Bioinformatics and its Applications

Setia Pramana

Educational Background

- Hasselt Universiteit, Belgium, MSc in Applied Statistics 2005-2006.
- Hasselt Universiteit, Belgium, MSc in Biostatistics 2006-2007.
- Hasselt Universiteit, Belgium, PhD Statistical Bioinformatics, 2007-2011.

Educational Background

Medical Epidemiology And Biostatistics Dept. Karolinska Institutet, Sweden, Postdoctoral, 2011-2014

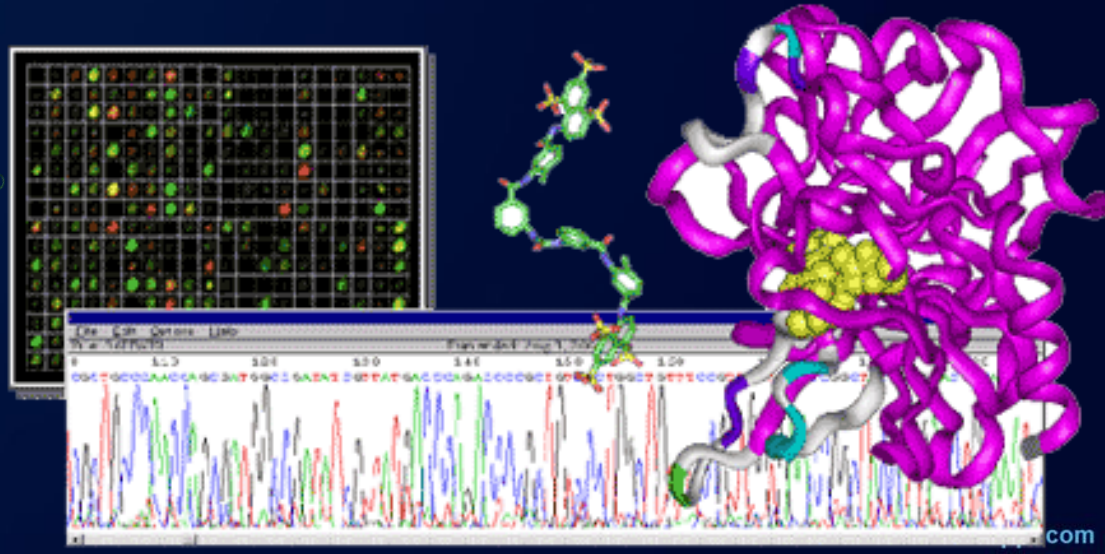
Now?

- Head of PPPM Politeknik Statistika STIS
- Adjunct Faculty at FKUI
- Board Member AIDI, ISI, MABBI.

What is Bioinformatics?

What is BioInformatics:

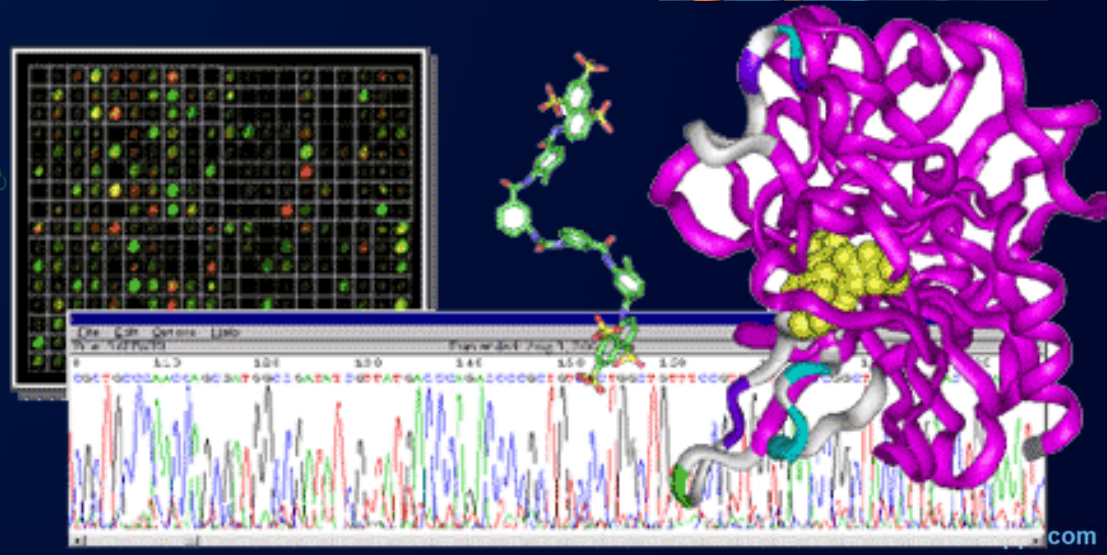
Bioinformatics is a field at the interface between biology, mathematics, computer science, and is rapidly becoming central discipline in medicine, life sciences, drug discovery, proteomics and systems-biology .



What is BioInformatics:

BioInformatics:
Bringing order and structure to chaos

proteomics and systems-biology .



What is Bioinformatics ?

Bioinformatics is the use of computers for the acquisition, management, and analysis of biological information.

It incorporates elements of molecular biology, computational biology, database computing, and the Internet…

… bioinformatics is clearly a multi-disciplinary field including:
computer systems management
networking, database design, computer programming,
molecular biology

From Using Computers for Molecular Biology, Stuart M. Brown, PhD, RCR, NYU Medical Center

What is Bioinformatics ?

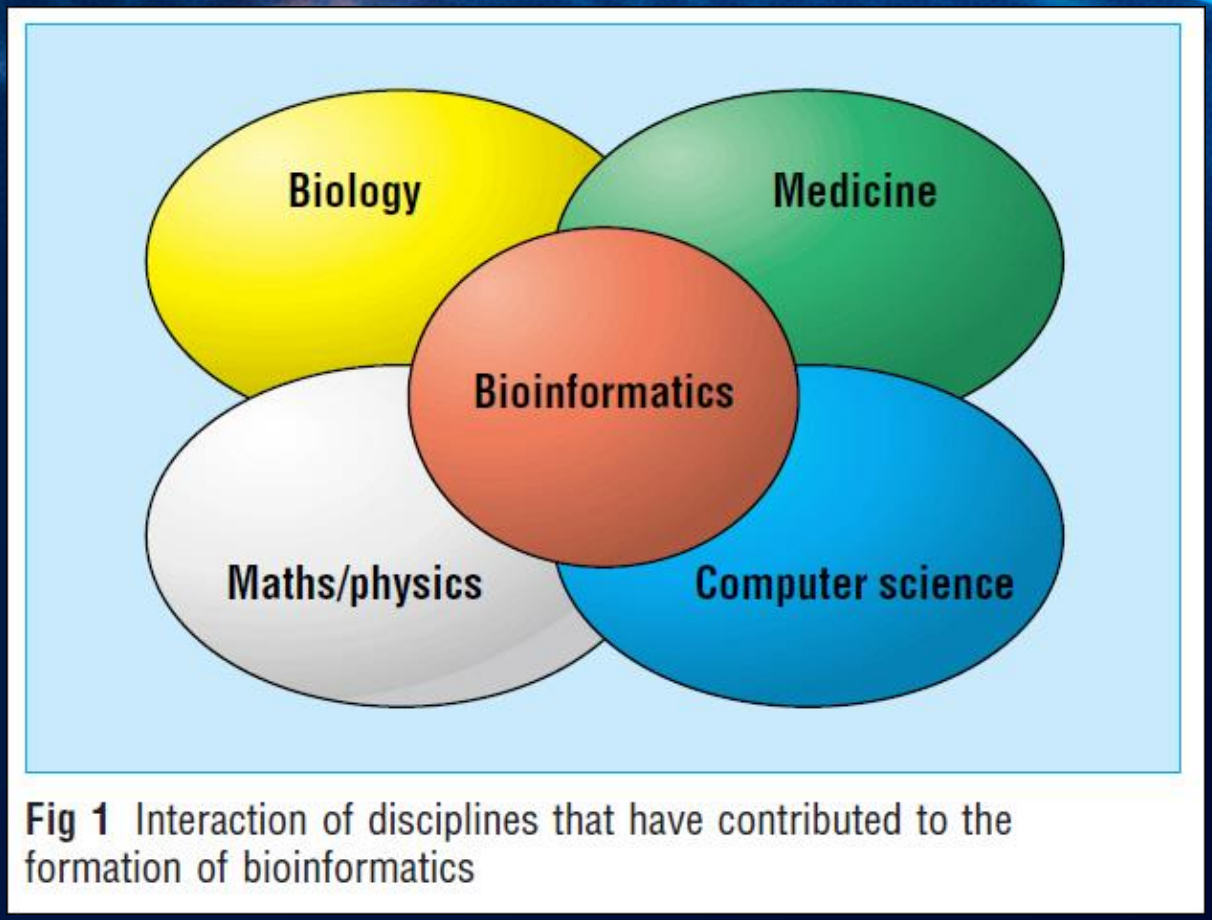


Fig 1 Interaction of disciplines that have contributed to the formation of bioinformatics

