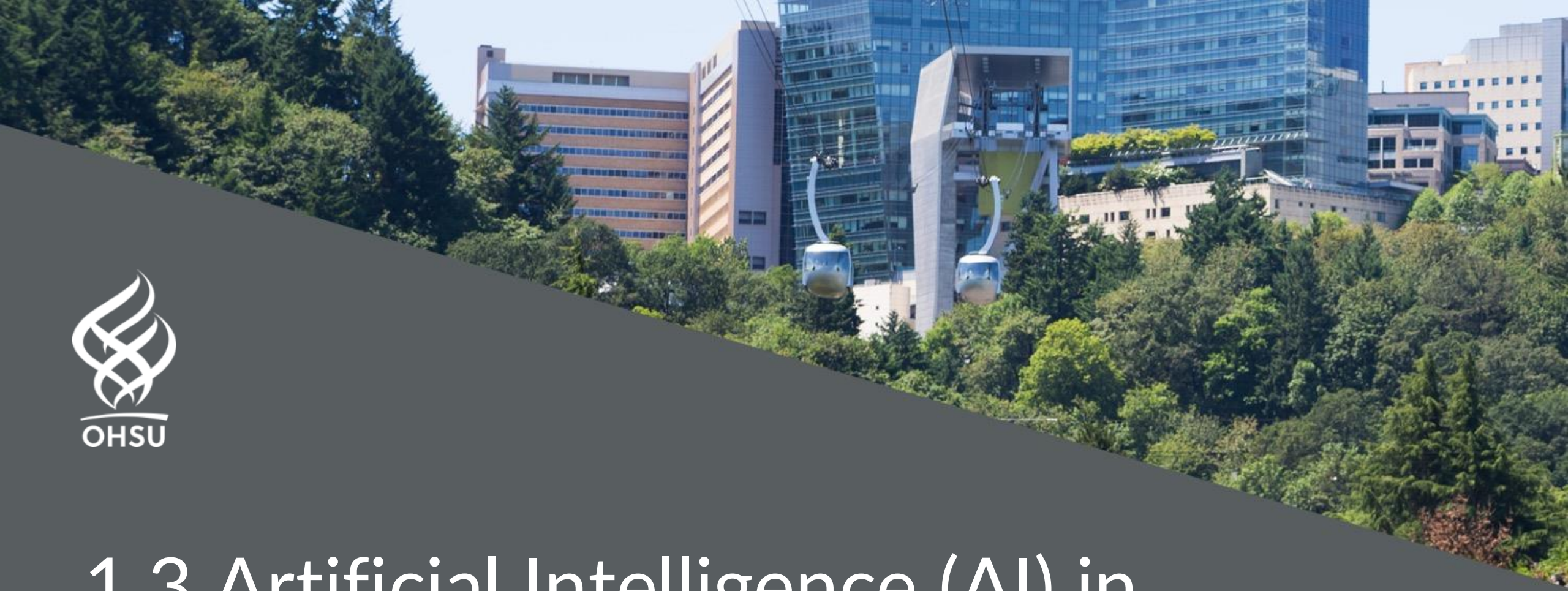


1.3 Artificial Intelligence (AI) in Biomedicine and Health

What is Biomedical and Health Informatics? - http://informatics.health/
William Hersh, MD, FACMI, FAMIA, FIAHSI

Definitions

- Artificial intelligence
- Machine learning
- Data science
  - Data analytics
- Big Data
- Data mining
  - Text mining
- Other terms

Artificial intelligence (AI)

