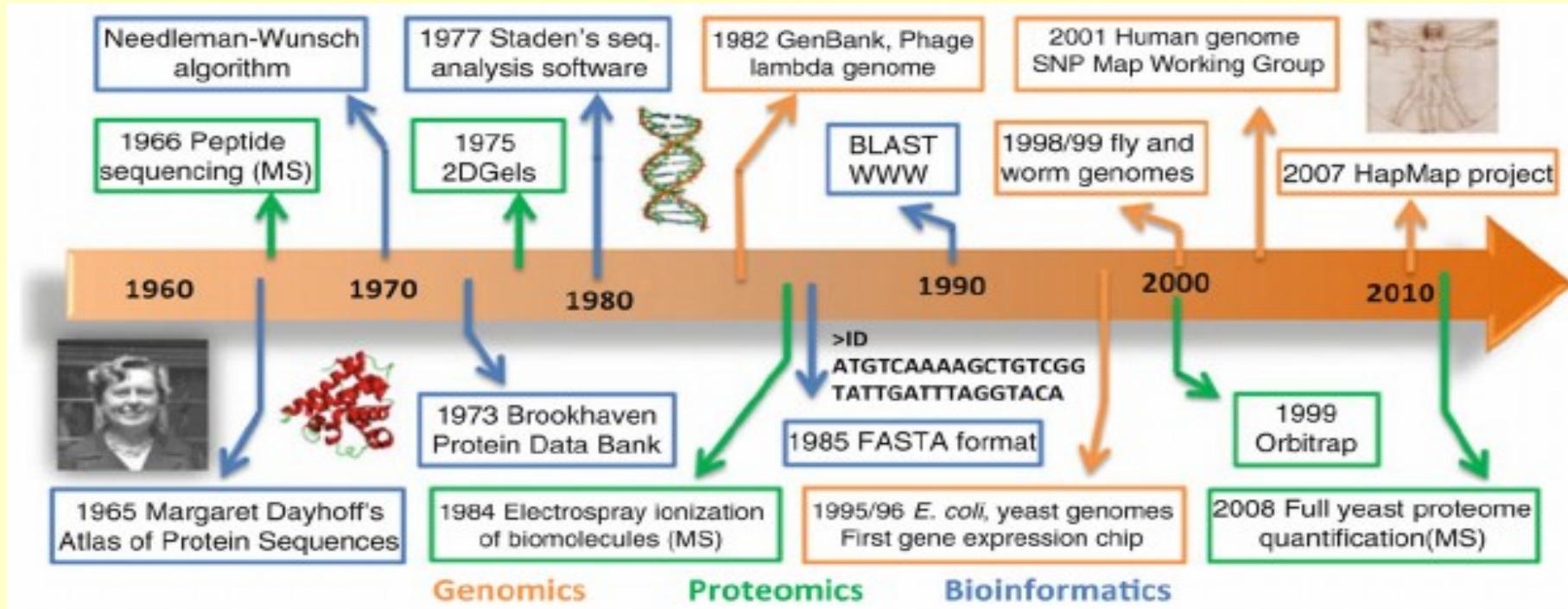


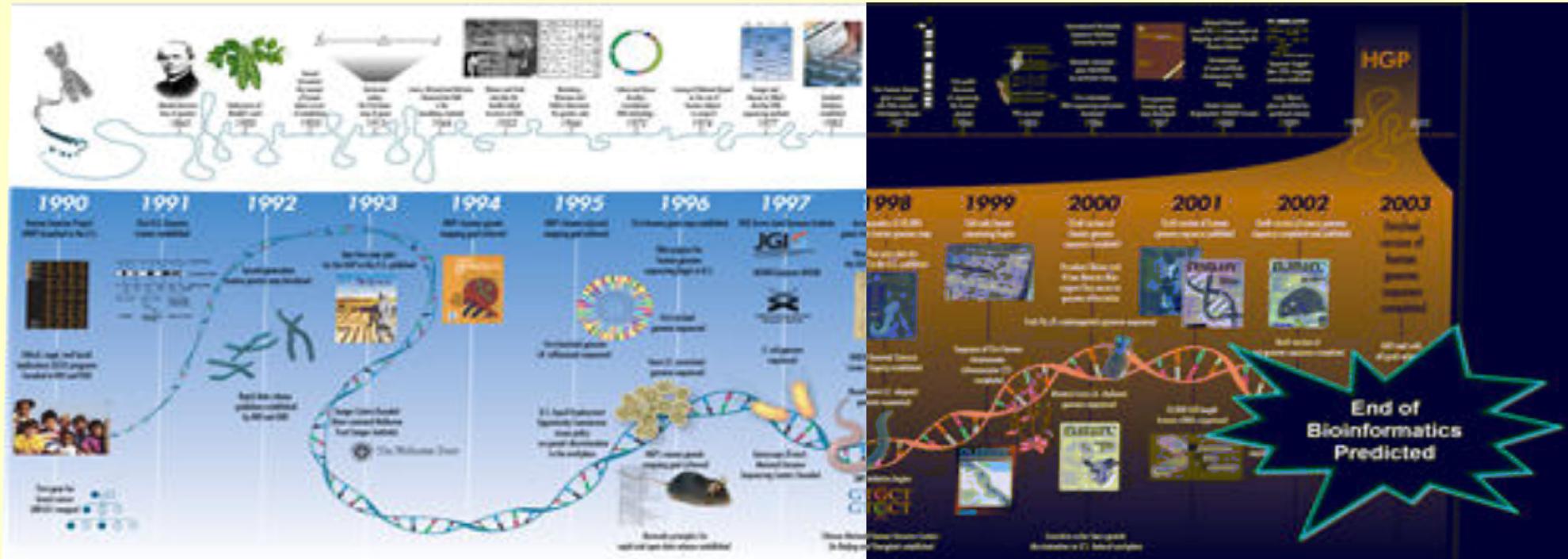
# Impacts of Bioinformatics

# Landmarks in Bioinformatics



Source: Comparative Interaction Networks: