



Harvard-MIT Division of Health Sciences and Technology

HST.950J: Engineering Biomedical Information: From Bioinformatics to Biosurveillance

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Slightly Controlled Information Exhibitionism in the Genomic Era

A Dangerous (but good?) Idea
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One Motivation

- Key promise of the genome sequencing projects was an era of “personalized medicine
- 99.9% of all human genomes are identical, but the remaining 0.1% produces the entire variability within the human population
- Research projects are collecting phenotypes about patients and correlating them with the precise variations present in the patients’ individual genomes



Points to be Made

- **Conventional methodologies** to obtain large **phenotypically annotated** populations are **not adequately productive**.
- **Concerns about privacy and disclosure of genotype and phenotype data** are **real but addressable** sociologically, technologically and legislatively.
- **Patient/consumer populations** are **ahead of the healthcare and research establishment** in thinking about these challenges.
- Methods for giving **patient control of health data disclosure** provide a mechanism for gathering research data, and
- **Harnessing the volunteerism of our populations** is an important and promising solution today.

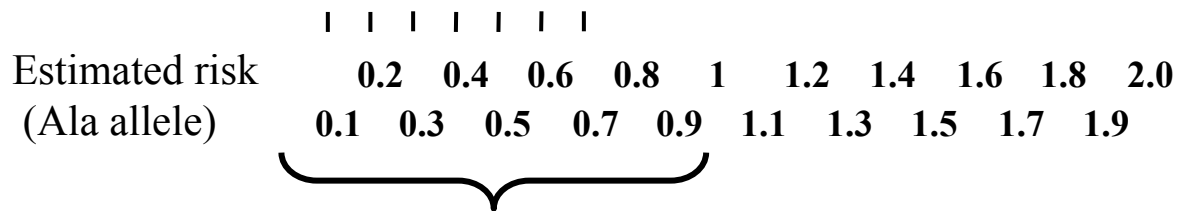
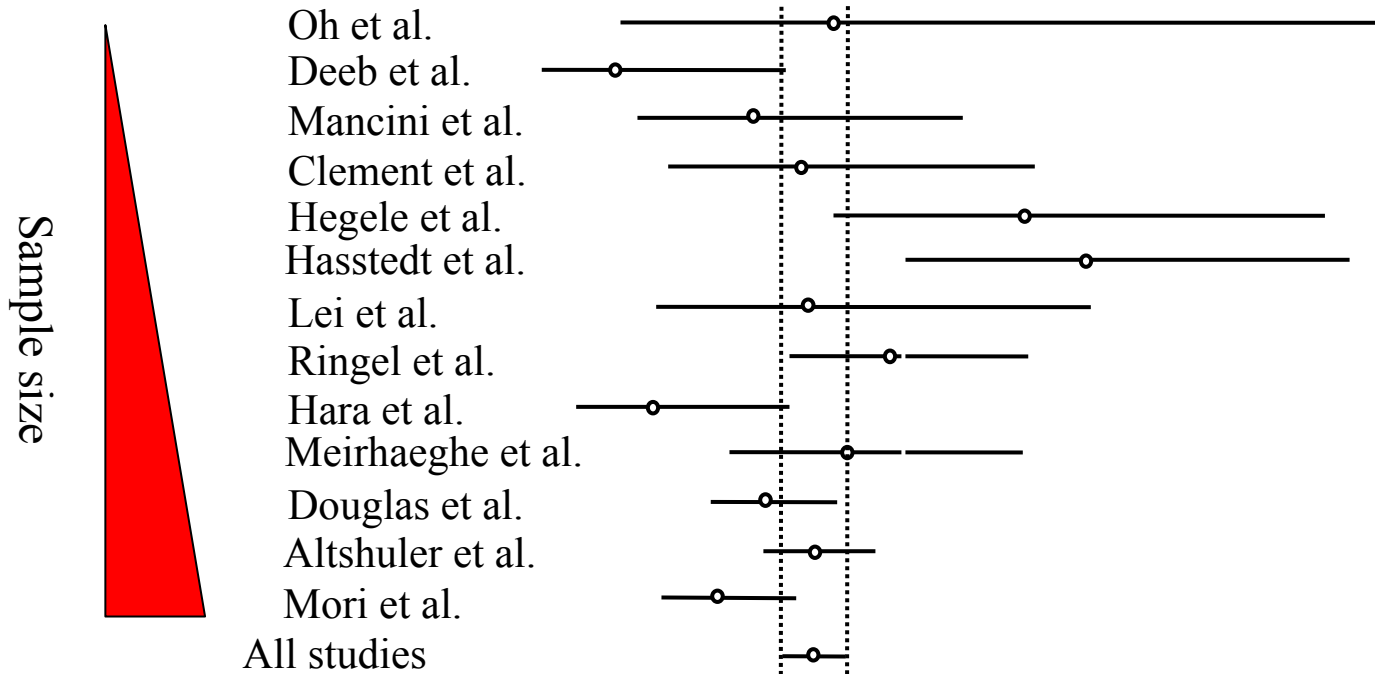