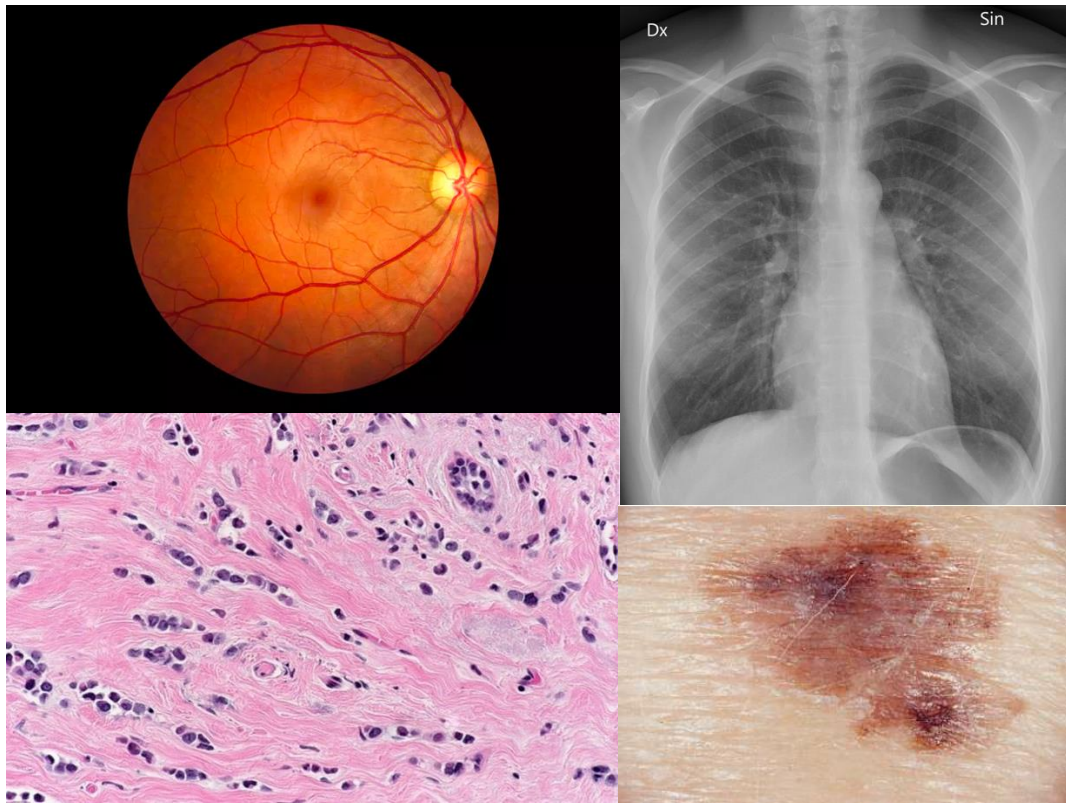
- AI – information systems and algorithms capable of performing tasks associated with human intelligence (Copeland, 2024)
  - Origin of term attributed to summer workshop held at Dartmouth College in 1956 (McCarthy, 2007)
- Some classify AI into two broad categories (Khare, 2023)
  - Predictive AI – use of data and algorithms to predict some output, e.g., diagnosis, treatment recommendation, prognosis, etc.
  - Generative AI – generates new output based on prompts, e.g., text, images, etc.

Machine learning (ML)

- Term originally attributed to Arthur Samuel in 1959: “Field of study that gives computers the ability to learn without being explicitly programmed” (McCarthy, 1990)
- Derived from
  - Mathematics/statistics – learning relationships from data
  - Computer science – emphasis on efficient algorithms, especially those involving large amounts of data
- Much recent success from deep learning (DL) (Shah, 2022)
  - ML associated with use of neural networks that have deep layers requiring substantial processing (Sevilla, 2022)
  - Initial success in predictive AI, in areas such as image classification, including in medicine (Esteva, 2021)
  - Also has facilitated large language models (LLMs) that drive generative AI (Telenti, 2023; Raschka, 2024)

Predictive AI in medicine

- Driven by advances in ML, increasing availability of data, and more powerful computers and networks (Topol, 2019; Rajpurkar, 2022)
  - Deep learning in imaging breakthroughs by Hinton (2006)
- Most success in 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
- Also beyond imaging
  - Adverse events in hospitalizations from EHR data (Rajkomar, 2018)
  - Predicting structure of protein from amino acid sequences (Jumper, 2021; Abramson, 2024)



And now, generative AI

- Introduction of ChatGPT on November 30, 2022 brought this new type of AI into focus
- Based on large language models (LLMs) that use deep neural networks, large amounts of training data, and tuning for specific tasks (Omiye, 2024)
  - Trained on massive amounts of text and other content, e.g., large Web crawls, books, Wikipedia, and more (Roberts, 2022)
  - Use transformer models that predict words in sequence from billions/trillions of words and add measures of importance to "attention" words (Raschka, 2024)
  - Fine-tuned with reinforcement learning from human feedback (RLHF) (Lambert, 2022)
  - Activated by (and importance of) prompting (Liu, 2023; Meskó, 2023)