Bridging Genotype to Phenotype  
Adv Exp Med Biol. 2012 ; 751: 139–156. doi:10.1007/978-1-4614-3567-9\_7

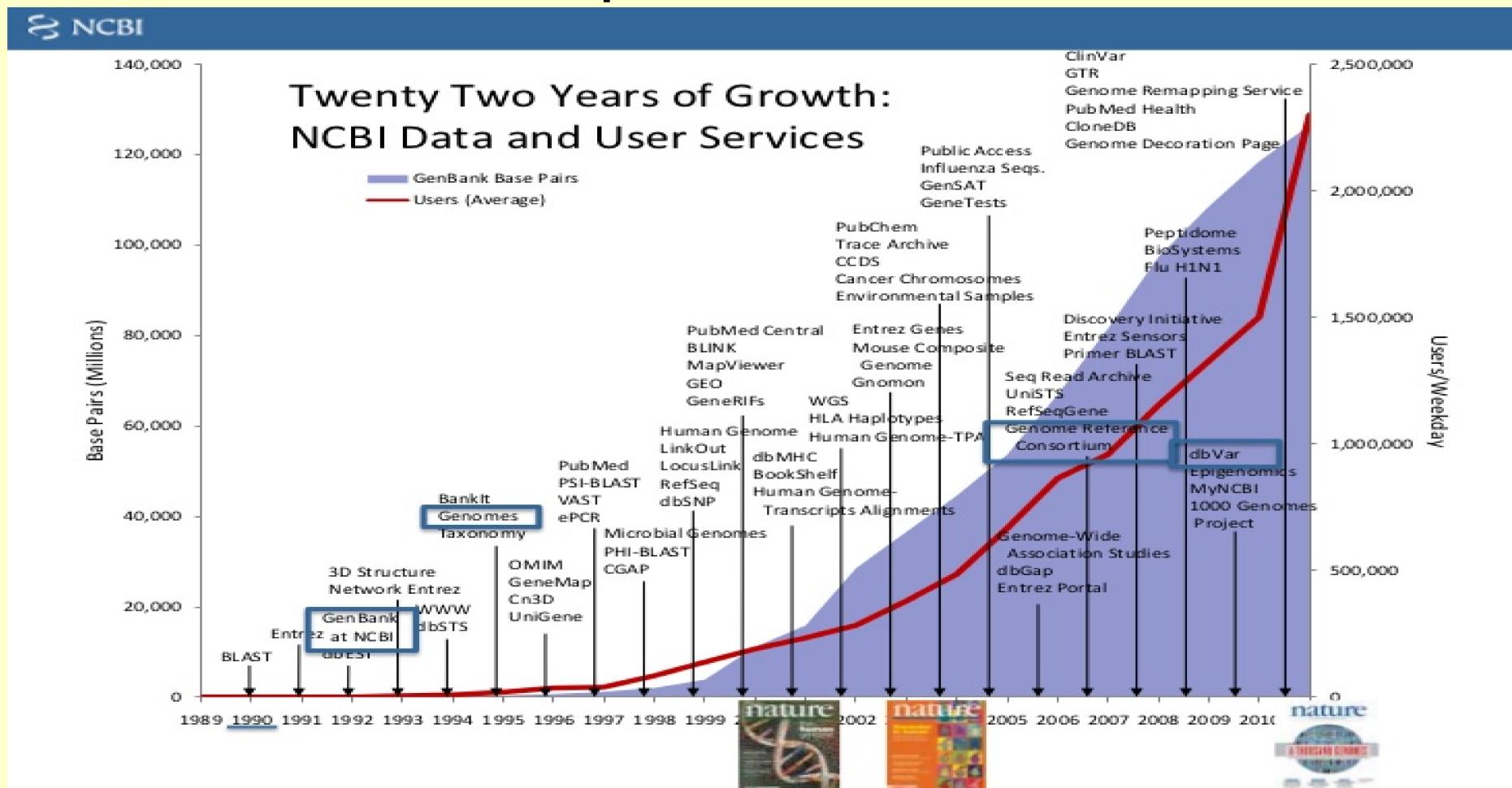
# Bioinformatics here to stay?



