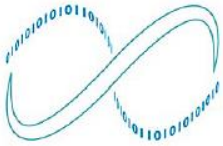
Centro di Competenza sul Calcolo Scientifico

Systems Biology & Systems Medicine



SysBioM

Centro Interdipartimentale
Molecular Systems Biology

Michele De Bortoli

Scienze Cliniche e Biologiche

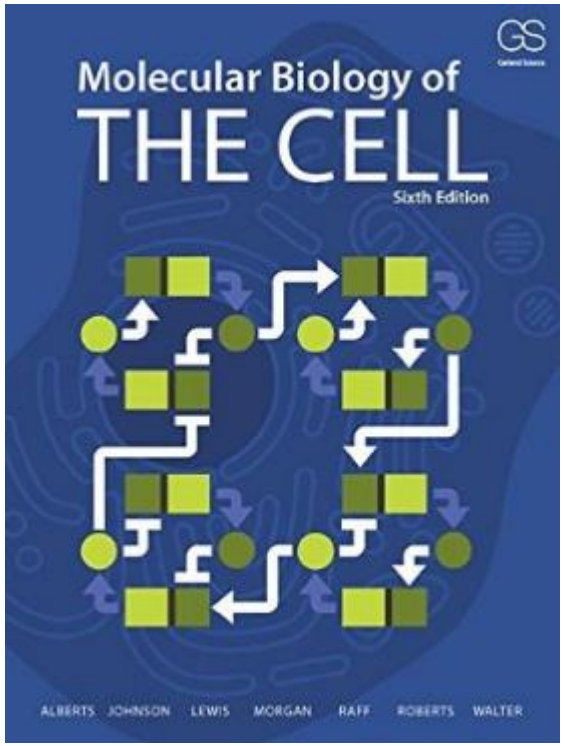
Polo San Luigi

Orbassano

1. *Sistemi complessi: programma di dottorato e Centro*

2. *Dipartimento di Scienze Cliniche e Bioloiche*

Le funzioni delle cellule e degli organismi sono costruite, regolate e si perpetuano grazie all'azione di centinaia di migliaia di entità molecolari diverse (geni, RNA, proteine, metaboliti,…) che interagiscono tra loro in modo coordinato, specifico e dinamico.



Studiarne il comportamento come ***sistema***, e non come singole molecole, è fondamentale.

Oggi questo è possibile, grazie all'avanzamento di:

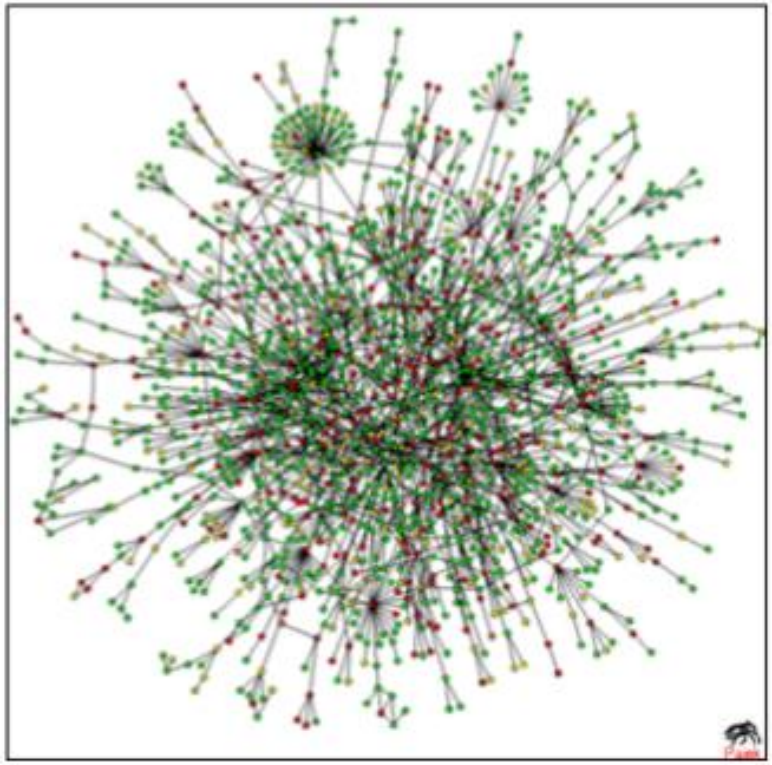
- Tecniche analitiche parallele
  (NGS, arrays, proteomica, metabolomica,…)
- Metodi di analisi dei dati e di modellizzazione
  (sistemi complessi, neural networks, …)
- Metodi informatici e computazionali
  (database, algoritmi di integrazione, …)