Bioinformatics is a multifaceted discipline combining many scientific fields including computational biology, statistics, mathematics, molecular biology and genetics (Fenstermacher, 2005, p. 440).

…from Bayat (2002), p 1018.

What is bioinformatics?

- Sequence analysis
  - Geneticists/ molecular biologists analyse genome sequence information to understand disease processes
- Molecular modeling
  - Crystallographers/ biochemists design drugs using computer-aided tools
- Phylogeny/evolution
  - Geneticists obtain information about the evolution of organisms by looking for similarities in gene sequences
- Ecology and population studies
  - Bioinformatics is used to handle large amounts of data obtained in population studies
- Medical informatics
  - Personalised medicine

General Aims in Bioinformatics

- Organizes data in a way that allows researchers to access existing information and to submit new entries as they are produced
- Develops tools and resources that aid in the analysis of data
- Use these tools to analyze the data and interpret the results in a biologically meaningful manner

Bioinformatics Data

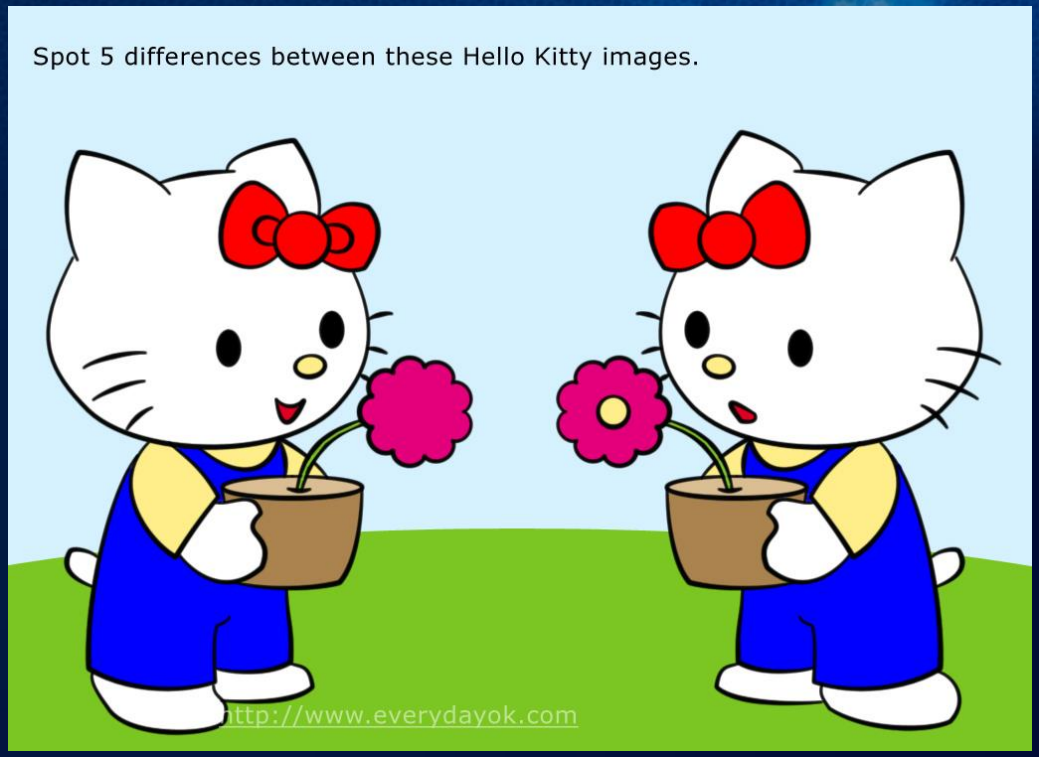
- Bioinformatics deals with
  - DNA and protein sequences
  - Gene expression (microarray)
  - Raw data collected from field or laboratory experiment, Clinical Data from hospital, etc
  - Images, virtual models, Software
  - Articles from literature and databases of citations
- Each type exist in different formats and structure

Bioinformatics Data

- More data in when we attempt to
  - determine structure of data
  - relate to transcriptomics, proteomics
  - relate to structure, physiology
  - relate to disease
  - relate to variation
- Automated discovery, experiments

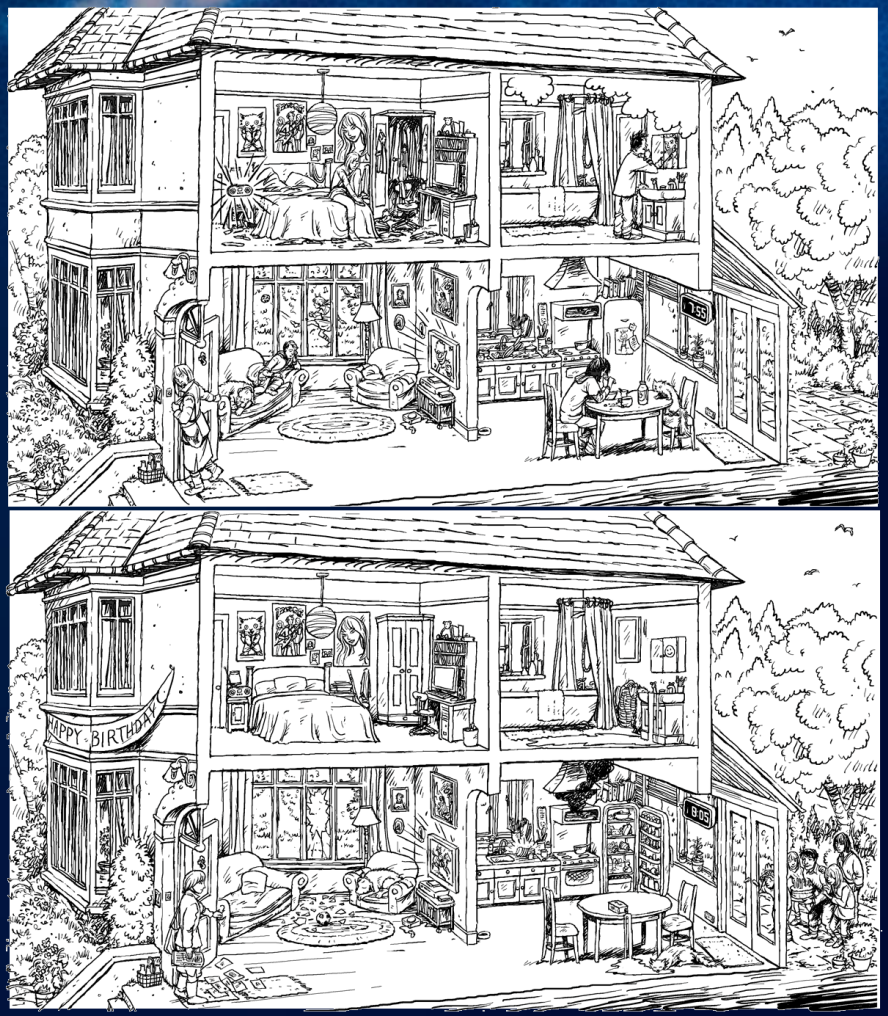
BioInformatics Goal 1: Find the difference