Clinical genomic research depends on large, well-phenotyped populations but...

- There are several challenges
 - ✓ In size of populations required
 - ✓ In obtaining the phenotypes



Example: PPAR γ Pro12Ala and diabetes



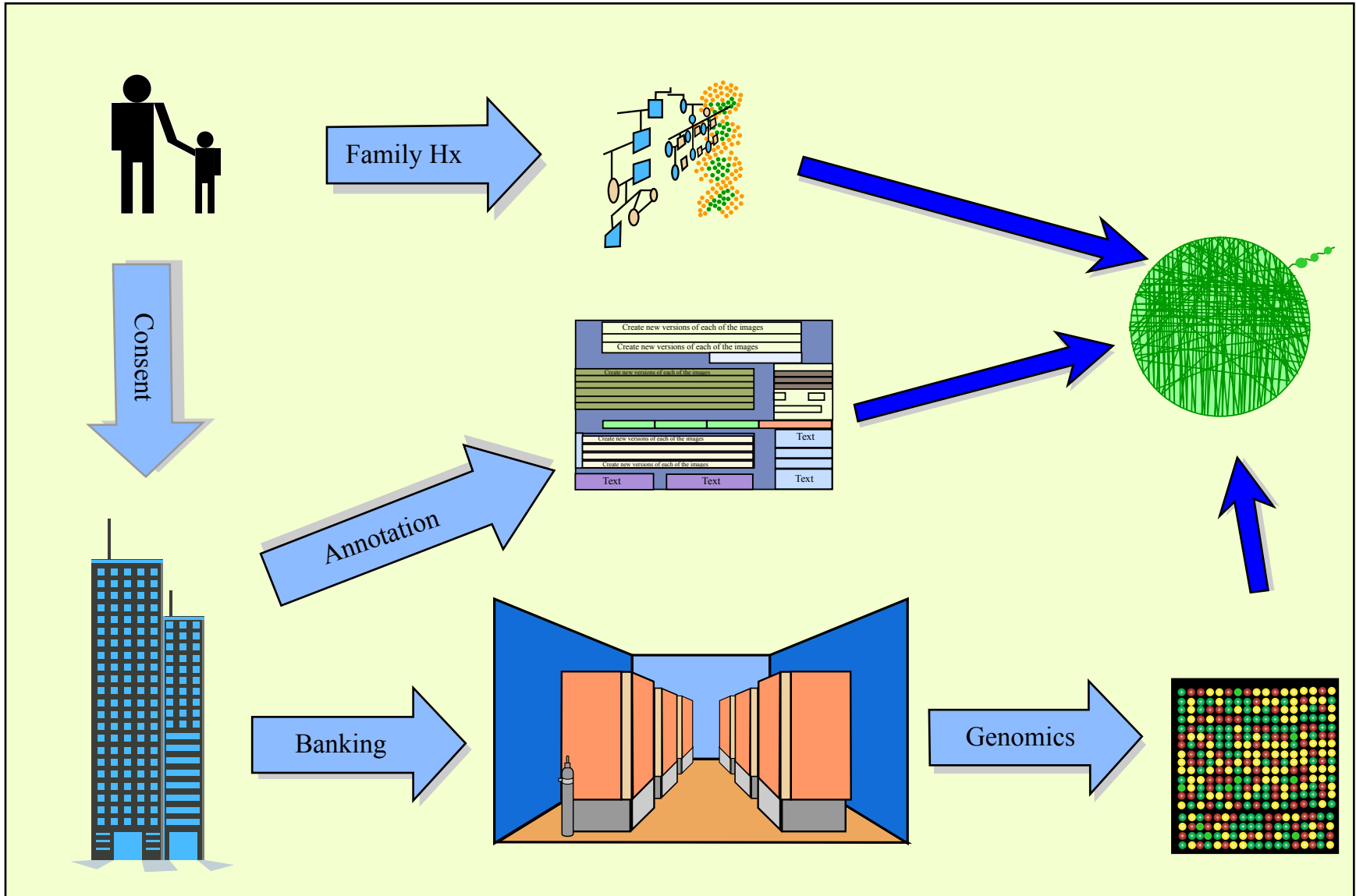
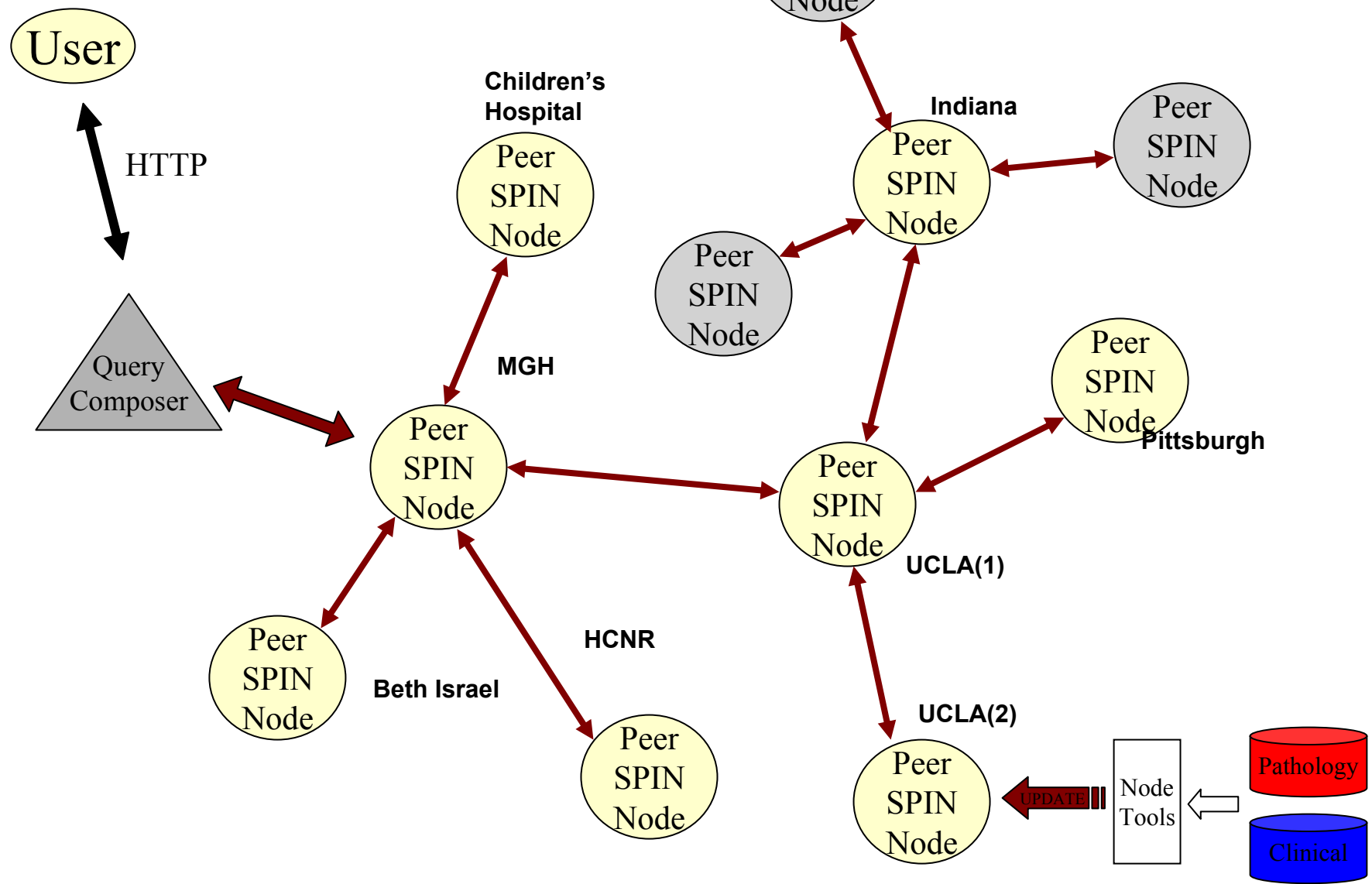


