BIOLOGICAL DATA SCIENCE

November 5–November 8, 2014

#biodata14

Anne Carpenter Broad Institute, @DrAnneCarpenter

Michael Schatz Cold Spring Harbor Laboratory, @mike_schatz

> Matt Wood Amazon Web Services, @mza





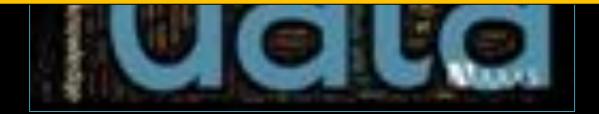


@JasonWilliamsNY

Charla Lambert

Data are interesting, but do not answer any of the thousands of possible questions:

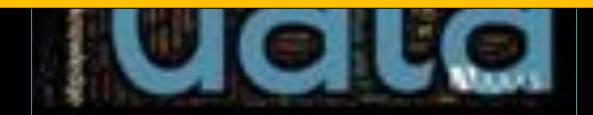
- How does my genome compare to yours?
- How does expression or methylation or chromatin change?
- What diseases are you at risk for, what pathogens have you been exposed to, and what medicines should we give you?

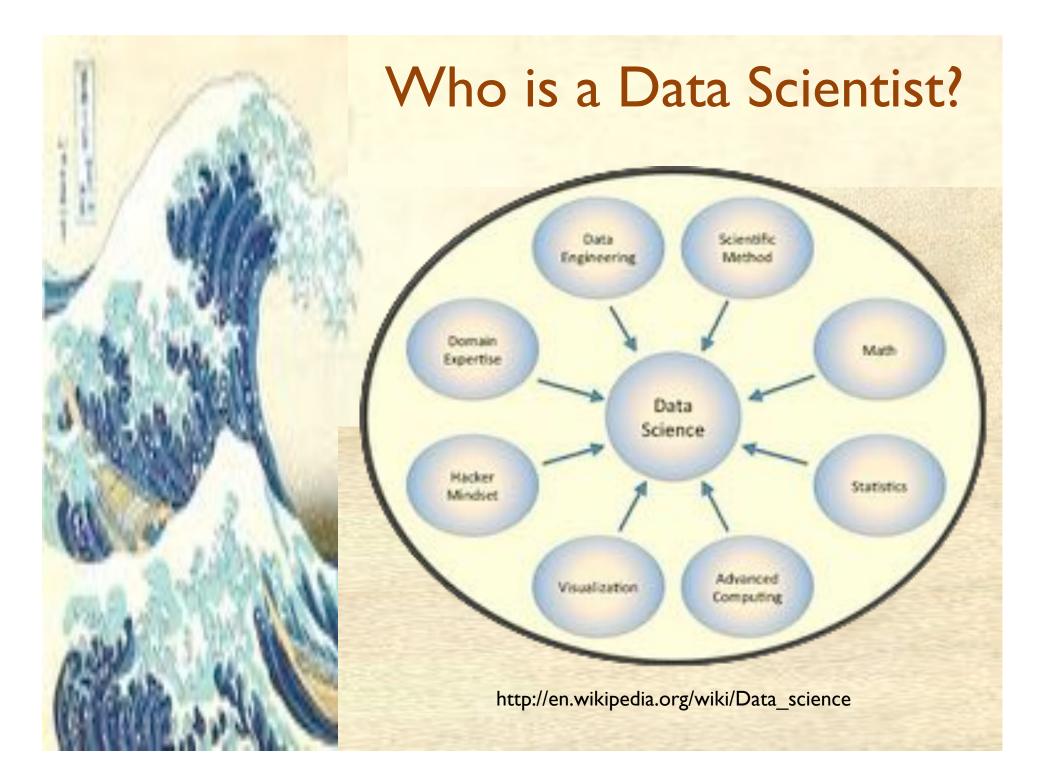


Data are interesting, but do not answer any of the thousands of possible questions:

- How does my genome compare to yours?
- How does expression or methylation or chromatin change?
- What diseases are you at risk for, what pathogens have you been exposed to, and what medicines should we give you?

Who will answer those questions? How will they do it?





Biological Data



1 Illumina X-Ten sequences a genome every 30 minutes ~100k whole human genomes sequenced Worldwide capacity exceeds 25 Pbp/year

How much is a petabyte?

| Unit | Size |
|----------|-----------------------|
| Byte | |
| Kilobyte | 1,000 |
| Megabyte | 1,000,000 |
| Gigabyte | 1,000,000,000 |
| Terabyte | I,000,000,000,000 |
| Petabyte | 1,000,000,000,000,000 |

*Technically a kilobyte is 2^{10} and a petabyte is 2^{50}

How much is a petabyte?



