The promise and challenge of individualized health

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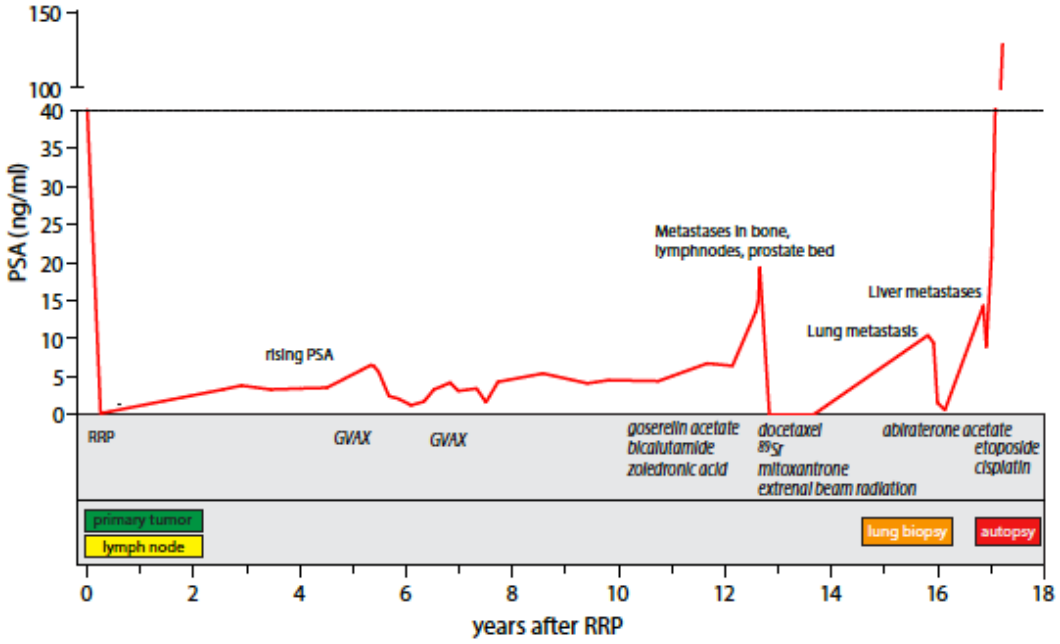
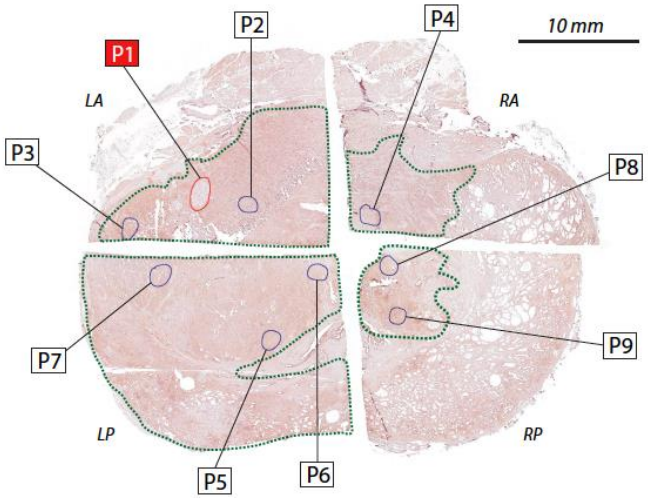
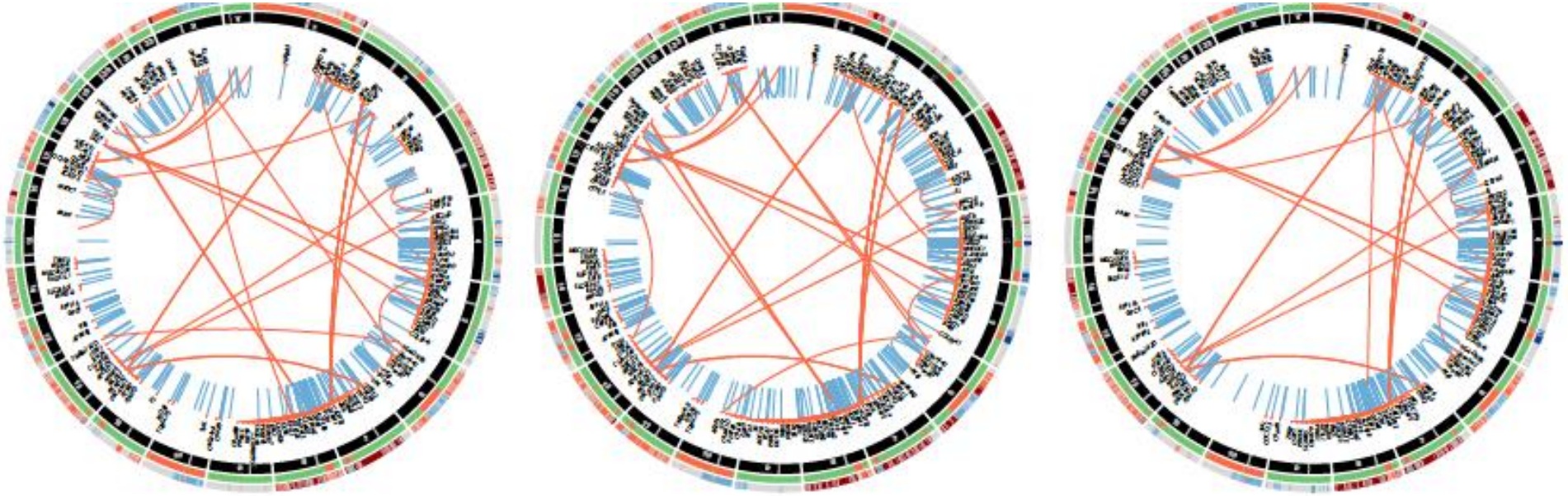




