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?

2007: $10^{-4}$/bp
2009: $10^{-5}$/bp
2011: $10^{-6}$/bp

... still going ...

Service RF, Science, 2006
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!!