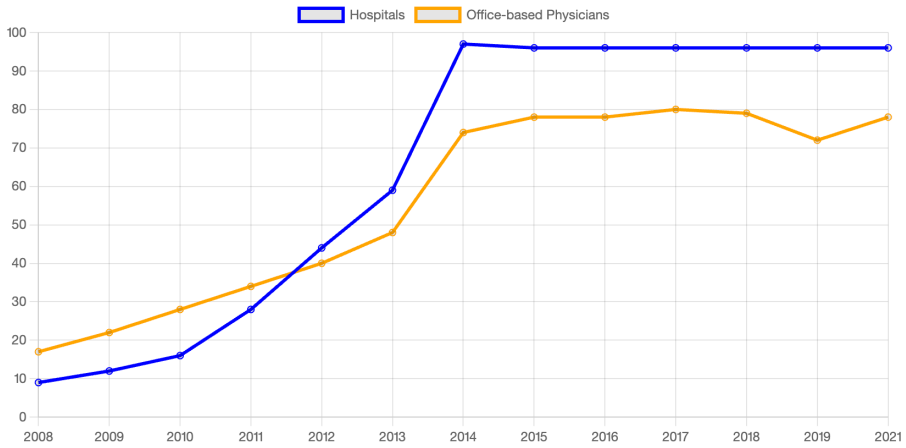
Has resulted in growth of EHR use in US



https://www.healthit.gov/data/quickstats/national-trends-hospital-and-physician-adoption-electronic-health-records

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Re-emergence of AI

- "Predictive AI" driven by advances in machine learning, increasing availability of data, and more powerful computers and networks (Topol, 2019; Rajpurkar, 2022)
- Most success has been with image interpretation (Rajpurkar, 2023); examples include
  - Radiology – chest x-rays for diagnosis of pneumonia and tuberculosis
  - Ophthalmology – retinal images for diagnosis of diabetic retinopathy
  - Dermatology – skin lesions for diagnosis of cancer
  - Pathology – breast cancer slides to predict metastasis
- But achievements in other areas
  - Predicting adverse events in hospitalizations (Rajkomar, 2018)
  - Generating clinical notes from patient and physician verbal interaction (Rajkomar, 2019)
  - Predicting protein folding from amino acid sequences (Jumper, 2021)

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Re-emergence of AI (cont.)

- Also success in "seeing" where humans cannot, e.g.,
  - Age and sex determination from retinal images (Poplin, 2018) and ECG (Attia, 2019)
  - Cardiac function and valvular heart diseases (Ueda, 2023) and diabetes (Pyrros, 2023) classified from chest x-rays
- But real-world use and evidence base still modest
  - Systematic review of clinical trials of predictive AI systems showed small number of trials (relative to predictive modeling papers), mediocre methodologies, and mixed results (Plana, 2022)

And now, "generative AI"

- Introduction of ChatGPT in November, 2022 brought a new type of AI into focus: generative AI
- LLMs based on transformer models trained with large amounts of text and achieving unprecedented ability to chat and answer questions (Thirunavukarasu, 2023)
- Although imperfect and sometimes prone to confabulation (Lee, 2023), impressive performance in medicine and beyond, e.g.,
  - US Medical Licensing Exam (USMLE) (Nori, 2023)
  - Board exams in e.g., radiology (Bhayana, 2023) and clinical informatics (Kumah-Crystal, 2023)
  - New England Journal of Medicine clinical cases (Kanjee, 2023)
  - Answering questions in social media forums (Ayers, 2023)
  - Drafting letters to patients (Ali, 2023)

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)
- Data standards and interoperability – HITECH led to systems that could share data and information (Adler-Milstein, 2017)
- Privacy and security – not limited to healthcare, but growing concern (Gostin, 2018)

Will AI help or hinder medicine?

- "AI won't replace radiologists, but radiologists who use AI will replace radiologists who don't," (Langlotz, 2019)
- Must address bias in data and algorithms
  - AI may compromise care (DeCamp, 2023)
  - Implementing responsible (Dorr, 2023) and fair (Chen, 2023) AI
- Need to determine appropriate role in medical education – usage and understanding (Meyer, 2023)

Who are the people of BMHI?

- Professionals
- Leaders
- Everyone else

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BMHI professionals

- Overlapping but distinctive from
  - Computer science (CS) and 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

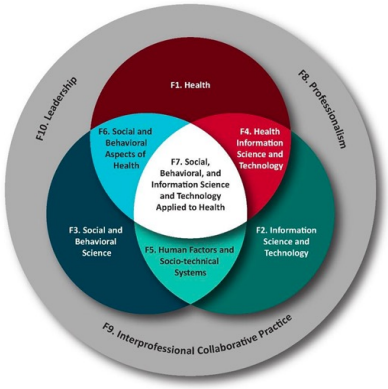
Health Informatics

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)

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)
- Need to prepare physicians for the "clinical algorithm era" (Goodman, 2023)

1. Find, search, and apply knowledge-based information to patient care and other clinical tasks
2. Effectively read from, and write to, the electronic health record for patient care and other clinical activities
3. Use and guide implementation of clinical decision support (CDS)
4. Provide care using population health management approaches
5. Protect patient privacy and security
6. Use information technology to improve patient safety
7. Engage in quality measurement selection and improvement
8. Use health information exchange (HIE) to identify and access patient information across clinical settings
9. Engage patients to improve their health and care delivery though personal health records and patient portals
10. Maintain professionalism through use of information technology tools
11. Provide clinical care via telemedicine and refer patients as indicated
12. Apply personalized/precision medicine
13. Participate in practice-based clinical and translational research
14. Apply machine learning applications in clinical care

Opportunities for BMHI in Africa

- Health and Human Heredity in Africa (H3Africa) – https://h3africa.org/
  - H3ABionet bioinformatics support (Mulder, 2016) – https://www.h3abionet.org/
- Data Science Initiative for Africa (DSI Africa) – 20+ research, training, ethics, and coordinating grants funded by US National Institutes of Health (NIH)
  - Computational Omics and Biomedical Informatics Program (COBIP) collaboration between UCT and OHSU, with MPIs
    - Tinashe Mutsvangwa, UCT
    - Bill Hersh, OHSU

DS-I Africa
Data Science for Health Discovery and Innovation in Africa

https://dsi-africa.org/

COBIP
COMPUTATIONAL OMICS AND BIOMEDICAL INFORMATICS PROGRAM

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Online courses and faculty expertise in clinical informatics and research informatics

Key challenges ahead for BMHI

- Improving usability of information 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 practice

Thank you!

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