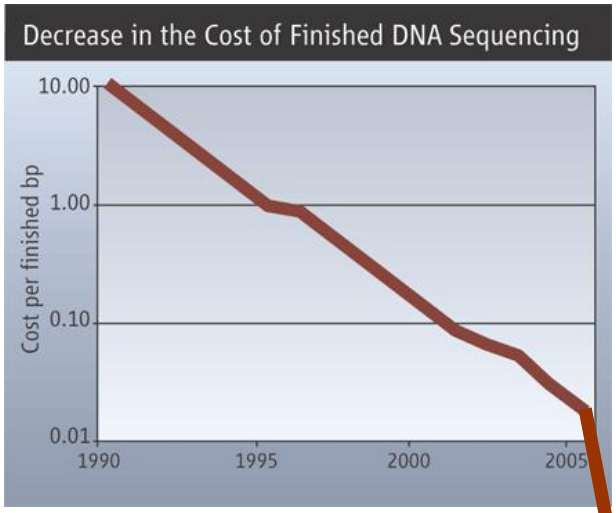


Haffner et al., JCI, 2013, in press.

Opportunities for individualized health

- Characterize an individual-specific constellation of genomic, epigenomic and phenotypic alterations
  - Improve outcomes by tailoring therapy
  - Avoid harm by withholding interventions that may be unnecessary and/or harmful to a given person
- The massive bet: that such an approach can allow improvements in health outcomes while at the same time improving efficiency of health care delivery.

Why the sudden buzz?