Systems Biology e *Systems Medicine* richiedono una profonda interazione ed integrazione tra le capacità e le conoscenze di varie discipline

Fisica, Biologia e Medicina, Matematica, Informatica, …

L'elaborazione si basa su:

- Teoria delle reti
- Modellizzazione quantitativa
- Ontologie



Centro Interdipartimentale
Molecular Systems Biology

Dottorato in Sistemi complessi nelle Scienze della Vita

PhD Programme: Complex Systems in Life Sciences

input

LM
Biologia
Biotecnologia
Medicina
Fisica
Matematica
Informatica
Ingegneria

Laureati stranieri

Collegio:
Medicina, Biologia, Fisica, Informatica, Matematica, Ingegneria, esterni

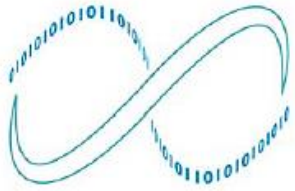
Didattica formale e seminariale, con programmi di azzeramento

Thesis:
- Regulatory Genomics
- Cellular regulatory pathways
- Bioinformatics
- Systems Biology
- Molecular networks
- Systems Medicine
- Modelling of biological phenomena
- Epidemiology
- Vaccinology

output

La disponibilità di una risorsa di super-calcolo scientifico in Open-Lab è fondamentale per il successo delle ricerche che impegnano i nostri studenti

… e non solo!



