A Short History of Biomedical & Health Informatics

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 challenges ahead
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)

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)
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., FLHILL; Lindberg, 1986) to PCs (e.g., Grateful Med; Lindberg, 1990) 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

Genomics and bioinformatics

• Human Genome Project to sequence human genome began in 1988
• In 2001, NIH-based project published “first draft” (Anonymous, 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
- Data resources from NLM National Center for Biotechnology Information (NCBI) (Nucleic Acids Research, 2018)

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* (Dick, 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* (Kohn, 2000) – medical errors are common and a systems problem

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 (IOM, 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
Other important IOM reports and concepts

- The “learning health system” must measure provision and outcomes of care to know what works (Eden, 2008)
- Components of learning health system (Smith 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 (Anonymous, 2012)

ARRA and the HITECH Act

- By mid-2000s, emergence of research supporting value of EHR and CDS for improving quality and safety of healthcare
- 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)
Second era of AI (Topol, 2019)

Screening for cardiac contractile dysfunction using an artificial intelligence-enabled electrocardiogram

Zachi AHIYa, SoniKrap, TriniBonillo-Lopez-Amere, Paul M. MaW(1), Dorothy L. Ludin(1),
GanetteSharian, Patrick A. Pohh(1), MauricioBritoGomes, Peter A. Bo$Clement(1),
Thomas R. M. Blagek, SerenaL. Askarzadeh, Christopher G. Scott(2), RickeyJ. Carter(1) and
PaulA. Taubman(1)

Cardiologist-level arrhythmia detection and classification in ambulatory electrocardiograms using a deep neural network

Avital Y. Manoura(1), PaulS. Applebaum(1), AdamK. Bakshish(1), DavidJ. Thoma(1),
CédricBourdier, WouterP. Verhulst(1) and Kenneth S. Ng(2)

Predicting the early risk of chronic kidney disease in patients with diabetes using real-world data

StanislavKrap(1), YongHeuser, AngelaAdler, LeonBolte, AlexanderBauer, FrederickF. Fletcher,
BatHeinzmann, HelenaKong, Scott M. McAlister, Daniel H. Robinson(3), TitusSchelter(1),
RennelSchwabler and WolfgangHieck(1)

A guide to deep learning in healthcare

AndreaEtzioni(1), AlexandreRicardou(1), BhavishKamdar, VolodymyrKhalidev, MarkDePristo,
KatherineChou, ClaireGul, GregGardiner, SebastianThran and JeffDoser

Medicine in the digital age

Deeper learning

Machine learning makes new sense of psychiatric symptoms

BriannaHoffman

Identifying facial phenotypes of genetic disorders using deep learning

HausBouwmeester(1), JanM. Wiel(2), PauRabot(1), ReinWillen(1), RichardNachreiner,
CordulaSchild, DavidE. Wiel(1), CorneelKampman(1), MartinLoetzer(1), LoppensAndré(1)
and KevinH. Kong(1)

A call for deep-learning healthcare

BeauNorgaot, BenjaminS. Glucksberg and AtulJ. Butte

The practical implementation of artificial intelligence technologies in medicine

JianjingHu(1), SatoshiKan(1), XiYu(1), ZhengRui(1), XuelingZhou and KangZhang(1)

Guidelines for reinforcement learning in healthcare

GlenKrnjevic, FarhadJafarzadeh, MathiasKustermann, HitoshiKashiba, DavidSang, PradeepScheule and
AngelaC. Stock

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)
• 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)
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