Lincoln Stein's prediction, Feb 2003\_ **Bioinformatics: Gone in 2012**

In 2008 he published a correction: **Bioinformatics: alive and kicking**  
Genome Biology 2008, 9:114 (doi:10.1186/gb-2008-9-12-114)

# Growth of sequence data and users



# Socially, what drives the process?

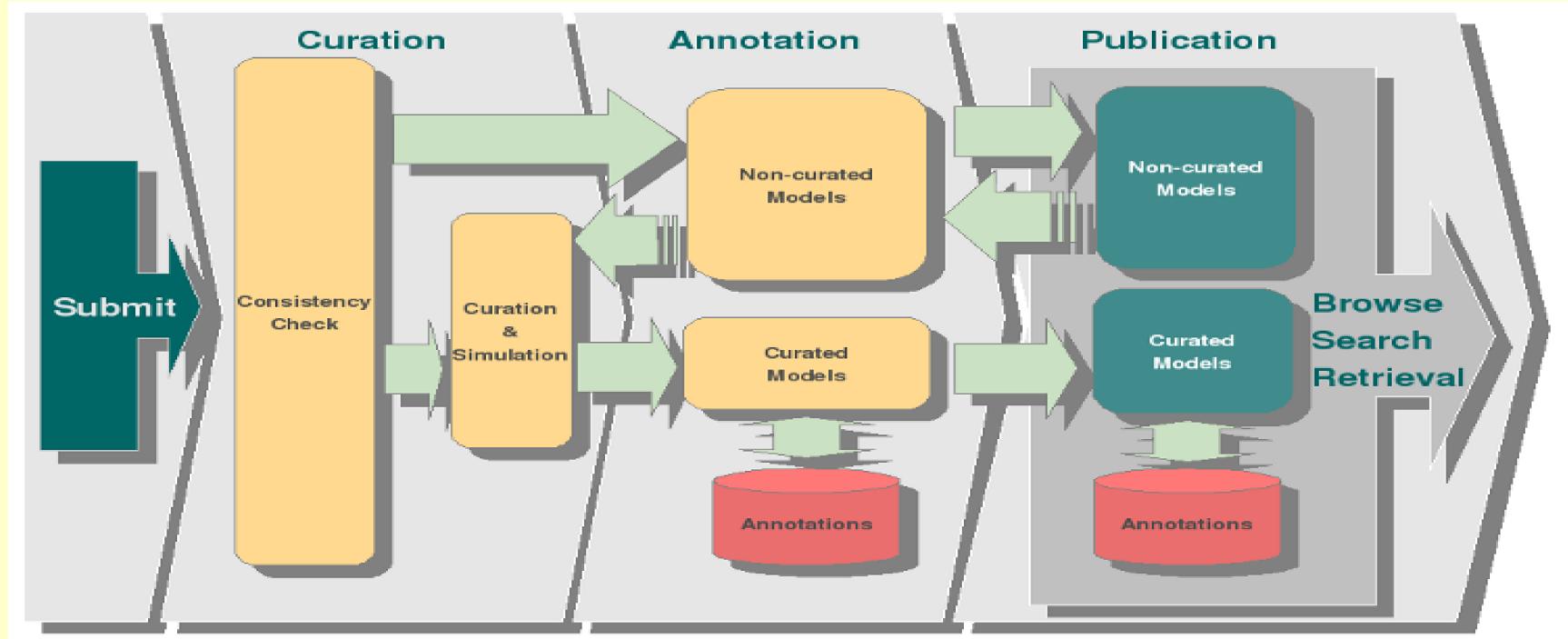
Some of the priority areas of investment that have triggered developments in Bioinformatics