SysBioM

Centro Interdipartimentale
Molecular Systems Biology

Polo Universitario del San Luigi di Orbassano

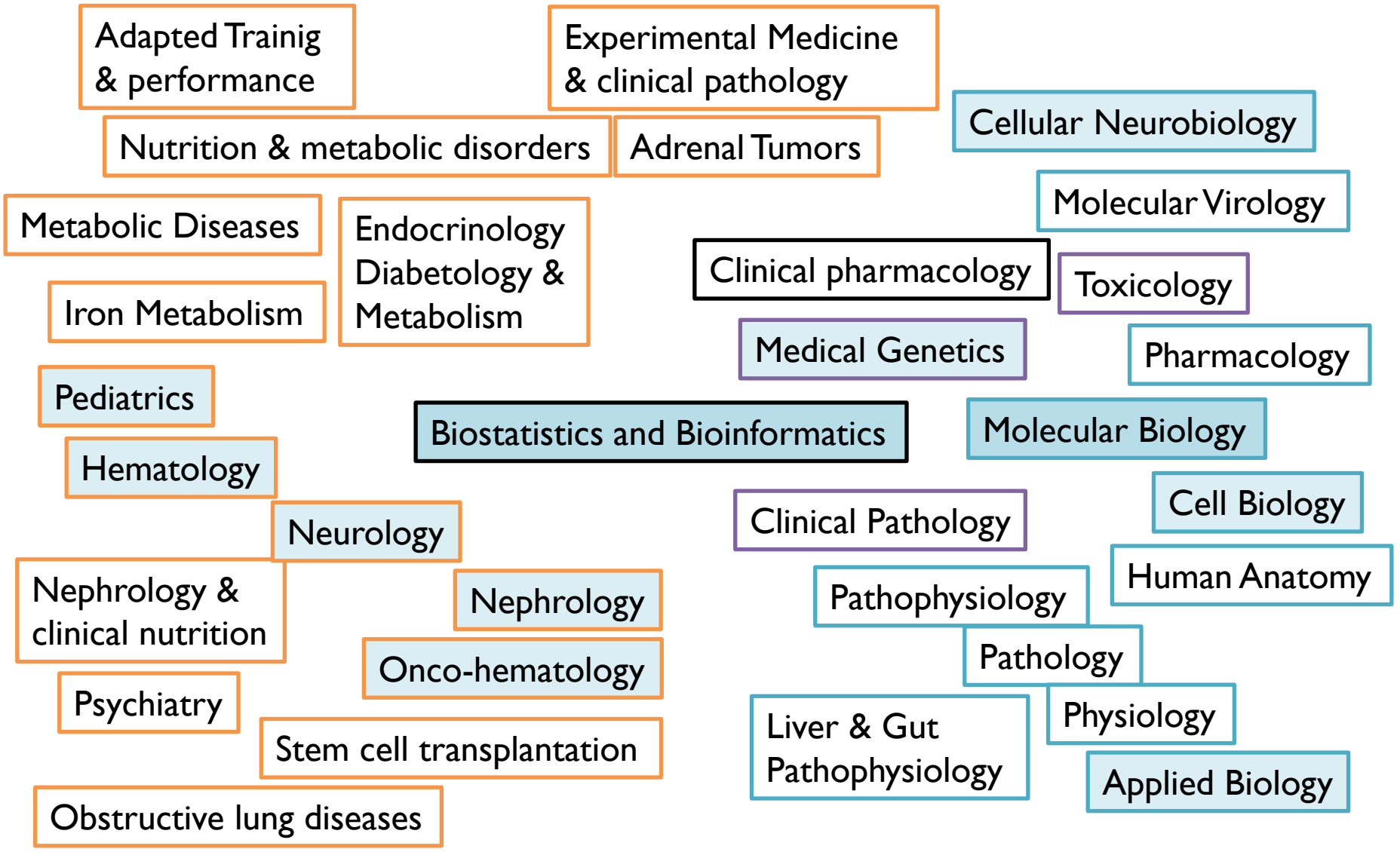
Ospedale – Università - NICO

Dipartimento di Oncologia
Dipartimento di Neuroscienze

Dipartimento di Scienze Cliniche e Biologiche

Corso di Laurea in Medicina e Chirurgia
Corsi di laurea delle professioni sanitarie
Master in Cellular & Molecular Biology

2014, Comm. Ricerca DSCB



Orange: clinical . Blue:basic. Blue shadow grades indicate involvement in computing.

Key Research Areas

Rare diseases

The last day of February every year is **Rare Disease Day.** In January 2016, 10 new projects kicked off focusing on the development of new treatments for rare diseases. The projects are funded under Horizon 2020, the EU's funding programme for research and innovation (2014 – 2020). Read more about these projects 321 KB

Many rare diseases cause chronic health problems or are even life-threatening. Genetic factors play a role in a majority of these diseases. The impact on the quality of life of affected patients, of whom many are children, is significant.

In the European Union, a disease is considered rare when it affects not more than 1 person in 2.000. This low prevalence is the common feature shared by all rare diseases, which altogether affect all biological systems. This nevertheless means that between 6 000 and 8 000 different rare diseases affect or will affect an estimated 30 million people in the European Union.