Find the 5 differences



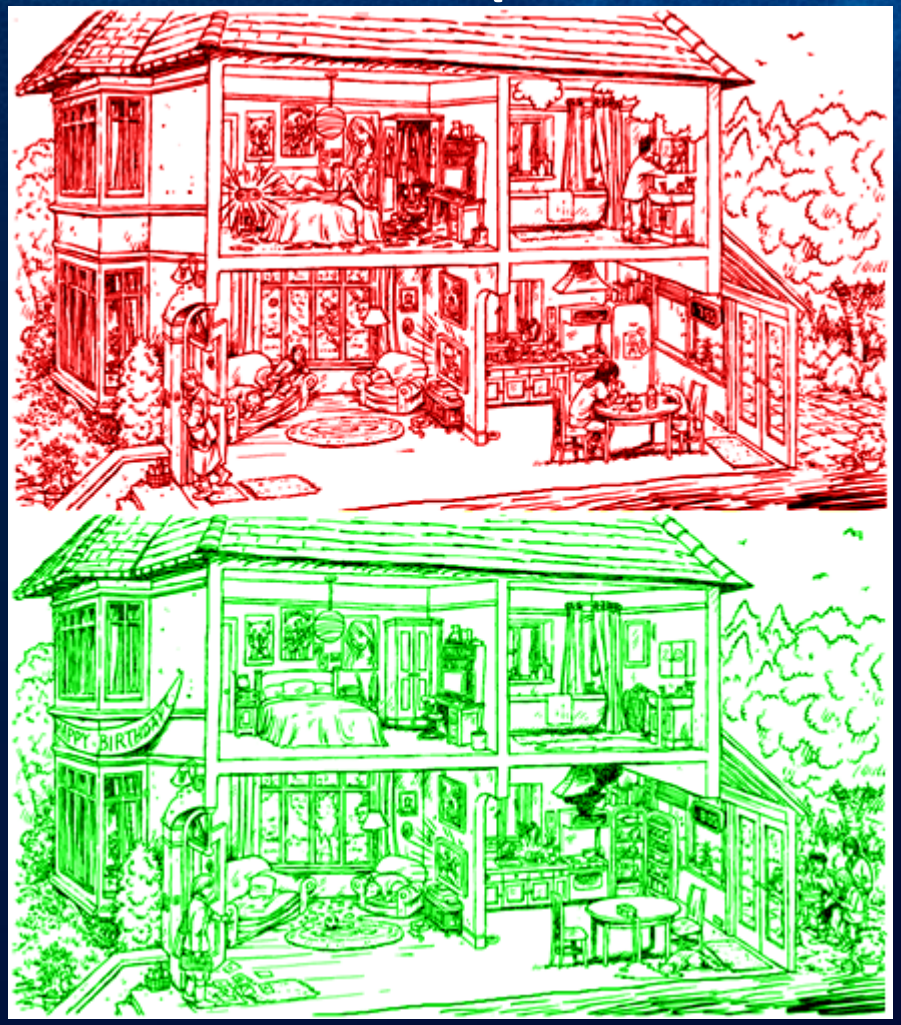
BioInformatics Goal 1: Find the difference

Find the differences (unknown number)



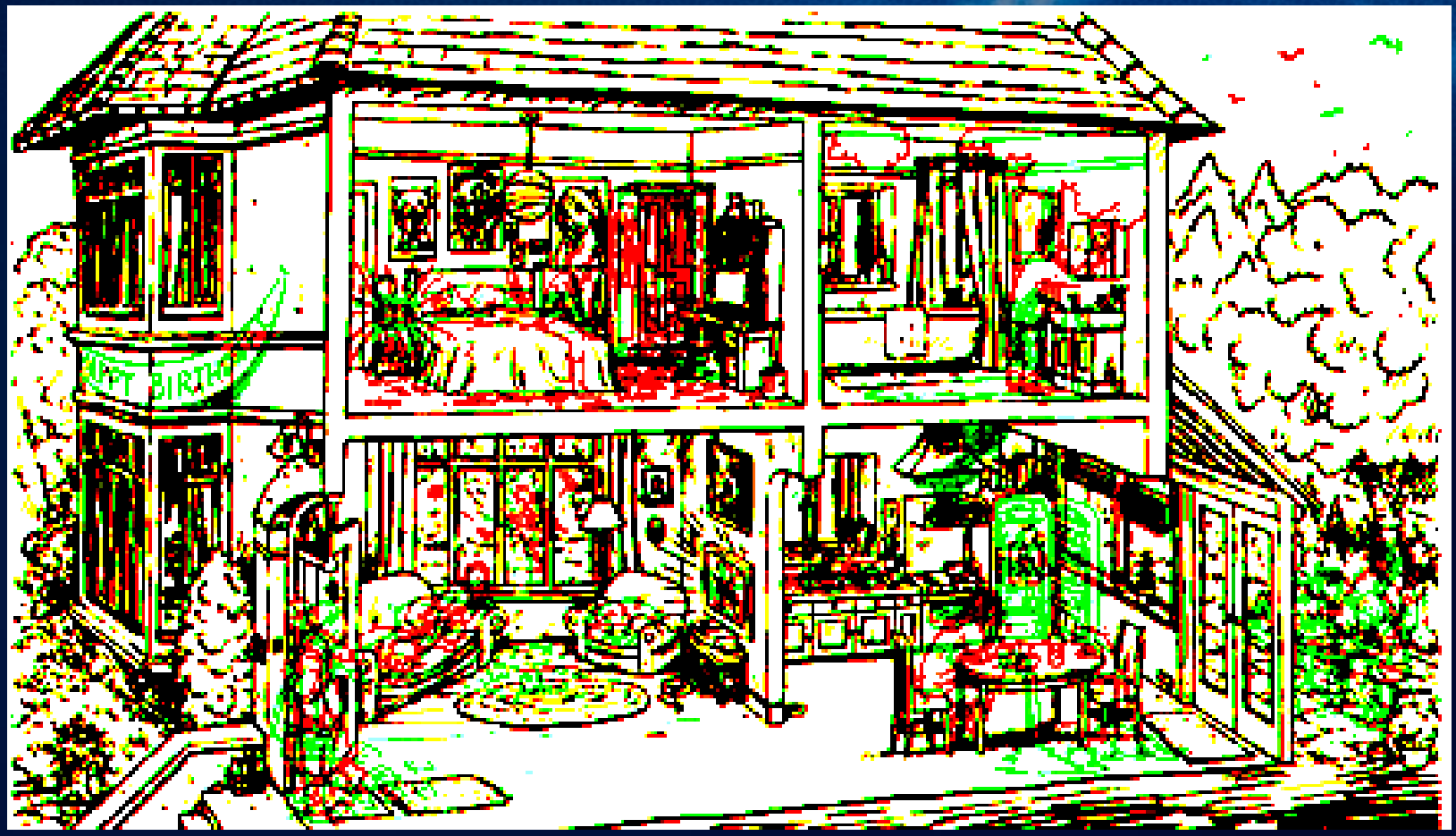
Goal 1: Find the difference
The BioInformatics Approach

Find the differences (unknown number)