- 1985 Biotechnology
- 2000 Personalised Medicine, Genomes & Health.
- 2010 Environmental Issues. GMOs & Food controls.
- 2015 Precision Medicine & Systems Biology
- 2020 ... Immunotherapy. Ecosystem (remediation?)

# Where is Bioinformatics heading?

- Coping with the Omic information deluge and growing service needs is a big challenge in itself
- But it is relatively small compared with what we anticipate will come in what concerns the need to
  - better interpret research data (recognise conserved domain architectures, use systems level reasoning in Biology and Medicine)
  - make more accurate predictions (inference)
  - understand complex genetic traits in health and disease

# From data to curated models

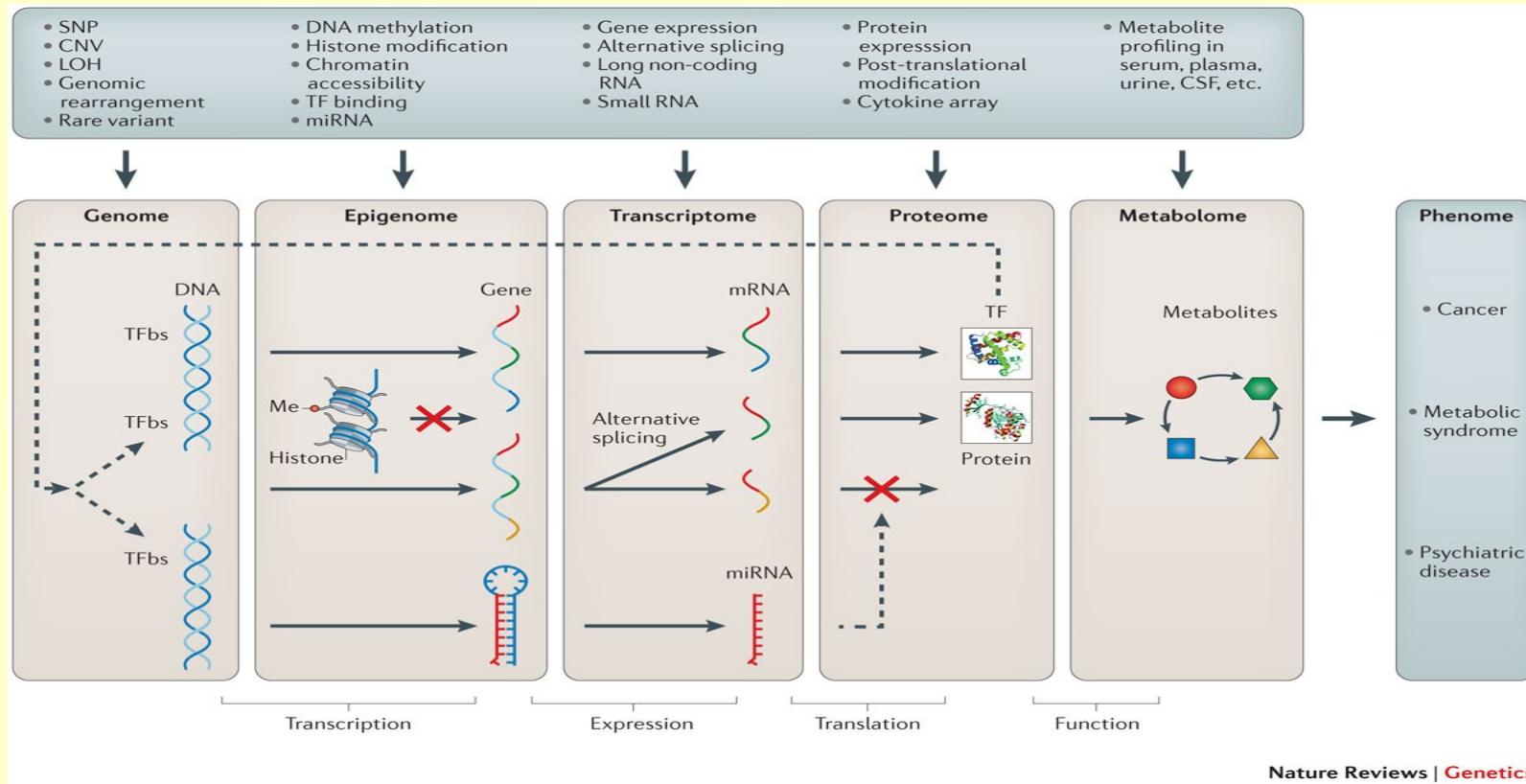


Curation and annotation pipeline in the BioModels Database

Source: European Bioinformatics Institute

<http://www.ebi.ac.uk/biomodels-main/develop>

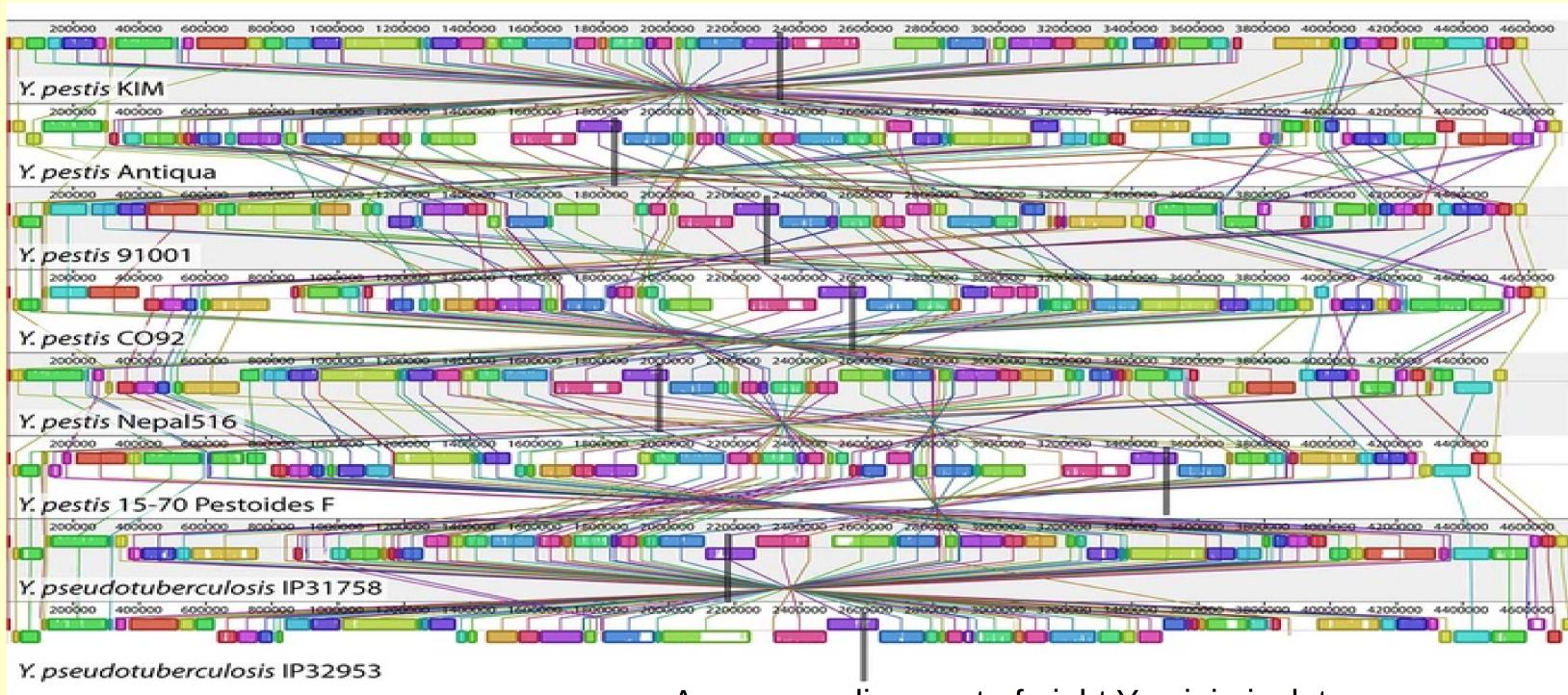
# Gains in knowledge via integration



Source: **Methods of integrating data to uncover genotype–phenotype interactions**

Marylyn D. Ritchie, Emily R. Holzinger, Ruowang Li, Sarah A. Pendergrass & Dokyoon Kim  
Nature Reviews Genetics 16, 85–97 (2015) doi:10.1038/nrg3868