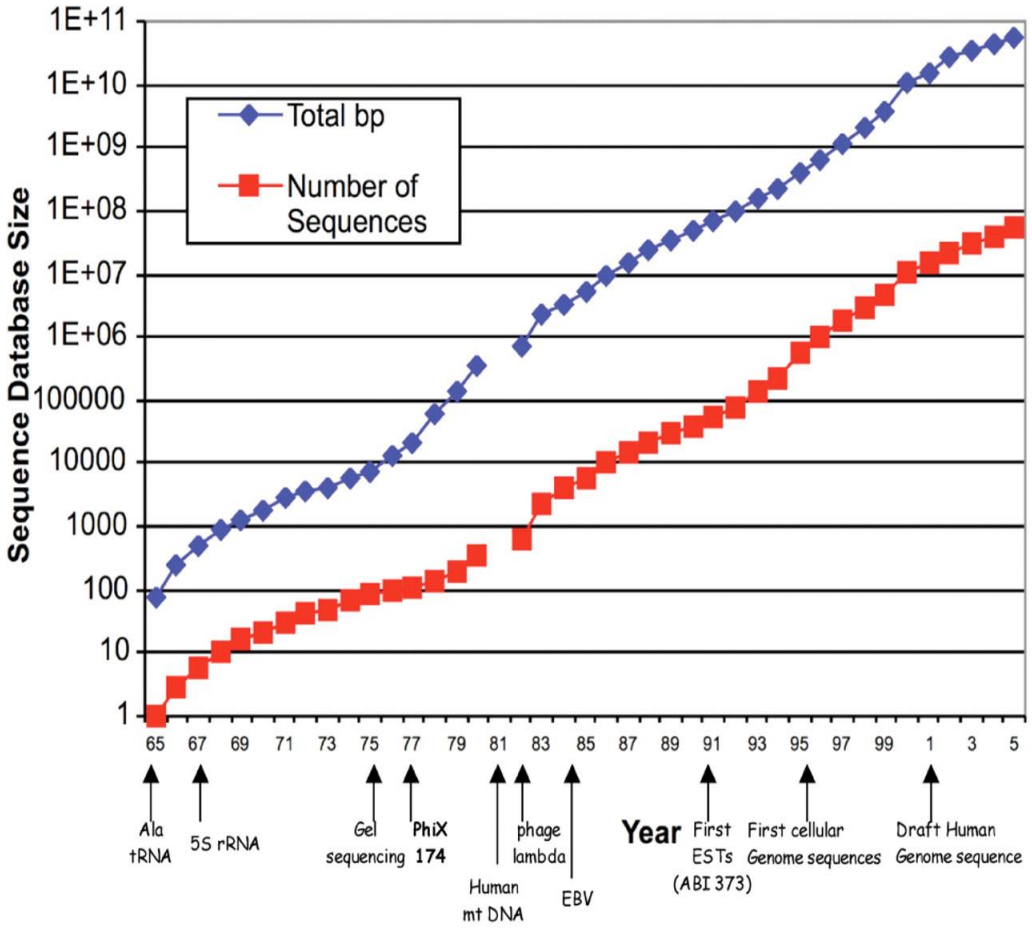
Service RF, Science, 2006

2007: $10^{-4}/bp

2009: $10^{-5}/bp

2011: $10^{-6}/bp

… still going …



Hutchison CA, Nucl Acids Res, 2007

Challenges

- Massive amounts of data:
  - Whole genome sequencing, Whole exome sequencing, RNA-seq, DNA methylation, -omics
  - ~300 GB to 1 TB per sample depending on scope and comfort level of what to keep.
- Longitudinal analysis: many samples per individual
- Overall, in the near future, a given individual may therefore accumulate up to dozens of TB of data through the course of a medical encounter

Challenges cont'd

- Massive number of people for whom this could be deployed in the future
  - SKCCC: 8,000 new diagnoses per year
  - Hopkins Health System covers >1.5 million people
- Data storage:
  - Where?
  - How?
  - Backup?
  - Integration with medical record
  - Integration with research databases
  - What types of new database architectures are needed?

Challenges Cont'd

- Data interpretation:
  - If we simply accumulate this data, how will it help achieve our goal?
  - How can we integrate and process these new genomics and other data to inform unmet clinical needs?
  - Data mining, pattern recognition, machine learning, directed hypothesis testing?
  - I/O needs for the computing infrastructure to move the massive data in and out?

Challenges Cont'd

- Individualized health needs advancement of individualized research!
- How can we craft novel clinical research paradigms to address opportunities in individualized health
- What will the computing needs of such clinical research be?
- Once opportunities are realized, how do we deploy to the community?

Funding

- Short term?
- Intermediate term.. Seeing the path towards sustainability
- Long term: true opportunity for sustainability:
  - If our health system can deliver more efficient health care as a result of the advances in individualized health, then they can save money and theoretically re-invest that in developing sustainable infrastructure!!