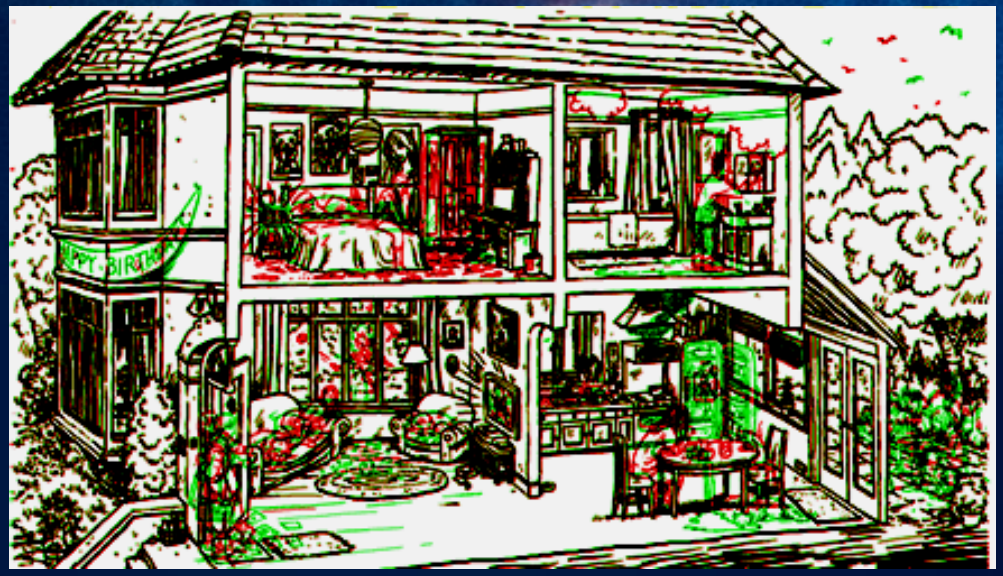


Goal 1: Find the difference
The BioInformatics Approach

Problem solved!



BioInformatics: Find the difference Relationship to Life-Science



Green: healthy patient

Red: Sick patient

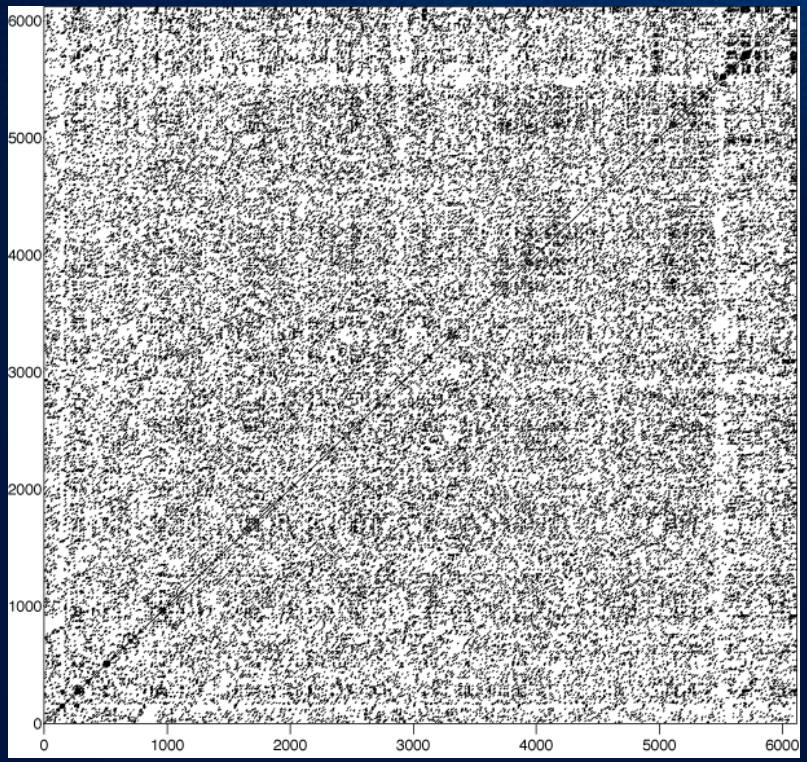
1. Sometimes things (proteins/genes) are only present in the healthy patient (green) *Example lactose intolerant people miss the enzyme to break down lactose.*

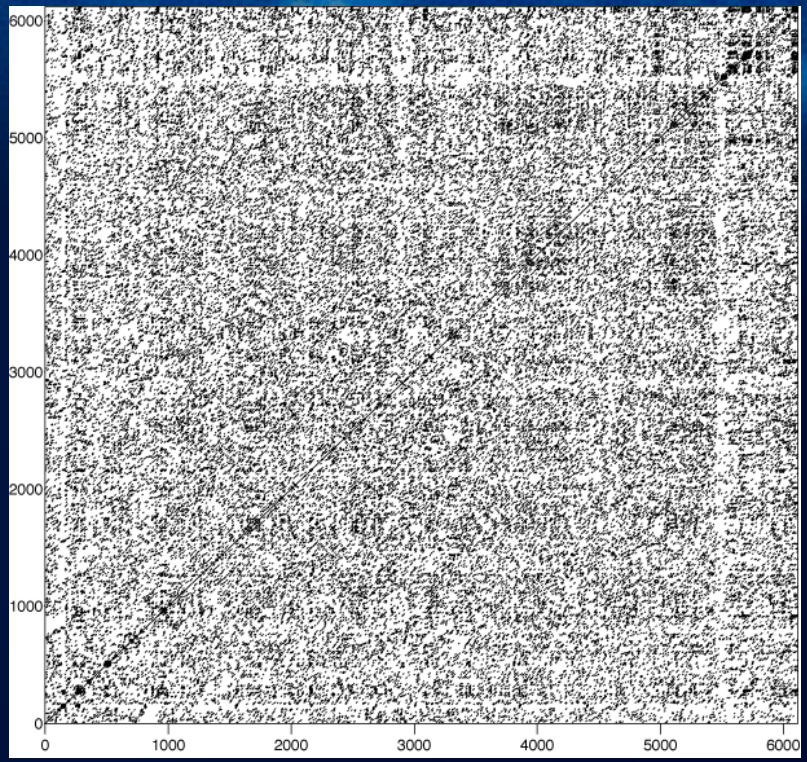
2. Sometimes things (proteins/genes) are only present in the sick patient (red)

Example Myc genes in many types of cancer.

BioInformatics: Find the difference
The complications of Life

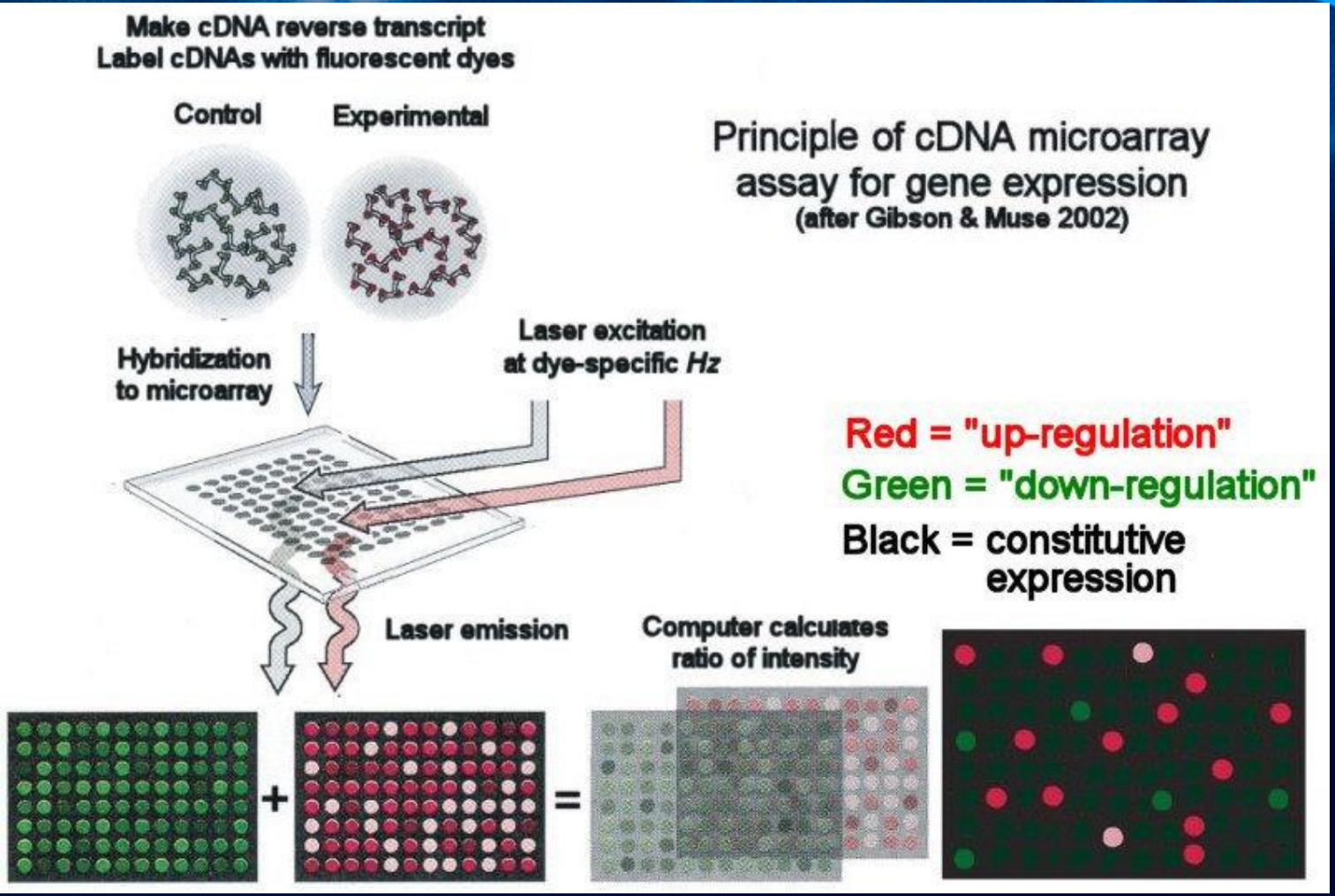
Find the difference in gene-expression in fruitflies (4 chromosomes)