# Deep understanding to fight disease



A genome alignment of eight *Yersinia* isolates.  
(source: PLOS Genetics on Wikimedia Commons).

## Deep learning meets genome biology

An interview with Brendan Frey about realizing new possibilities in genomic medicine.

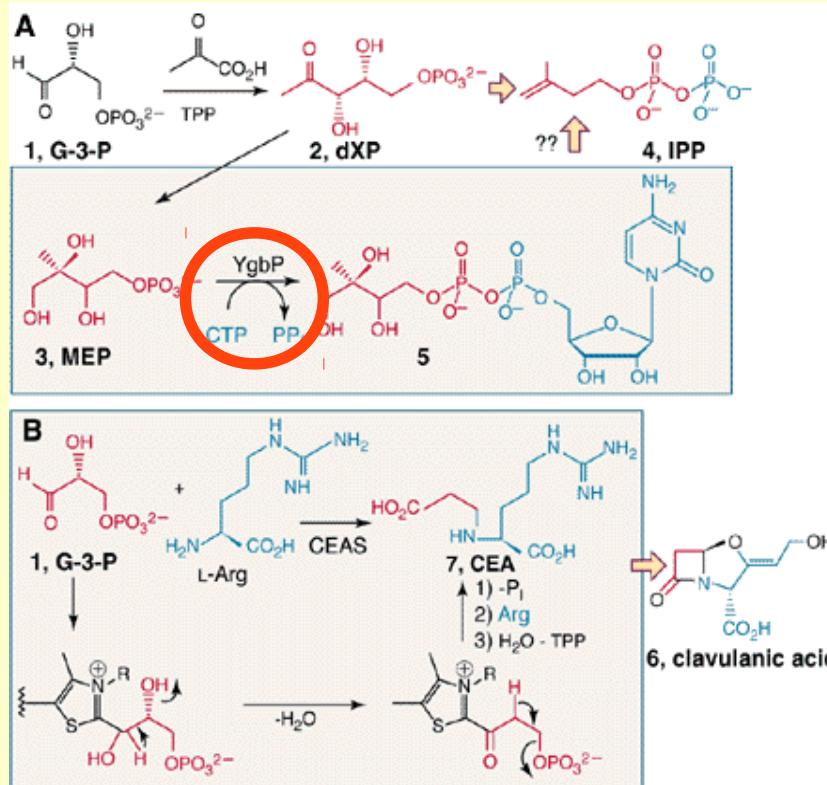
<https://www.oreilly.com/ideas/deep-learning-meets-genome-biology>

# Using large datasets

Using large datasets together with powerful analytics and machine learning allows for generating and testing hypotheses in unprecedented ways

It does require specific skills but the most serious bottleneck is still.. human!