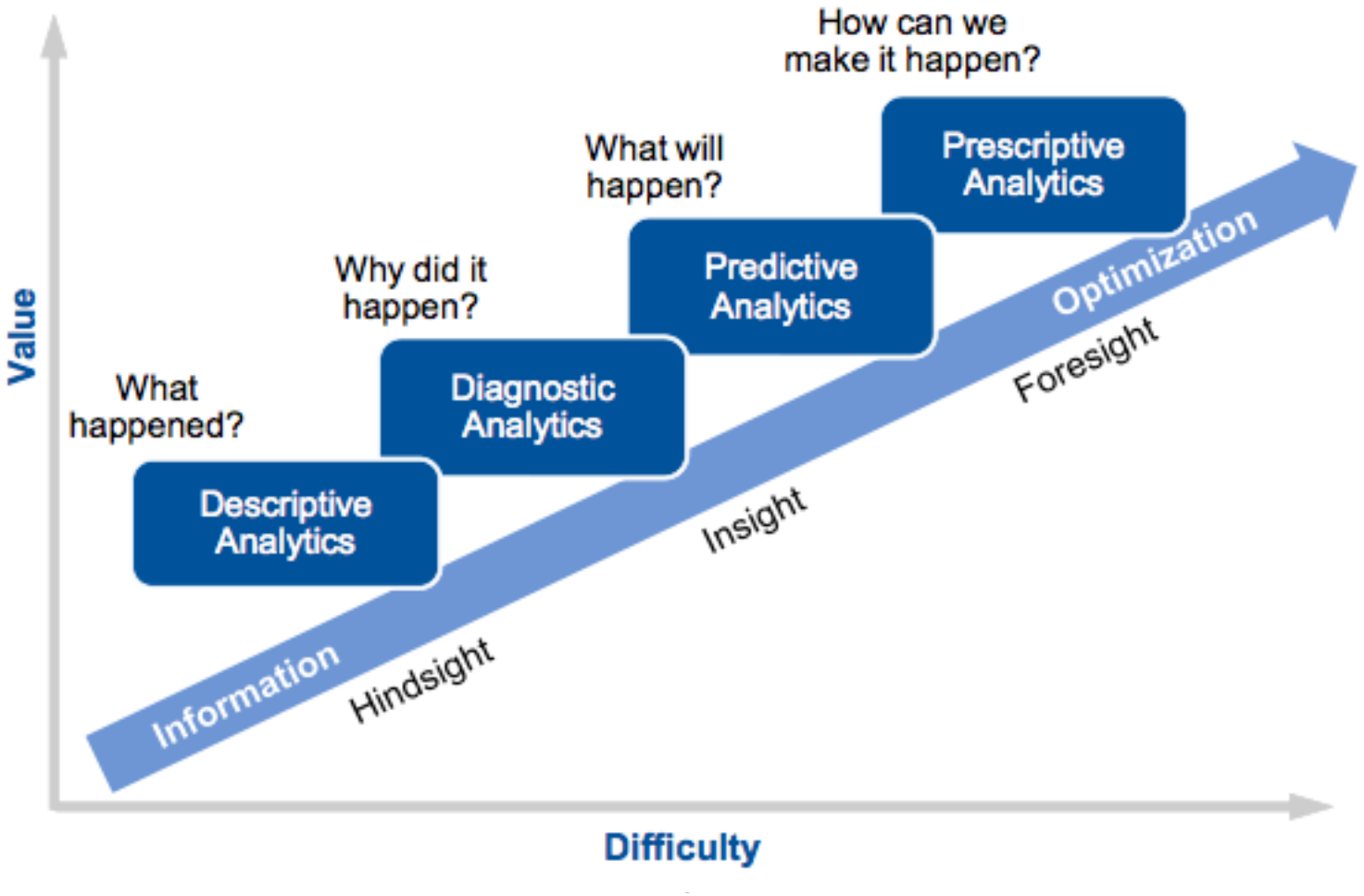
Data science

- “The science of learning from data; it studies the methods involved in the analysis and processing of data and proposes technology to improve methods in an evidence-based manner” (Donoho, 2017)
- “Data science encompasses a set of principles, problem definitions, algorithms, and processes for extracting nonobvious and useful patterns from large data sets” (Kelleher, 2018)
- Applications in biomedicine (Hoyt, 2019; Topol, 2019; Patrishkoff, 2023)
- Technical but non-mathematical overview (Spector, 2022)