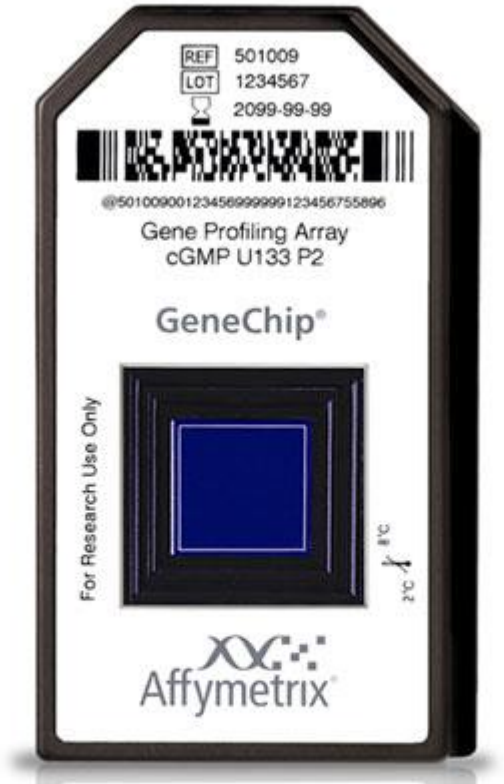


Imagine how this is for humans with 23 pairs of chromosomes (46 chromosomes)

BioInformatics: Find the difference
Real Life Application: Gene Array



What Do You See?



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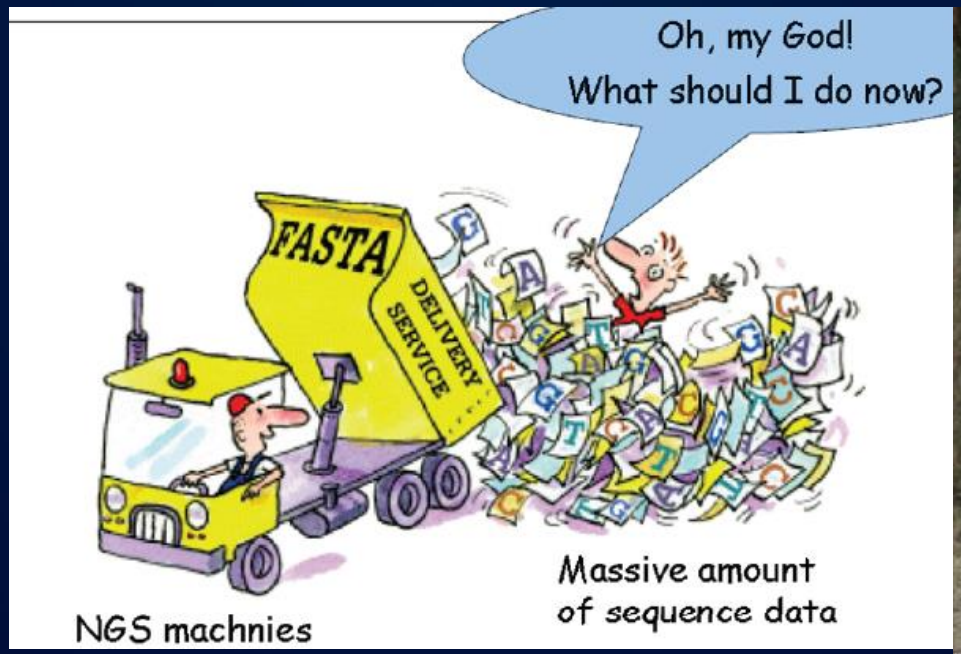
Advanced Biotech

- Biological data are being produced at a phenomenal rate
- A flood of massive and fast data



BioInformatics
Goal 2: Sort, Analyze, Compare and Predict

Keith Haring (untitled)

Questions:

how many elements (shapes) are there?
how many color schemes?
how do the shapes relate to the color scheme?
How did the artist developed over time?
What can we predict for the future?
How does the artist relate to other artists in the same field (graffiti and pop art)?

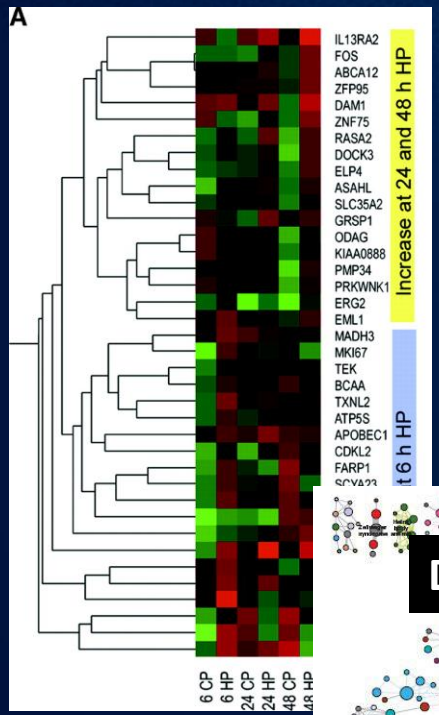
BioInformatics Goal 2: Sort, Analyze, Predict Relationship to Life Science

1. **Sorting the different elements** makes it possible **to find relationships** between proteins and genes within a person.
2. **Comparing the sorted elements to previous findings will tell us about how a disease develops.**
3. **Comparison to other patients** either healthy or with similar disease will **give us insight in what changes** during disease (comparing to healthy) **or** find **similar** changes in closely related diseases (for instance various cancers)
4. **Comparing the effects of treatment** in patients by taking a sample before and after treatment.

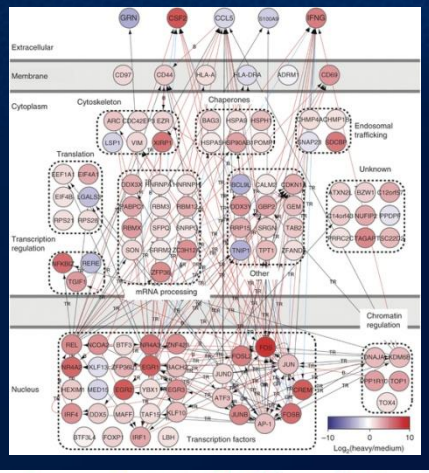
The information on relationship and change will enable scientists in various fields to determine what could be valid targets for drug discovery.