# Ex.1 Arguably difficult without Bioinformatics

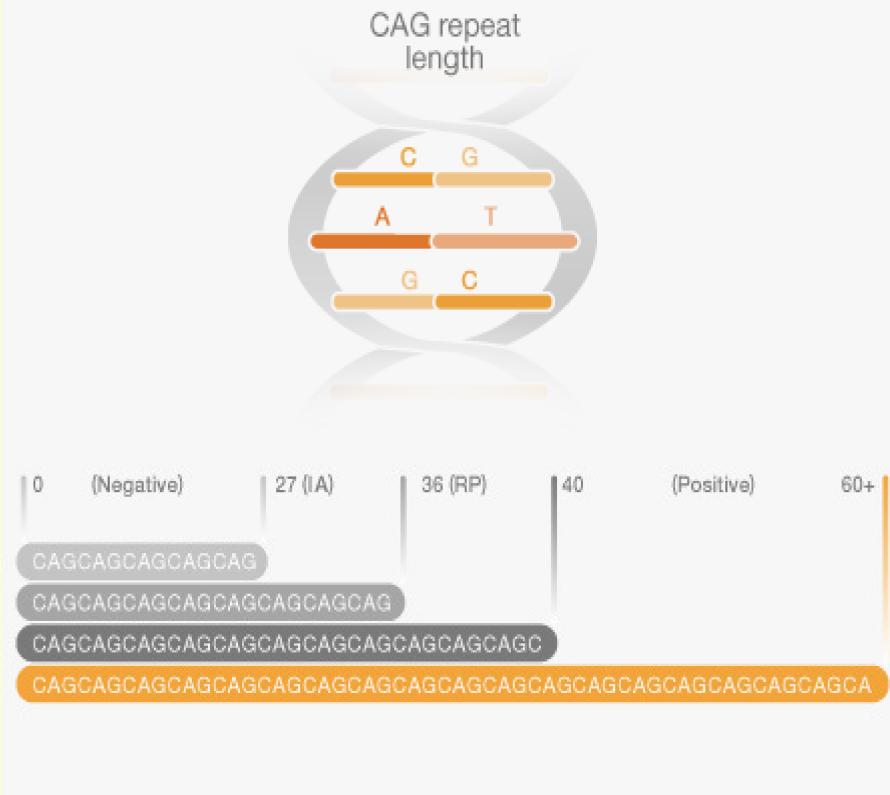


Science. 2000 Feb 4;287(5454):818-9.  
**Perspectives: biosynthetic pathways.**  
**Biosynthesis meets bioinformatics.**  
Cane DE

Clavulanic Acid is a beta-Lactamase inhibitor that helps penicillin-like antibiotics fight resistant bacteria.

**YgbP** was found in natural organisms by searching sequence databases for its genomic Signature. This led to dramatic reductions in production costs.

# Ex.2 Arguably difficult without Bioinformatics



The number of CAG repeats in the HD (Huntington Disease) gene on Chr4, discovered in 1993, determines disease severity.

This relationship was drawn by classifying sequencing reads in large quantities and comparing clinical observations in a large scale.

This has allowed for designing genetic tests that are useful in family genetic counselling, to reduce the chances of anticipation, as the repeats go longer in successive generations.

# The human resource issues

## Education

- The need for Bioinformatics-aware biomedical professionals is horizontal. All Life Science related professionals need to know at least the basics.

## Training

- Bioinformatics tools and data resources evolve rapidly and their correct usage requires skills. We need trained people that can use their skills autonomously.

# Motivation to carry-on

- This is why teaching introductory Bioinformatics in a large scale becomes so important.
- Should motivate you to deepen your knowledge in this course and acquire specific skills by attending training sessions.
- It did motivate all of us to collaborate in bringing the IBT course to life!