Figure by MIT OCW.





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Risks

- Real Risks
- Cultural-specific perceptions



Risks: There is no perfect anonymity

- Sweeney
- Altman

Figure removed due to copyright reasons.
Please see:

Figure in Lin, Z., A. B. Owen, and R. B. Altman. "Genetics. Genomic research and human subject privacy." *Science* 305, no. 5681 (July 9, 2004): 183.



Risks

- The decision to share genetic data affects other people. ‘
DNA is shared with immediate family members, extended family,
and ethnic groups
- Carelessness.

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The cat is already out of the bag

- Most people are not fully aware of the degree to which their blood samples **can be and are used by the pharmaceutical industry**
 - ✓ When they learn, some wonder if they should participate in the profits that result from their samples.
- Insurance companies, hospitals **routinely share data** for reimbursement and research.



Perceptions of Risk Vary

- Culturally and personally determined.
- Not surprisingly, the level of concern for health privacy shows a distribution (just as many other phenotypes!) in the population



Personal Data Exchange

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Implanted RFID

- ISO standard representation of data
- FDA approved
- 16 bytes of healthcare identifiers
- Implanted in the right triceps
- MRI safe
- Airport security safe
- John's RFID identifier is linked at CareGroup to his medical history



California

Figure removed due to copyright reasons.

Please see:

Strassman, Marc. New bioactivist group calls for universal, voluntary individual genome sequencing for all California residents. Menlo Park, CA. Etopia Media News Networks, September 6, 2004.



Iceland

Figure removed due to copyright reasons.
Please see:

Annas, G. J. "Rules for research on human genetic variation--lessons from Iceland."
N Engl J Med 342, no. 24 (Jun 15, 2000) 1830-3.



US

- An Alpha-1 patient has one of the most obvious genetic discrimination cases in the country ...Patient, who lost a 30-year-old brother to Alpha-1, was identified as having the disorder ... As a preventative treatment her doctor put her on a plasma augmentation therapy used to slow the progression of lung damage. Shortly after receiving the bill for her first treatment, her employer, who self-insured, abruptly fired her, in spite of the fact that she continued to perform her job, in a manner, which her employer had always referred to, as exemplary.