BioInformatics
Goal 2: Sort, Analyze, Compare and Predict Real Life Applications

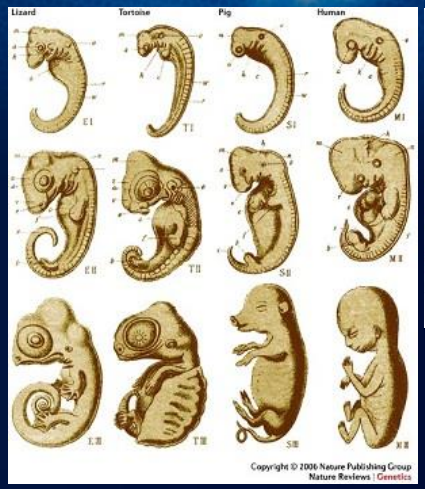
Hierachical Clustering

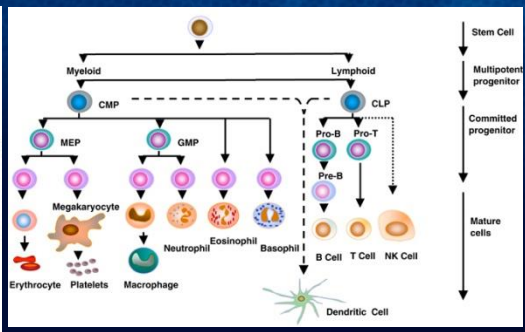


Pathway analysis

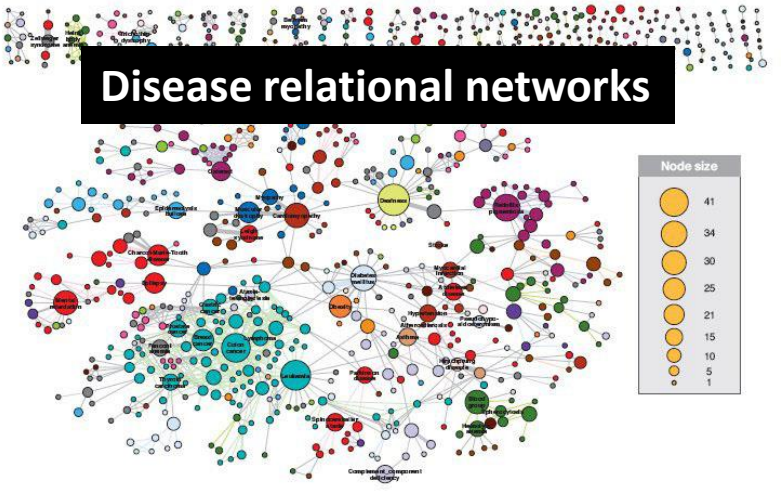


Ontogeny

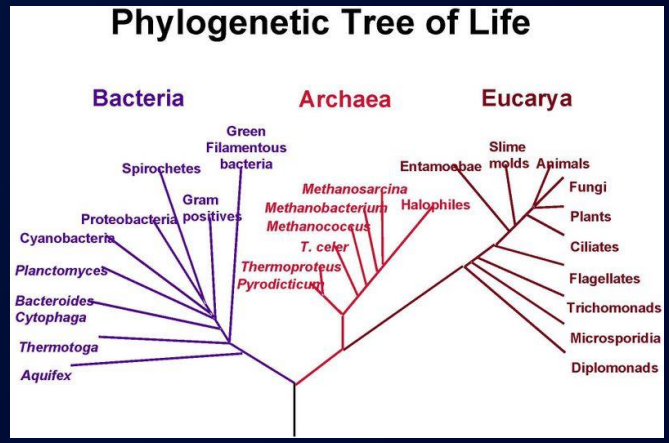




Disease relational networks



Phylogenetics



Common Features of Projects

- High throughput
- Use of technology, in particular
  - Automation (Robotics, AI)
  - Databases
  - Visualization, simulation/computational models
  - Groupware: Coordination and communication
- Public domain tools
- Open sharing of data

Large scale bioinformatics: genome projects

- Mapping
  - Identifying the location of clones and markers on the chromosome by genetic linkage analysis and physical mapping
- Sequencing
  - Assembling clone sequence reads into large (eventually complete) genome sequences
- Gene discovery
  - Identifying coding regions in genomic DNA by database searching and other methods
- Function assignment
  - Using database searches, pattern searches, protein family analysis and structure prediction to assign a function to each predicted gene
  - Data mining
  - Searching for relationships and correlations in the information
- Genome comparison
  - Comparing different complete genomes to infer evolutionary history and genome rearrangements

Applications

- Early detection of genetic predispositions to diseases
- Improved diagnosis of disease; Molecular diagnosis of leukemia, breast and prostate cancer.
- Pharmacogenomics
  - Appropriate treatment for genetic signature
  - Potential new drug targets Customized drugs
  - Individualized drugs selection
  - Appropriate doses determination

Applications

- Gene therapy and control systems for drugs
- Biological discovery
  - new and better molecular diagnostics
  - new molecular targets for therapy
  - finding and refining biological pathways
- Improve nutritional quality
- Alternative energy sources?

Application: Personalized Medicine

- The Drug Development Stages:
  - Drug Discovery
  - Pre-clinical Development
  - Clinical Development 4 Phases
- Stages are highly regulated
- Result is based on most of patients
- But .. Patients are created differently!

Murder case solved by BioInformatics: The dentist who infected his patients with HIV

In 1990 a young woman attracted HIV and blamed her dentist for infecting her.

Blood was taken from over 1100 patients and tested for HIV, 5 were found positive (patient A-E)

Blood was taken from the dentist and his lover, who was also a patient (patient F)

HIV mutates very fast and two most different variations found are compared (x and y)

Person	Sex	Known risk factor	Clinical status*
Dentist	M	Yes	AIDS
Patient A	F	No	AIDS
Patient B	F	No	Asymptomatic (CD4 = 222/μl)
Patient C	M	No§	Asymptomatic (CD4 = <50/μl)
Patient E	F	No	Asymptomatic (CD4 = 567/μl)
Patient G	M	No	Asymptomatic (CD4 = 400/μl)
Patient D	M	Yes	AIDS
Patient F	M	Yes	Asymptomatic (CD4 = 253/μl)

