A Short History of Biomedical and Health Informatics

What is Biomedical & Health Informatics?
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Key historical developments of informatics

- Ledley & Lusted paper
- First era of artificial intelligence (AI)
- Early electronic health records (EHRs)
- National Library of Medicine (NLM) and early information retrieval (IR) systems
- Internet and World Wide Web
- Genomics and bioinformatics
- Institute of Medicine reports
- Health Information Technology for Clinical & Economic Health (HITECH) Act
- Second era of AI
- Backlash and recalibrating: 21st Century Cures Act
- Informatics and COVID-19
- Key challenges ahead

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Ledley & Lusted (1959)

- Attributed as scientific origin of field
- Aimed to model and understand physician reasoning through
  - Symbolic logic – representing concepts such as patient findings, tests, diagnoses, etc.
  - Probability – likelihood of outcomes (e.g., diagnosis) based on concepts (symbols)
  - Value theory – complexity of values going into medical decision-making
- Led to early attempts at computer-based decision-making in medicine
- Widely cited (Beck, 1984)

First era of 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)

First era of AI

- 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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Era also saw 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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NLM and early IR systems

- A critical organization in history of informatics was NLM
  - 30-year leadership of Donald Lindberg, MD, with torch passed to Patricia Brennan, PhD, RN (Brennan, 2016)
- Early application was IR 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 research as well as training grants and other education

NATIONAL LIBRARY OF MEDICINE

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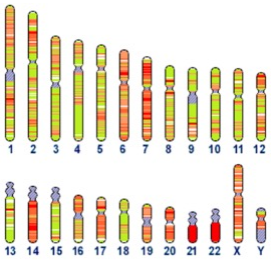
Genomics and bioinformatics

- Human Genome Project to sequence human genome began in 1988
- In 2001, NIH-based project published "first draft" (NIH, 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



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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, 2020) and others (Rigden, 2020)
- Manifested in precision medicine and other clinical advances (Denny, 2019; Collins, 2021)
- 20-year retrospectives noted accomplishments but also challenges still ahead in science and policy (Gates, 2021; Jones, 2021)

Internet and World Wide Web

- Emergence in 1980s of Internet – network of networks
  - Initial use focused on sharing information, e.g., file transfer, email
- Major application empowering Internet was World Wide Web
  - 1990s boom and bust in dot-com era
  - Subsequent success of business models, e.g., Facebook/Apple/Amazon/Netflix/Google (FANG)
- Ubiquitous now with wired (broadband) and wireless (wifi, cellular) connectivity

Important thought leadership led by initial IOM reports

- The Computer-Based Patient Record (1997) – paper records illegible, inefficient, and error-prone; computer-based record vital to modern healthcare
- For the Record: Protecting Electronic Health Information (1997) – benefits of electronic health information compromised by inadequate protection; informed HIPAA legislation
- Networking Health (2000) – value of networks important but do not need separate health Internet
- To Err is Human (2000) – medical errors are common and a systems problem

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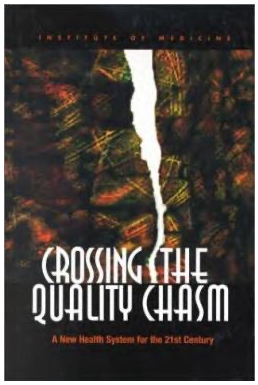
Next round of IOM reports laid out vision for better healthcare system



- Crossing the Quality Chasm – set of aims and rules for high-quality 21st century healthcare (2001)
- Aims included care that was
  - Safe – avoid injuries from care intended to help
  - Effective – provide service based on scientific knowledge and avoid care unlikely to benefit
  - Patient-centered – care respectful of patients' preferences, needs, and values
  - Timely – reduce waits and delays in care
  - Efficient – avoid waste of equipment, supplies, and energy
  - Equitable – provide care that does not vary based on personal characteristics

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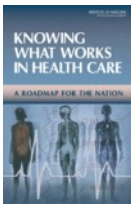
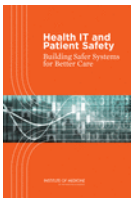
Other important IOM reports and concepts

- The "learning health system" must measure provision and outcomes of care to know what works (2008)
- Components of learning health system (2012) included
  - Transparency of data and information
  - Reward outcomes and value, not volume
  - Errors promptly identified and corrected
- Health IT systems that improve healthcare may also introduce error and cause harm if not designed and applied properly (2012)





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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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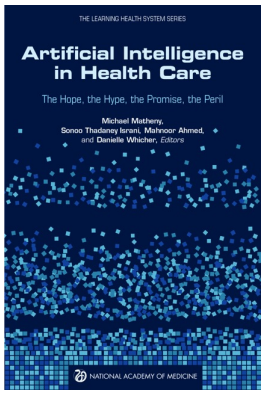
Second era of AI

- Reinvigorated with success of machine learning, which occurred from increasing availability of data, more powerful computers, and advances in deep learning (Topol, 2019; Esteva, 2021)
- Popularized by Topol (2019)
- Retrospective from Shortliffe (2019)
- Drawing attention from leading policy bodies, such as National Academy of Medicine (Matheny, 2019)



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Backlash

- 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)
- 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 (Health Care Industry Cybersecurity Task Force, 2017)

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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 (Sweeney, 2019)
  - 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/

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)
- 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)

Key challenges ahead based on historical perspectives

- Improving usability of systems in clinical care, especially EHR
- Access to data, information, and knowledge
- Learning from data while protecting privacy and security
- Integrating new AI into healthcare professions and activities