100 GB / Genome 4.7GB / DVD ~20 DVDs / Genome

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10,000 Genomes

=

1PB Data 200,000 DVDs



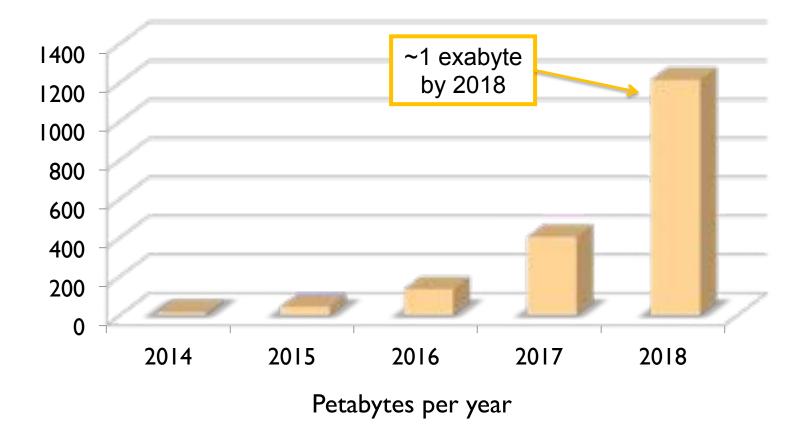
787 feet of DVDs ~1/6 of a mile tall



500 2 TB drives \$500k

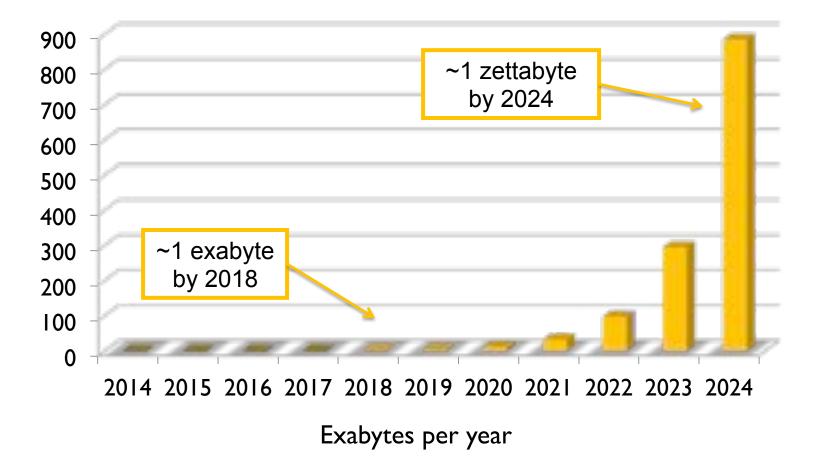
DNA Data Tsunami

Current world-wide sequencing capacity is growing at $\sim 3x$ per year!



DNA Data Tsunami

Current world-wide sequencing capacity is growing at $\sim 3x$ per year!



How much is a zettabyte?

| Unit | Size |
|-----------|-------------------------------|
| Byte | |
| Kilobyte | I,000 |
| Megabyte | 1,000,000 |
| Gigabyte | I,000,000,000 |
| Terabyte | I,000,000,000,000 |
| Petabyte | 1,000,000,000,000,000 |
| Exabyte | I,000,000,000,000,000,000 |
| Zettabyte | 1,000,000,000,000,000,000,000 |

How much is a zettabyte?