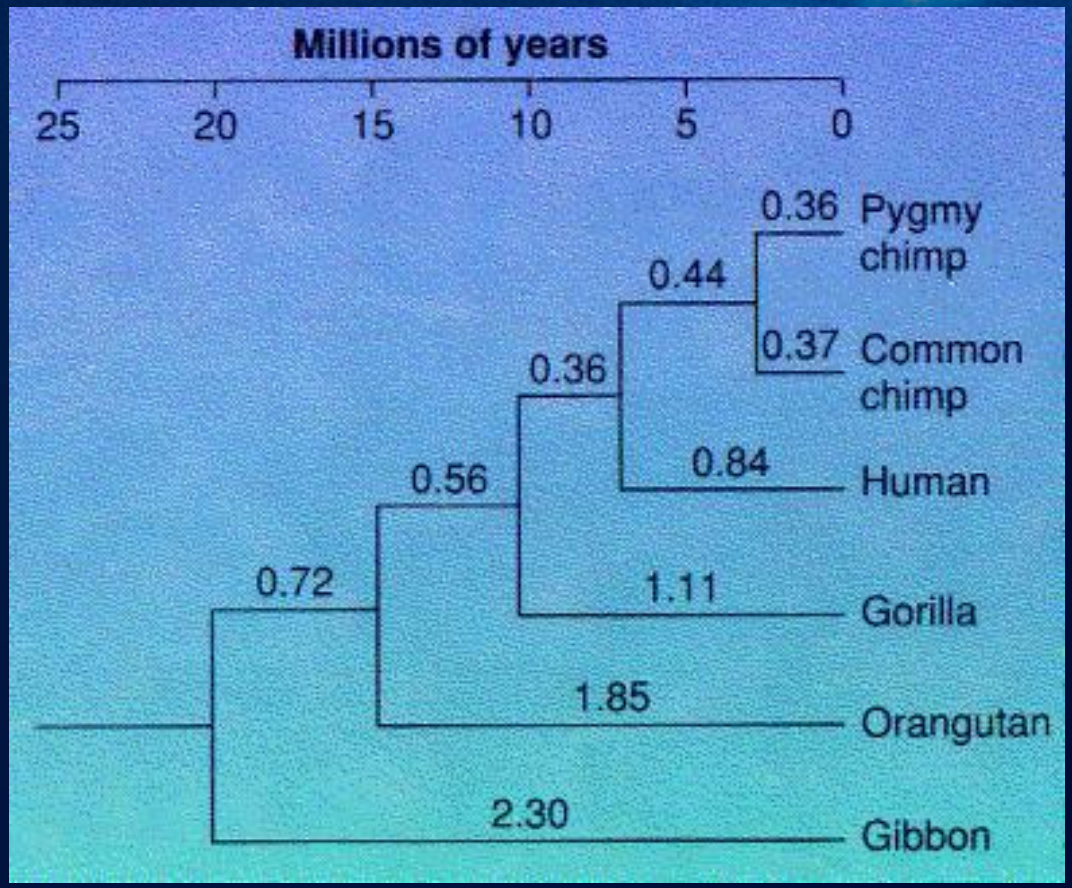
How did the court use bio-informatics to solve this case?

(Science, 256, 1992)

Phylogenetic analysis

Or…

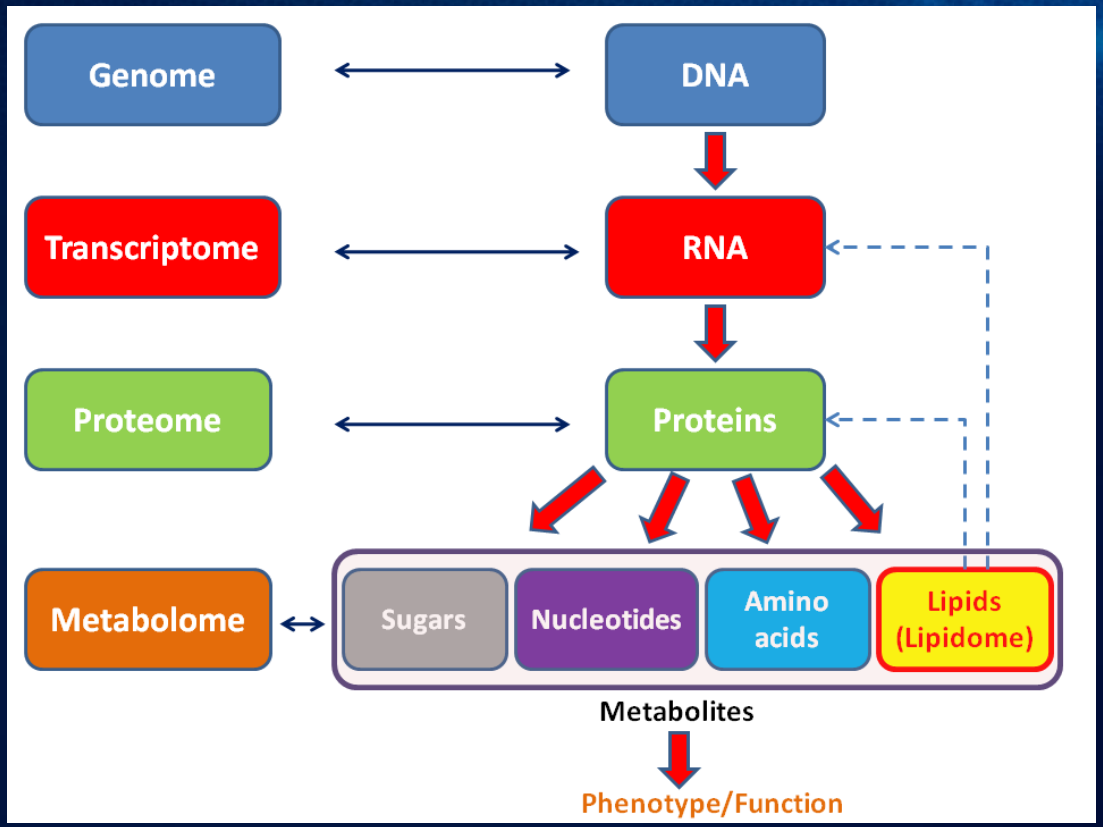
Finding evolutionary relationship based on the molecules of life: DNA, RNA, protein



Humans and chimpanzees are closely related

Humans and gibbons are far relatives

Bioinformaticians analyze any Biological material: They just add -omics to it



Genomics (~1970)

Transcriptomics (1997 – yeast, 2013 RNAseq)

Proteomics (1997)**/ Lipidomics**

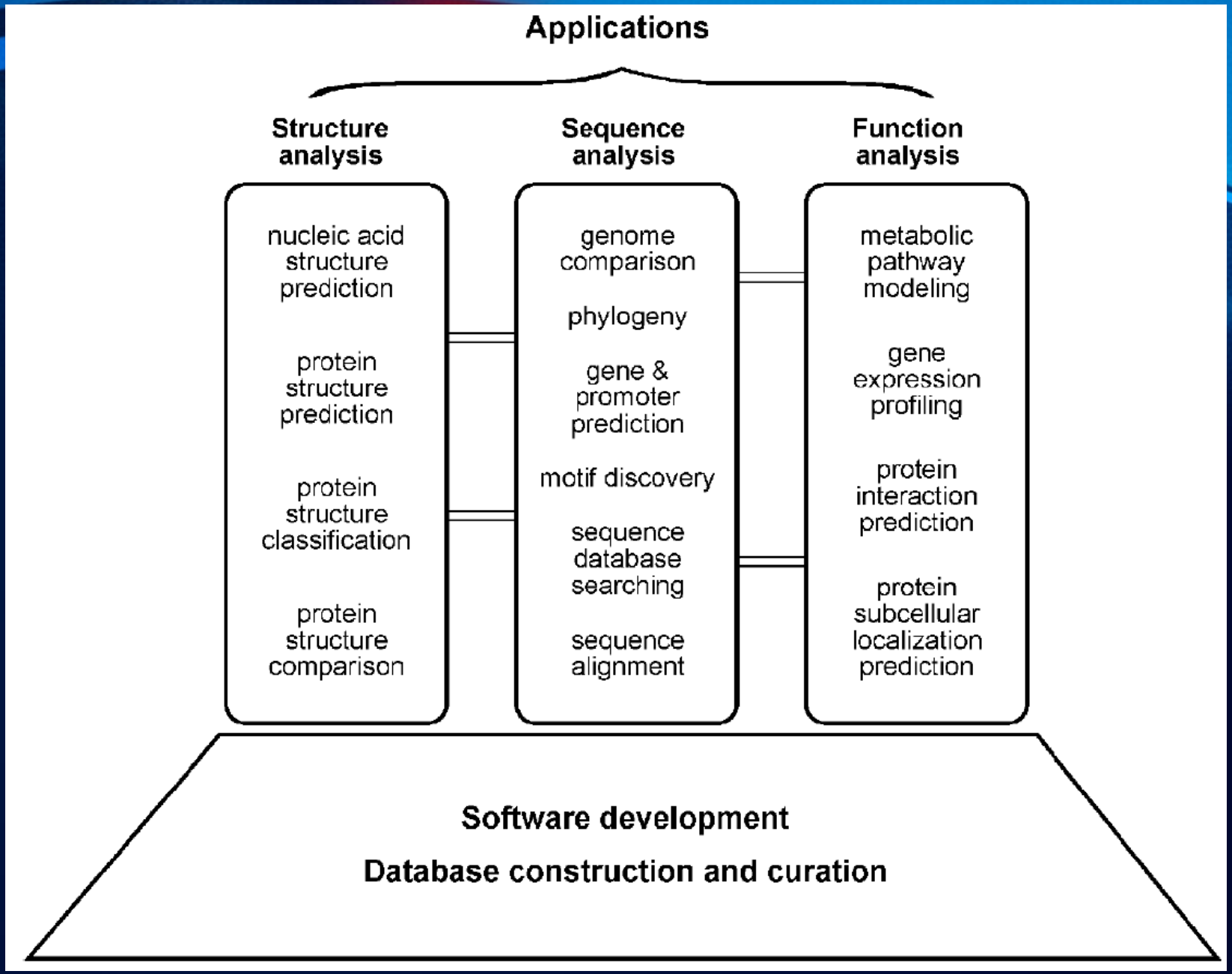
Metabolomics (first database 2005)

