

From Physics to Daily Life

Technology fallout in Bioinformatics

- A personal recollection -

Paolo Zanella



- PROGRESS in Science and Society is driven by discoveries, inventions and disruptive technologies
- One must invest in new technologies to trigger the virtuous chain research – technology – innovation
- Technology Transfer has many problems.
To involve in the transfer key developers of a technology it helps
- ICT and innovation are bound together from the middle of the XX Century.

The first electronic digital universal computer (ENIAC) built in 1945 by Eckert and Mauchly followed 300 years of mechanical attempts and mathematical studies, from the arithmetic machines of Schickard, Pascal and Leibniz to the big programmable machine of Babbage.

ICT revolution and evolution triggered by:

- The transistor (1949)
- The integrated circuits (1971)

CERN AS A PIONEER



26/09/2014

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at CERN

and before speed electronic machines took the power



Wim Klein worked in the Theory Division from 1958 as a human computer.

In August 1976 he calculated in the Main Auditorium the seventy-third root of a 507-digit number in 2 minutes 43 seconds.

CERN as a pioneer in ICT

CERN started surfing the digital computer waves at the end of the 1950's

It took 25 years of doubts and struggles with early generations of hardware and software before adopting it massively

CERN pioneered:

- 1) the development of networks,
- 2) visual computing,
- 3) big data analysis,
- 4) pattern recognition,
- 5) experimental data acquisition with real-time computer decisions,
- 6) modeling and simulations,
- 7) advanced computing systems,...



at CERN

Ferranti Mercury (1958-1965)

1024 40-bit words

four magnetic drums each holding $32K \times 20$ bits



(Courtesy of CERN)

Control Data 6600 (1965)

128K words of 60 bits ~ 0.94 MB

for the analysis of bubble chambers tracks



(Courtesy of CERN)

Computer language
FORTRAN



(Courtesy of CERN)

UA1 Experiment

168E Emulators (1983)

Computer Centre (1983)

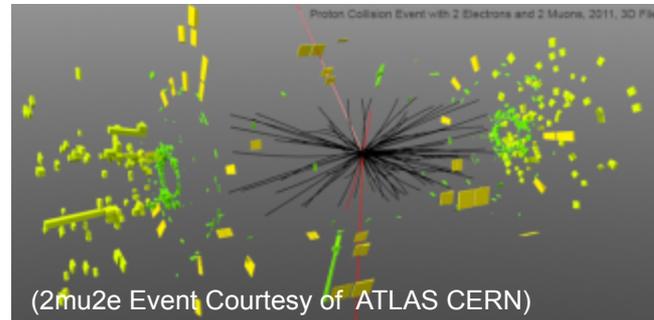


(Courtesy of CERN)



CERN as leader in digital systems for HEP

Over the last 60 years CERN made advances that changed radically the way experiments were designed and carried out.

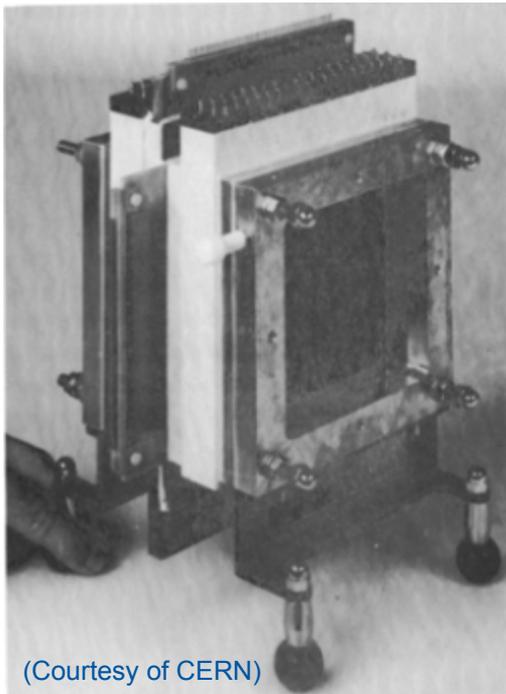


at CERN

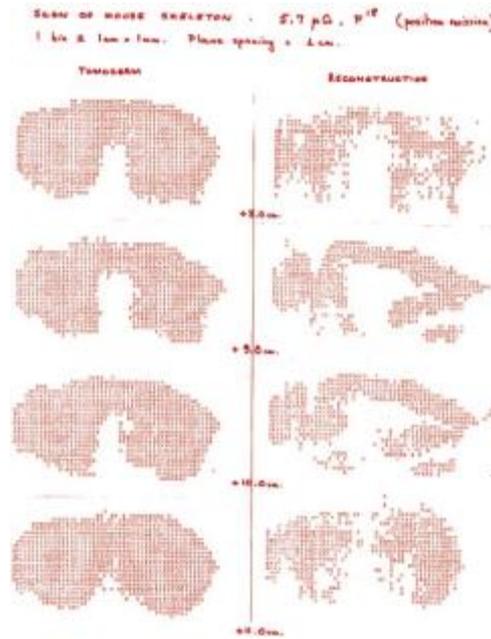
Some techniques that inspired the information revolution that is changing the world.

Positron emission camera (1977)

A. P. Jeavons et al. CERN-PRE-78-054



(Courtesy of CERN)



(Courtesy of CERN)

First PET mouse image (CERN 1977)

The PET, Positron Emission Tomography, has growing impact in medical practice.