Data analytics

- “The extensive use of data, statistical and quantitative analysis, explanatory and predictive models, and fact-based management to drive decisions and actions” (Davenport, 2017)
- “The systematic use of data and related business insights developed through applied analytical disciplines (e.g. statistical, contextual, quantitative, predictive, cognitive, other [including emerging] models) to drive fact-based decision-making for planning, management, measurement and learning” (IBM, 2012)

Levels of data analytics from Gartner (Schaap, 2020)



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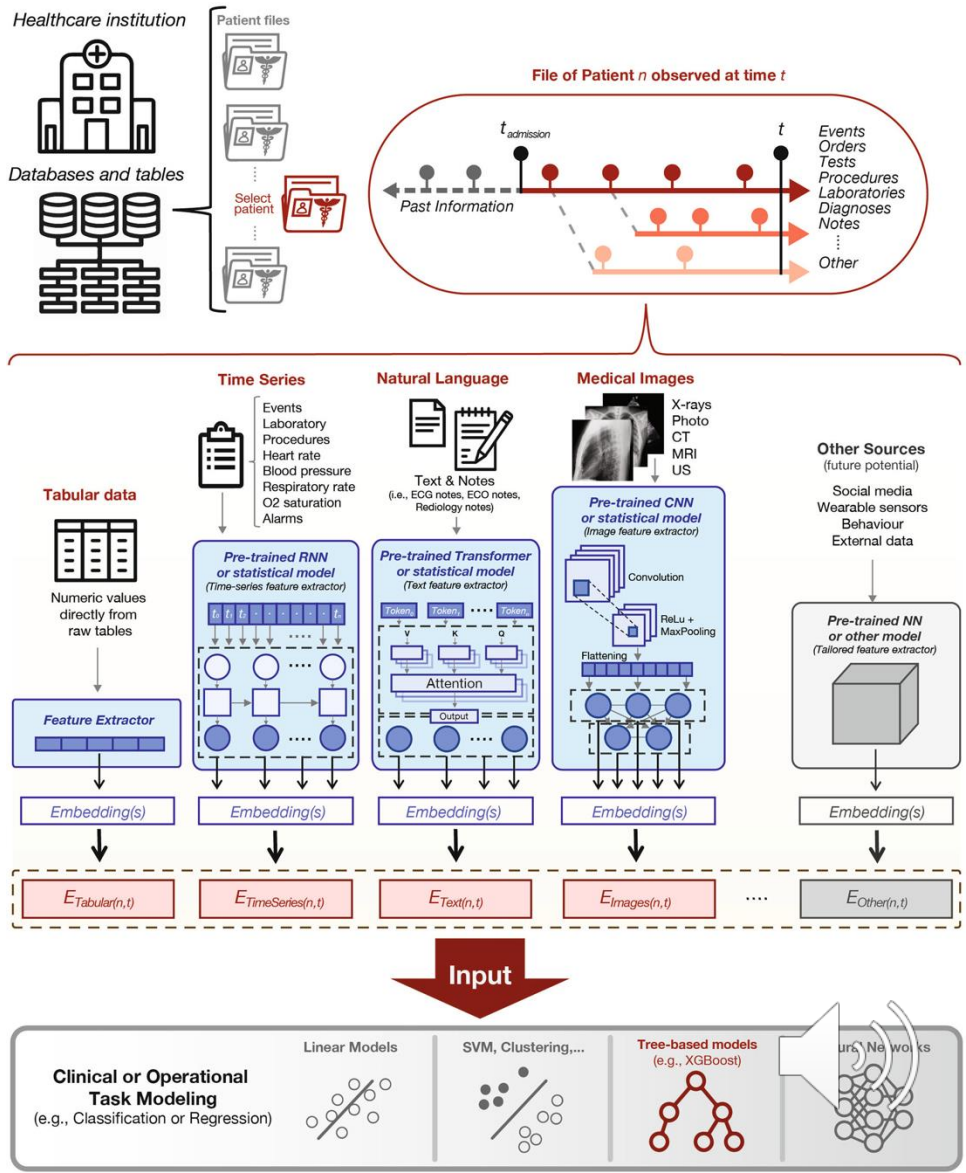
Big Data