Microbe-host interactions

Human

Microbes

Microbiomics


Applications
Structure analysis
Sequence analysis
Function analysis
nucleic acid structure prediction
protein structure prediction
protein structure classification
protein structure comparison
genome comparison
phylogeny
gene & promoter prediction
motif discovery
sequence database searching
sequence alignment
metabolic pathway modeling
gene expression profiling
protein interaction prediction
protein subcellular localization prediction
Software development
Database construction and curation

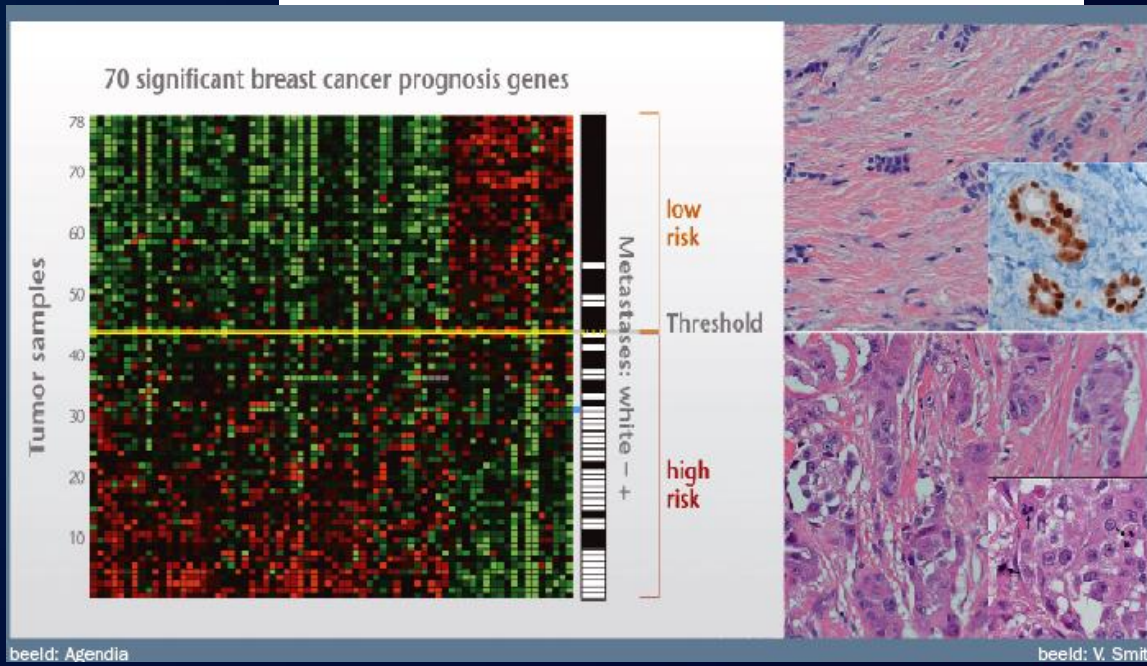
Molecular Diagnostics: Some examples

- Mammaprint.

Use expression 70 genes from an early-stage breast cancer tissue sample to figure out if the cancer has a low or high risk of recurrence within 10 years after diagnosis.

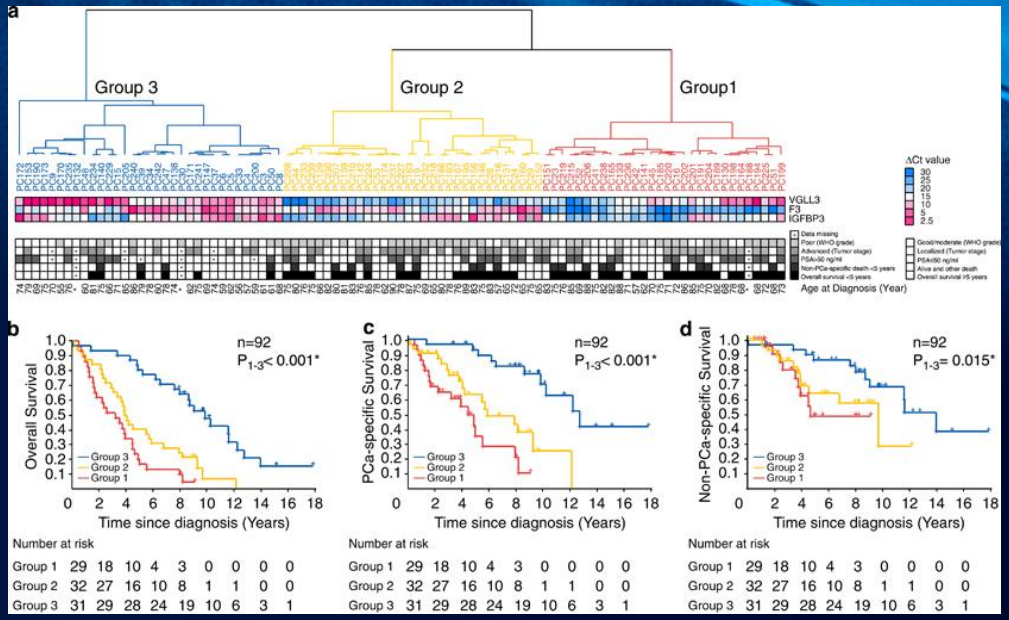
mammaprint™

decoding breast cancer.



Molecular Diagnostics: Some examples

- Prostate cancer (*Prostatype®Test System*). Tumor subtypes classification based on the embryonic stem cell gene predictor (ESCGP) signature (VGLL3, IGFBP3 and F3).



OPEN

Prostate Cancer and Prostatic Disease (2014) 17, 81–90

www.nature.com/pcan

ORIGINAL ARTICLE

An expression signature at diagnosis to estimate prostate cancer patients' overall survival

Z Peng[1], L Skoog[1,2], H Hellborg[3], G Jonstam[4], I-L Wingmo[2], M Hjälm-Eriksson[1,4], U Harmenberg[1,4], GC Cedermark[1,4], K Andersson[5], L Ährlund-Richter[6], S Pramana[7], Y Pawitan[7], M Nistér[1,2], S Nilsson[1,4] and C Li[1,4]

Personalized Medicine

- The ability to determine an individual's unique molecular characteristics and to use those genetic distinctions to diagnose more finely an individual's disease, select treatments that increase the chances of a successful outcome and reduce possible adverse reactions.
- Personalized medicine also is the ability to predict an individual's susceptibility to diseases and thus to try to shape steps that may help avoid or reduce the extent to which an individual will experience a disease

Subgroup Identification and Targeted Treatment

- Determine subgroups of patients who share certain characteristics and would get better on a particular treatment
- Discover biomarkers which can identify the subgroup
- Focus on finding and treating a subgroup

Subgroup Identification and Targeted Treatment

Genotype	Phenotype	Intervention	Outcome
Mutations/SNP Gene/Protein Expression Epigenetics	Diseases Disability Etc.	Drugs Therapies Regimes	Personalized medicine

Thank you for your attention!!!

Setia.pramana@stis.ac.id