- The problem:
 - Highly mobile patients
 - No formalized data exchange
 - Mobile patients
 - Disparate providers
 - Incompatible medical data storage
 - Lack of unified view of patient record



Standards in Healthcare

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Solving this in a centralized fashion is another major risk



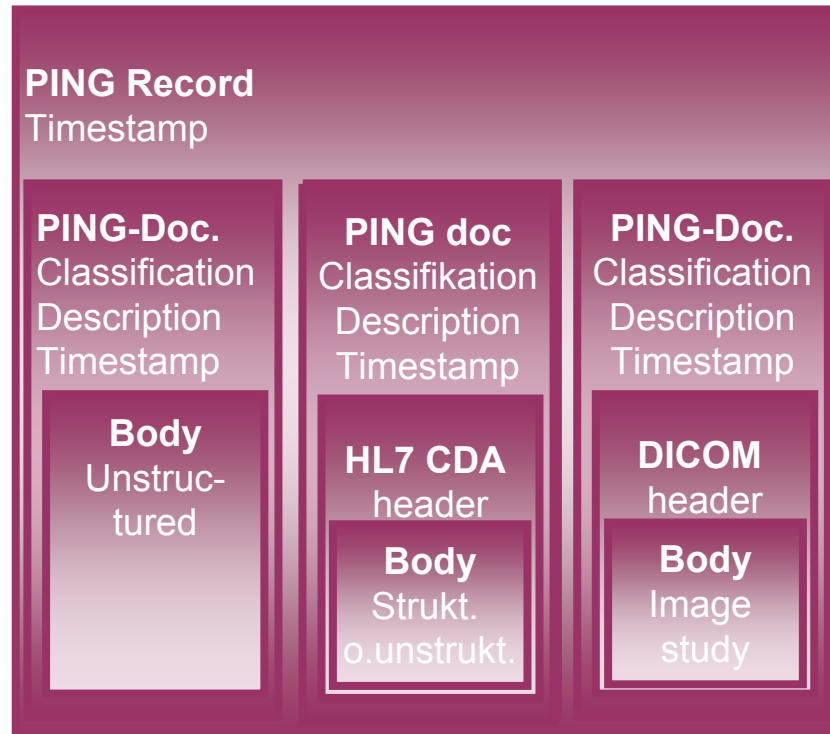
Solution: PING

- Personal Internetnetworked Notary and Guardian
- Personally controlled records
- Ubiquitous
- Open standards
- Open source
- Web interface
- Public key infrastructure
- XML-based data



Genomic Data for a PHR

Example SNP-Test





Header

Body

SNP Test
description

Sequence
information

Interpretation



Overall Proposed solution

- Seek out volunteers who exist on the end of the spectrum near the exhibitionists.
- Potential damages can be outlined, but the subjects will elect to take the risk in the hopes of helping to save lives
- Volunteers would donate DNA sequence data as well as molecular and cellular phenotypic data



Modest (?) Necessary Desiderata

- **Actions by policy makers and legislators** could clear the field for these studies.
 - ✓ **illegal** to act upon the health information contained in research databases would send a simple message to those considering mining these data:
 - ☞ **you can do it, but you are not allowed**
- Researchers who curate genetic databases should have some protection of activities,
 - ✓ as long as they follow an **agreed set of operating guidelines**.
 - ✓ Guidelines **would not guarantee privacy** (a standard that is too high),
 - ✓ **guarantee reasonable physical security** and data quality.
- Patients should be granted explicit control over the disclosure process.



Real test

- Create pilot studies to test the feasibility of asking patients to accept lower levels of privacy guarantees.
- Require the development of new consents.
- Studies would also limit the disclosed data to include only those subsets of information that the patient is willing to share.
- At the same time, policy makers would provide some protections for the patients and the researchers



In Summary

- Why should you fear this idea?
- Why should you rejoice that it's beginning to be implemented?
- What should you tell your mom about it?
- What's your most interesting discovery?
 - ✓ Oncogenesis recapitulates ontogenesis
- What's your most recent discovery?
 - ✓ It's all over after 40.