- Various 4 Vs (e.g., Zikopolous, 2011); most recent from NIST (Chang, 2019)
  - Volume
  - Velocity
  - Variety
  - Variability
- Multimodal data (Acosta, 2022; Soenksen, 2022; Topol, 2023)
  - Data from many modalities, from cells and molecules; tissues and organs; and organisms, populations, and the global environment



Data mining

- Processing and modeling of data to discover previously unknown patterns or relationships (Bellazzi, 2008; Zaki, 2020)
- Text mining – applying data mining to unstructured textual data (Aggarwal, 2012; Cohen, 2013)
- Terms have faded from vernacular but methods still used

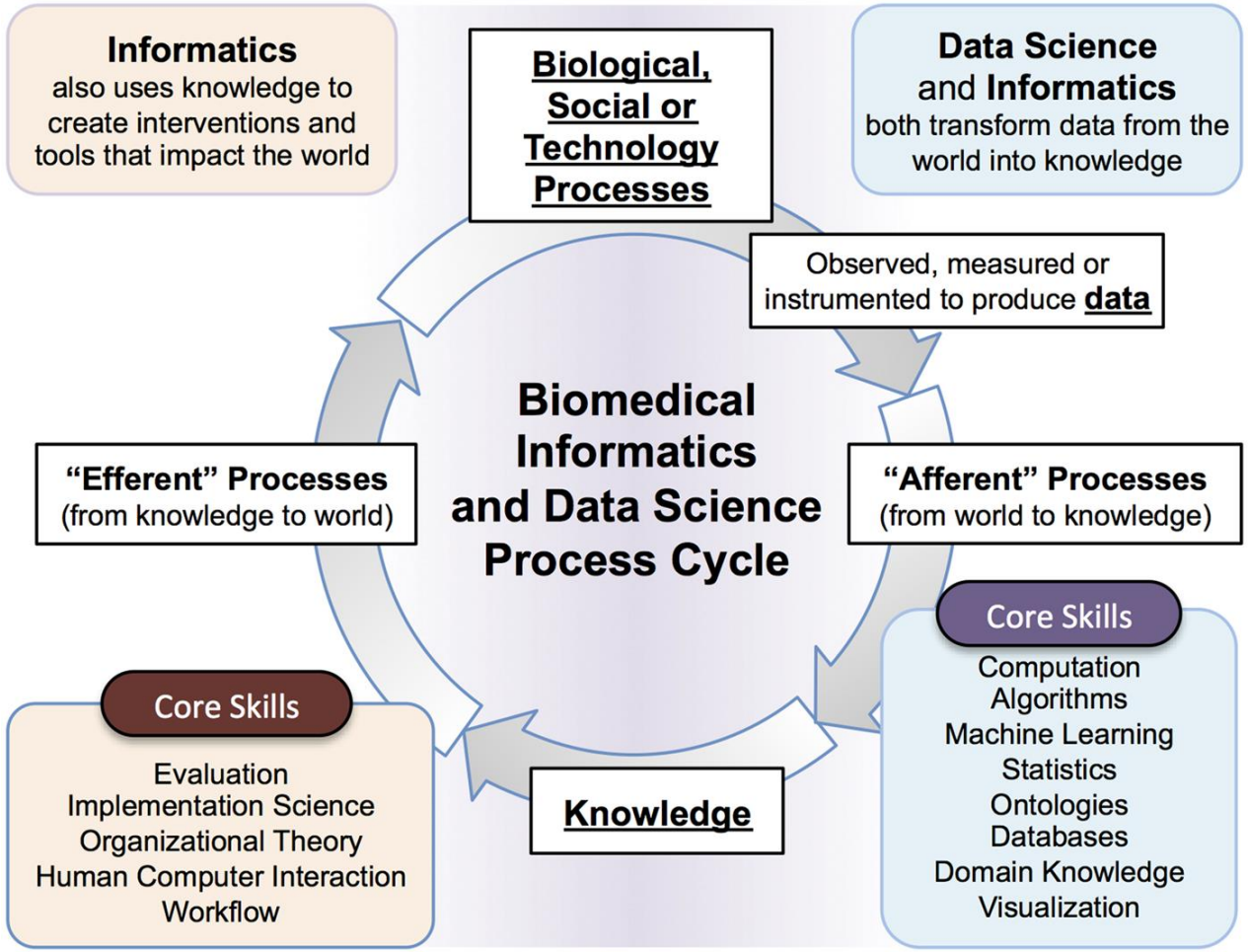
Some other terms

- Data provenance – origin and trustworthiness of data (Doan, 2012)
- Business intelligence – use of data to obtain timely, valuable insights into business and clinical data (Brijs, 2012)
- Precision medicine (IOM, 2011; Collins, 2015; Denny, 2019); formerly personalized (Hamburg, 2010) or computational medicine (Winslow, 2012)
- “Re-use” or “secondary use” of clinical data, especially from the electronic health record (EHR) (Safran, 2007; Meystre, 2017) and other sources (Näher, 2023)

Other terms (cont.)