WWW invented at CERN in 1989

Initial release 24th December 1990
The NeXT Computer used by Tim Berners-Lee became the **World first Web Server**



(Courtesy of CERN)



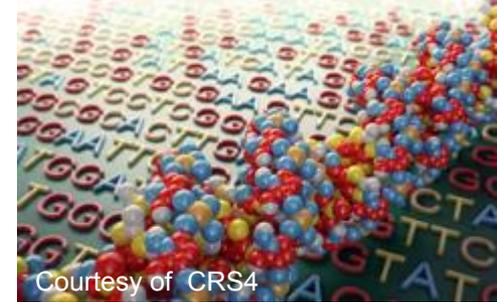
ICT and Life sciences

Crick and Watson determined the structure of the DNA in 1953 and a few years later, also at Cambridge, UK, Fred Sanger won 2 Nobel prizes for the invention of the sequencers.

- A protein data bank founded in 1971.
- Bacterial sequences were produced in the 80's and a new discipline, **bioinformatics was born**.
- The human genome project was launched in 1990. It took 13 years to publish the first genome – 3 billions bases.
- Today, the life sciences, exploring nature at the molecular level, as well as astrophysics, looking at the universe, are submerged by data.
- The new frontiers are biomed, pharma, healthcare, and bioinformatics with all its 'omics'.

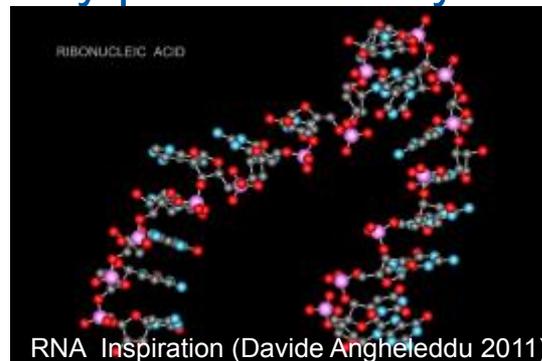


Bioinformatics in a nutshell



Courtesy of CRS4

The human genome is represented by two strings of symbols ATCG, corresponding to the bases aligned in pairs along the double helix packed into the 23 chromosomes. It contains the genes coding for proteins, necessary to support life and maintain health in the body. Problems may arise because of accidental deletions or mutations of bases. The code is about 3 billions base long and each human being has a different code. Errors may be the cause of diseases and may be transmitted to children. The fundamental hypothesis of biology was... one gene-one protein-one function, but after the last 2 decades of research and discovery it became many genes-many proteins-many functions...



Bioinformatics goes across boundaries

To determine a genome one needs **sequencers**, instruments that are evolving rapidly, reducing time and money needed to understand the many aspects of its information contents.

One requires of course very powerful computers and large disks to compare, analyze and store entire genomes.



Bioinformatics is the new discipline across the boundaries of genomics, proteomics and computer science technologies.



The European Bioinformatics Institute (EBI)

EBI is a branch of EMBL, created in **1993** in the Genome Campus of the Wellcome Trust at Hinxton, Cambridge, UK.



Courtesy of EMBL-EBI

It has been growing fast and now over 500 people from all European Countries and beyond works there. Its mission is to help the community of biologists and bioinformaticians, to carry out research, vital also for biomedicine, biotechnology and the Pharma Industry. It maintains, grows and develop a variety of databases and software applications for use by researchers.

The EBI runs a large computer centre making available database queries and executions of programs to users all over the world.

To spread all over Europe the knowledge of bioinformatics, the EU launched the ELIXIR project, a network of centres of competence to promote this discipline and its methods in view of the radical changes expected in Medical and Healthcare practices.



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TT in ICT@



CRS4 was founded in 1990, as an operational consortium including the Sardinian Regional Government, IBM-ITALY, STM Microelectronics, SARAS, etc... with the participation of a small team of CERN physicists and computer scientists to bring the island onto the map of the Information Society.

1993 - CRS4 was the first Italian www site;

1994 - the first 2 newspapers to go on-line were: the Washington Post and L'Unione Sarda;

in the 90's, successful collaboration with ENI-prospection division to discover new oil fields.

1998 - birth of Tiscali in Cagliari (big ICT company that provides internet and telecommunication services, expanded in Europe and was number 1 on the Italian Stock Exchange).



Most Recent research at



Growing interest from the cultural heritage Agency and contributions to the art presentation of the Monte Prama statues dating from 3000 years ago.



In recent years strong contribution to the data acquisition and analysis in a collaboration with CNR and NIH to sequence over 3600 DNA of Sardinian people and investigate genetic predispositions for diseases as Multiple Sclerosis, Diabetes 1, affecting the local population. This research is producing results in many related fields of the biomedical sciences and are being published by major scientific journals.

In 2014, “The Population Genomic Analysis of Ancient and Modern Genomes Yields. New Insights into the Genetic Ancestry of the Tyrolean Iceman and the Genetic Structure of Europe” has been published.



Bioinformatics: towards the future

1st step: **General to Cohort-based** medicine
followed by

“Cohort-based” medicine leading to **Personalized medicine**

Medical prevention, diagnosis and treatments

- 1) *knowledge of variants involved on the most common diseases,*
- 2) *genomics, proteomics, metabolomics , nutrigenomics etc.,*
- 3) *targeted drugs and therapy treatments,*
- 4) *availability and sharing of information between doctors and disciplines world wide*



More Information at

Wiley Book: From Physics to Daily Life

Applications in Biology, Medicine, and Healthcare, Bressan, Beatrice (ed.)

Paolo Zanella 30 years of computing at CERN **CERN-CN-90-2**

<http://information-technology.web.cern.ch/about/computer-centre/computing-history>

<http://www.ebi.ac.uk/>

<http://www.crs4.it/>



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