100 GB / Genome 4.7GB / DVD ~20 DVDs / Genome

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10,000,000,000 Genomes



1ZB Data 200,000,000,000 DVDs







150,000 miles of DVDs $\sim \frac{1}{2}$ distance to moon

Both currently ~100Pb And growing exponentially

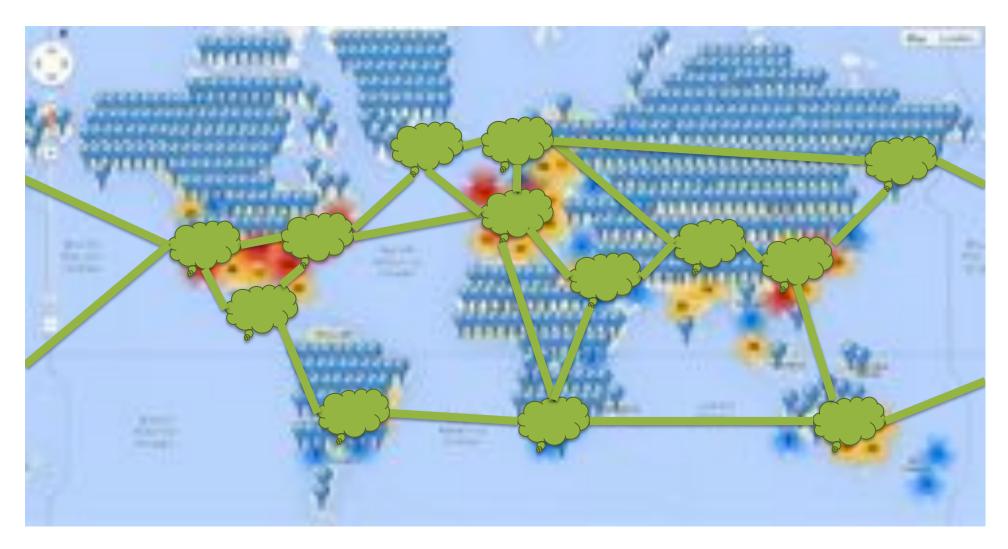
Sequencing Centers 2014



Next Generation Genomics: World Map of High-throughput Sequencers

http://omicsmaps.com

Informatics Centers 2014

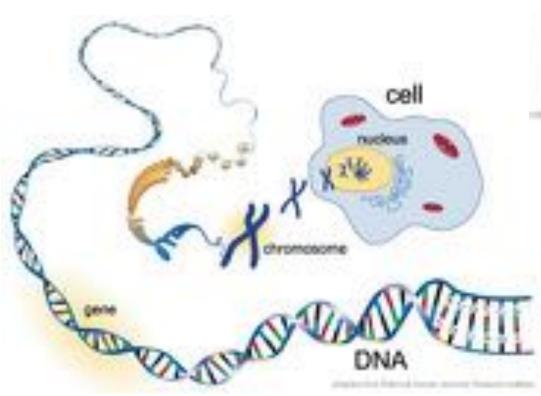


The DNA Data Deluge

Schatz, MC and Langmead, B (2013) IEEE Spectrum. July, 2013

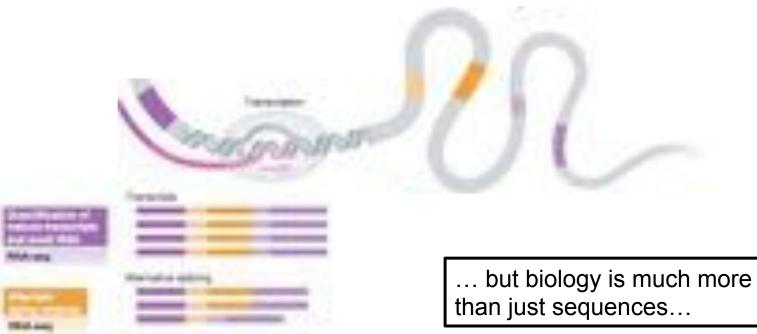
Biological Data

Much of the capacity is used to sequence genomes (or exomes) of individuals...





... but biology is much more than just genomes...



Soon et al., Molecular Systems Biology, 2013



http://www.slideshare.net/pebourne/wiki-mania080914

Biological Data Science

ARTICLE

An integrated encyclopedia of DNA elements in the human genome

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LETTER

A cis-regulatory map of the Drosophila genome

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CLUSTAL W: improving the sensitivity of progressive multiple sequence alignment through sequence weighting, position-specific gap penalties and weight matrix choice

ALC: NOT THE OWNER.

Jule D Thompson, Desmand G/Hggire* and Tuby J.Otteon* Surgroup Molecular Bulkgy Lateratory, Postach 10209, Meyertotatees 1, 0-60112 resolutions. Germany

Penalmel 20/y 12, 1984; Revised and Asserted Department 28, 1994

ABSTRACT

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The contribution of de novo coding mutations to autism spectrum disorder

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A map of rice genome variation reveals the origin of cultivated rice

Barden Tanayi, Yan Daniya, Hanghao Wali, A. Xuan Mund, Y., Aman Mund, Yuan Ziao, Yuan Xiao, Yuan Xiao, Jiao Yuan, J. Shanayi, Y. Yuan Xiao, Yuan Yuan, Y. Yuan, Yuan, Y. Shanayi, Yanayi, Y. Shanayi, Yanayi, Yuan Yuan, Yu

ARTICLE

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THURSES:

A framework for human microbiome research

MapRobuce: Simplified Data Processing on Large Clusters

Artisty Dean and Bargay Chamanas

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Create Inc.