- Metadata – data about data (Riley, 2017)
- Model – representation of data, structure, and relationships for ML (and other tasks) (Shin, 2020)
- Data visualization – applying visual methods to “tell the story” about the data (Wilke, 2019)
- Data wrangling – processing data into format suitable for analytics, learning, visualization, etc.

Where does informatics fit into data science/AI/ML (Payne, 2018)?



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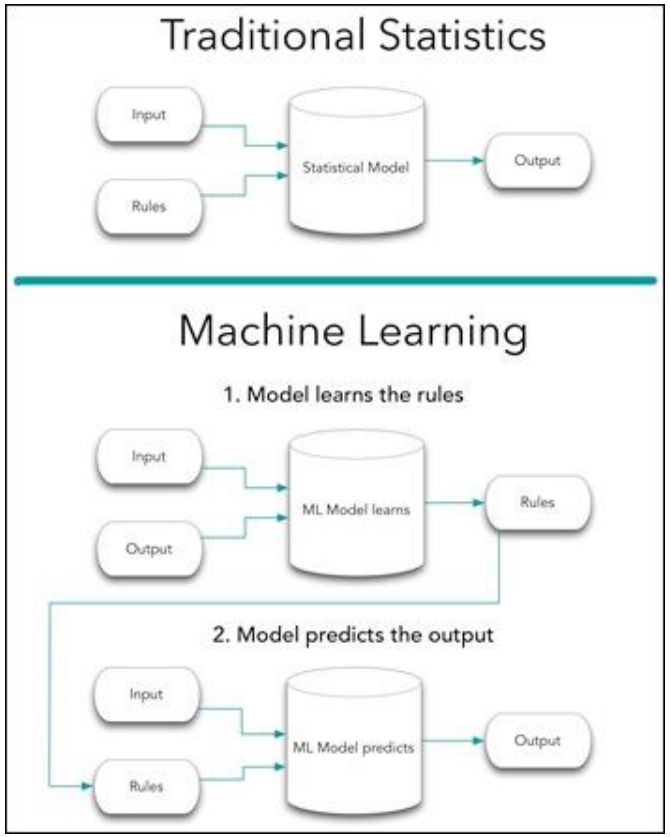
How does data science/ML differ from statistics?

- Statistics draws population inferences from samples; ML finds generalizable predictive patterns (Bzdok, 2018)
  - Diagrammed by Scarlat (2019)
  - A false dichotomy? (Finlayson, 2023)
- Data scientist is a “person who is better at statistics than any software engineer and better at software engineering than any statistician” (Donoho, 2017)



Historical developments of AI in biomedicine

- Early focus on “expert systems” that performed diagnosis
- Functions of systems tightly linked to methods for knowledge representation
- Four general approaches
  - Clinical algorithms (flowcharts)
  - Bayesian statistics
  - Production rules (if-then rules) (Shortliffe, 1975)
  - Scoring and heuristics (profiles of diseases and findings) (Miller, 1982)

End of first era of (20th century) AI

- By the late 1980s and early 1990s, it was apparent that
  - Diagnostic process too complex for computer programs
  - Systems took long time to use and did not provide information clinicians truly needed
  - Manual construction of algorithms, rules, and knowledge bases not efficient or scalable (Haigh, 2024)
  - “Greek Oracle” model inappropriate to medical usefulness (Miller, 1990)
- Rule-based approaches evolved into clinical decision support in the 1990s with recognition of value within EHR

Into second era of (21st century) AI

- From “AI winter” to resurgence of predictive AI due to advances in
  - Machine learning, especially deep learning applied to image classification (Hinton, 2006)
  - Growth in quantity of data, including electronic health records (EHRs), omics, social media, mobile devices, etc.
  - Improvements in computers and networks
- Popularized by Topol (2019)
- Further advances in generative AI and large language models (LLMs) starting in late 2022