Harvard-MIT Division of Health Sciences and Technology HST.950J: Engineering Biomedical Information: From Bioinformatics to Biosurveillance Course Directors: Dr. Isaac Kohane, Dr. Marco Ramoni



Harvard Medical School



Massachusetts Institute of Technology

Biomedical Computing

Introductory Lecture September 8, 2005

6.872 / HST 950



Introduction

- Definition: Development and application of information science and technology for/to/in biomedical sciences.
- Background: Biomedical sciences and engineering.
- For (support): Improve management, research and care in terms of speed, accuracy, efficiency (electronic clinical records).
- To (enablement): Do something that would be otherwise impossible (CAT scan and shotgun sequencing).
- In (embedment): Be integral part of biomedical research (bioinformatics).



History of Biomedical Informatics

- **1950**: R. Ledley's dental data at National Bureau of Standards.
- **1959**: Reasoning foundations of medical diagnosis, *Science*.
- **1964**: NIH Computer Research Study Section.
- 1966: MGH starts the development of COSTAR/MUMPS.
- 1971: MEDLARS Online, National Library of Medicine.
- **1972**: NLM fellowship training program started.
- 1974: Mycin/Internist: dawn of Artificial Intelligence in Medicine.
- **1988**: Human Genome Initiative and NCBI started.
- **1995**: Shotgun sequencing (computer intensive) developed.
- 2001: Publication of Human Genome draft.
- 2003: Medical Informatics becomes Biomedical at Columbia.
- 2003: Computer scientist moves to Genetics, Stanford.
- 2006: HMS Dean announces Center for Biomedical Informatics





For (Support) Biomedical Informatics

Scope: Processing of biomedical information.

- Goal: Support, streamline, and make more efficient the processing of biomedical information (clinical records, lab tests, etc).
- Impact: Empower patients with control and dissemination of information; make healthcare delivery more efficient and cheaper; improve healthcare quality (96,000 death/year in US due to medical error).
- Applications: Electronic medical records, information security and confidentiality, telemedicine.





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To (Enabling) Biomedical Informatics

Scope: Use computer to .

- Goal: Develop applications leveraging on information science and technology.
- Impact: Change the way medical care is delivered; develop new tests and therapies; deliver new methods to control and manage disease outbreaks.
- Applications: Genomic sequencing; image processing; real-time surveillance and early detection of population-wide events.



In (Embedded) Biomedical Informatics

Scope: Analysis of biomedical information.

- Goal: Develop methods to analyze biomedical data as part of the overall research endeavor.
- Impact: Change the way biomedical data are analyzed; deliver new views of natural phenomena; develop new methods to discover the bases of diseases.
- Applications: Genomics, proteomics, bioinformatics.



Topic Areas

Bioinformatics: Application of information technology to genomic information.

- Clinical Informatics: Application of information technology to medical (clinical) information.
- Biosecurity and Public Health: Application of information technology to population data to monitor and infer global behavior and detect unexpected events.
- Note: there are for/to/in components of biomedical informatics in all of these areas.





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Bioinformatics

Bioinformatics: Research, development, or application of computational tools and approaches for expanding the use of biological, medical, behavioral or health data, including those to acquire, store, organize, analyze, or visualize such data.

Computational Biology: Development and application of dataanalytical, theoretical methods, mathematical modeling and computational simulation to the study of biological, behavioral, and social systems.

BISTIC Definition Committee, NIH, 2000.

Flesch Readability Index: 0.1 - Bush Inaugural (75), Sports Illustr. (65); NY Times (39); Auto Insurance Policy (10).

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Bioinformatics

- Since work started in the past decade, we now have the complete genetic sequences of over 600 organisms.
- Complete human genome draft published, with final quality sequence, on April 25, 2003.
- * At least 40-60% of genes in genome have unknown function. Many, many fewer have a solved structure.



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Bioinformatics

- Genome: sequencing and genotyping.
- Transcriptome: expression and regulation.
- Proteome: protein expression and interactions.
- Metabolome: biological process on a larger scale.
- Bibliome: navigation and mining of annotation databases.
- Pharmacogenomics: genomics to create/test terapies.
- Genomic Privacy: data security.







Clinical Informatics

Clinical Databases: electronic medical records.

- Patient Privacy and Confidentiality: cryptology and distributed systems.
- Just in time clinical information: real-time delivery of medical information.
- Telemedicine: remote distribution/collection of medical information.
- Pharmaco-economics: Optimization and prediction of drug discovery and development.





Biosecurity

- Public Health Informatics: Collection and distribution of population-wide medical information.
- Geospatial Analysis: Analysis of population-wide medical data.
- Population Confidentiality: Collecting population data without endangering the single individual.
- Biosecurity: Real-time surveillance of man-made or natural disease outbreaks.





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Syllabus

Class: Tuesdays and Thursdays, 2:30-4:00pm.

Text: Shortliffe EH, Perreault LE, Wiederhold G and Fagan LM, Medical Informatics: Computer Applications in Health Care and Biomedicine, 2nd Edition. Springer 2001.





Goals and Rationale

Objective: Introduction to the basic notions and current trends of biomedical informatics.

Strategy: In-depth description of basic notions through their presentation in basic trends:

Basic Notion Lectures: Necessary and sufficient information covering a particular topic.

Case Study Lectures: Guest lecturers describe a particular project/problem/trend in a domain.

Lecturers: Computer scientists, CIOs, computational biologists, geneticists, clinical trials leaders, biosecurity experts.



Grading

- **Class participation (30%)** Attendance and contribution to discussions are a critical component of the class. Much of the material will be taught by guest lecturers who are uniquely knowledgeable in their areas.
- Homework assignments (30%) We plan to give a half-dozen homework assignments Some of these will include programming tasks as well as thinking and writing.
 Note: With the exception of medical excuses, assignments will be penalized 50% if they are turned in up to two days late, and will receive no credit thereafter.
- **Project (40%)** Students will work on projects of their own choice related to the topic of the class. Grading will be based on both a written paper due at the end of the semester and oral class presentations on each project. Ask guest lecturers for help!





Faculty

Bioinformatics

Alvin Kho, HMS/CHIP Stefano Monti, Broad/MIT Gil Alterovitz, HMS/CHIP Scott Weiss, HMS/BWH Steve Sonis, HSDM/BWH

Clinical Informatics

Aneel Advani, HMS/CHIP Octo Barnett, HMS/MGH Daniel Nigrin, HMS/CHIP John Halamka, HMS/BI John Glaser, Partners Hamish Fraser, HMS/CHIP Stan Finkelstein, Sloan/MIT

Public Health and Biosecurity Kenneth Mandl, HMS/CHIP

Ben Reis, HMS/CHIP Chris Cassa, HMS/CHIP