Abstract

Highster is a preparate mole and as your and implementation for processing and generating large data and. Easts specify a map benchm that processes a implement pair or processes a set of intermediate large value pairs and a maker function that surgets all intermediant values associated with the same intermediant key. Many real world tasks are supersafile in this model, as itsner in the paints

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Privacy & Security

Identifying Personal Genomes by Surname Inference

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Predicting Social Security numbers from public data

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information aloud an individual's place and date of lartit can be exploited to preside his or her Contal Genurity number (50%) Using only publicly available information, we assumed a contactuation between individually 50% and their birth data and found that he promps colorests the secondation alread states and found that he promps (50%). The information alread states and found that he provide (50%). The information are reade presiden by the public mailability of the field becarity indeconstruction's fourth Waster tits and the nodespressi accessibility of personal information from multiple secures, such as data broken or profiles or social ratentities of the contactor resolutions are profiles of social ratentities of the contactor resolutions around allows and sequences of the contactor resolutions around alloys the sequences of the contactor resolutions and social private stores of the contactor resolutions and social private the stores of the contactor resolutions and social private the stores of the contactor resolutions and social private the stores of the contactor resolutions and social private social stores of the contactor resolutions and social private the tasebar DNO, The KBA specify provides information almost the process through which A/Na, GNa, and DNa are insued (1). A/No ant currently sengred based on the specific of the training address pervalues in the MIP application form DEMERGOLUM! (1) Low-population white and outstate U.S. presentence or admented I A/N each, whereas other mater admented are of A/Na (for instance, an individual applying from a signable which A/Na (for instance, an individual applying from a specific for Size York more may be using out of 15 pointies for 3 500 Eight). Written each MIA with, CPH are assigned in a process bit neurosciencement order between (ii) and 90 (KMIRER), 2001 (1). Buth the sets of A/N assigned to different sizes and the sequence in terms and the applying from the sequence of the sequence.

Extrapolating to the U.S. living population, this would imply the potential identification of millions of SSNs for individuals whose birth data were available. Such findings highlight the hidden privacy costs of widespread information dissemination and the complex interactions among multiple data sources in modern information economies (11), underscoring the role of public records as breeder documents (12) of more sensitive data.

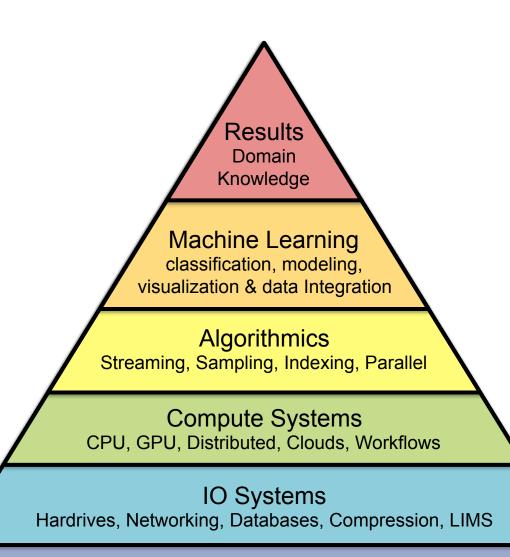
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How?

- Integration of multiple data types
- Massively scalable
- Geographically distributed
- Computationally flexible
- Tolerate noise, errors, and artifacts
- Support data exploration and ambiguity
- Reliable, reproducible, and secure



Data Science Technologies



Sensors & Metadata Sequencers, Microscopy, Imaging, Mass spec, Metadata & Ontologies



BIOLOGICAL DATA SCIENCE



| Wednesday | 7:30 pm 8:00 pm | Introduction Keynote Speaker |
|-----------|--------------------|-----------------------------------|
| Thursday | 9:00 am | 1 Data and Data Mining I |
| Thursday | 1:30 pm | 2 Data and Data Mining II |
| Thursday | 3:00 pm | 3 Poster Session I |
| Thursday | 4:30 pm | Wine and Cheese Party |
| Thursday | 7:30 pm | 4 Compute Infrastructure |
| Friday | 9:00 am | 5 Algorithmics |
| Friday | 1:30 pm | 6 Diclogical Software |
| Friday | 4:30 pm | Master Locture |
| Friday | 5:30 pm | 7 Poster Session II and Cocktails |
| Friday | 7:00 pm | Benquet |
| Saturday | 9:00 am | # Human Biology |

Master Lecture



"Homomorphic encryption as a tool to preserve privacy in genomic computation"

Friday @ 4:30pm

Kristin Lauter, Ph.D. Microsoft Research

Schedule Change



Saturday Morning: Human Biology

Mark Gerstein will present first in the session

Plan to break for lunch at 11:40am instead of noon

Eric Perakslis, Ph.D. Harvard Medical School

Keynote Introduction



Ph.D. in CS from the Univ. of Colorado at Boulder in 1982

Member of the NAS and the American Academy of Arts and Sciences; Fellow of AAAS and AAAI

Research combines mathematics, computer science, and molecular biology

- Pioneered the use of HMMs and other machine learning techniques for analyzing biological sequences
- Major efforts in the human genome project, and developing the UCSC Genome Browser
- Recently focused on understanding and fighting cancer; sharing of data through the Global Alliance for Genomics and Health

David Haussler, Ph.D.

Distinguished Professor of Biomolecular Engineering at UCSC Investigator, Howard Hughes Medical Institute Scientific Director, UC Santa Cruz Genomics Institute Thank you! @mike_schatz / #biodata 14