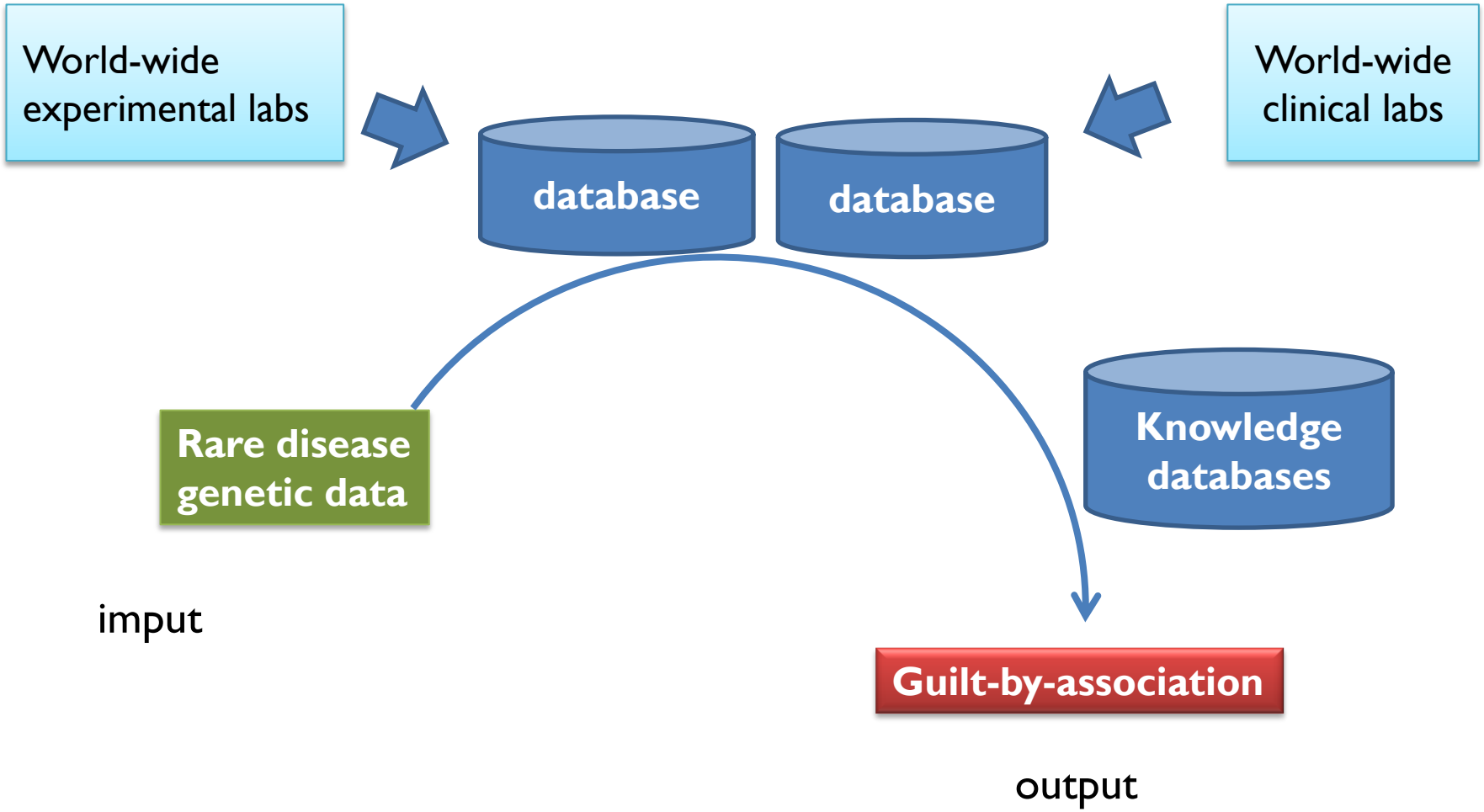
Links

- Horizon 2020
- International Rare Diseases Research Consortium (IRDiRC)
- E-RARE ERANET Co-fund
- EU policies for rare diseases

Video

- In search of IRDiRC

Rare disease: genetics and molecular characterization



Single clinical case requires computational power

- girl with uncorrect 9p-syndrome diagnosis by karyotype
- Reassesment: balanced translocation
- Further analysis with NGS techniques

complex chromosomal rearrangement (>30 breakpoints)

Breakpoints, epigenetics mapping point to pathways of neurodevelopment

- Mate-pair sequencing of the patient DNA
- WGS on trio DNA (mother-father-patient)
- RNA-sequencing on trio blood

Assay	Sample	Fastq file size (GB)	N°Reads (Million)
Mate-Pair Sequencing	Child	18	57
Whole Genome Seq	Child	315	908
Whole Genome Seq	Father	(106 the BAM file)	1369
Whole Genome Seq	Mother	272	784
RNA-Seq	Child	11	55
RNA-Seq	Father	7	39
RNA-Seq	Mother	9	46

NGS-based rare-disease-causing-gene discovery

Whole Exome Sequencing: ~1% protein coding portion of the human genome (**the exome**) is sequenced by NGS.

Autism sequencing consortium

Large international effort (Autism Sequencing Consortium, New York, USA) - exome sequencing of 50,000 trios (one proband and parents). Our group contributes 500 trios.

Need: analysis of WES data aligned to the reference human exome (.bam files).

Alfredo Brusco

Project

"Machine Learning Techniques in Food Risk Assessment"
funded by the European Food Safety Agency.

Dealing with Extensive Literature Searches

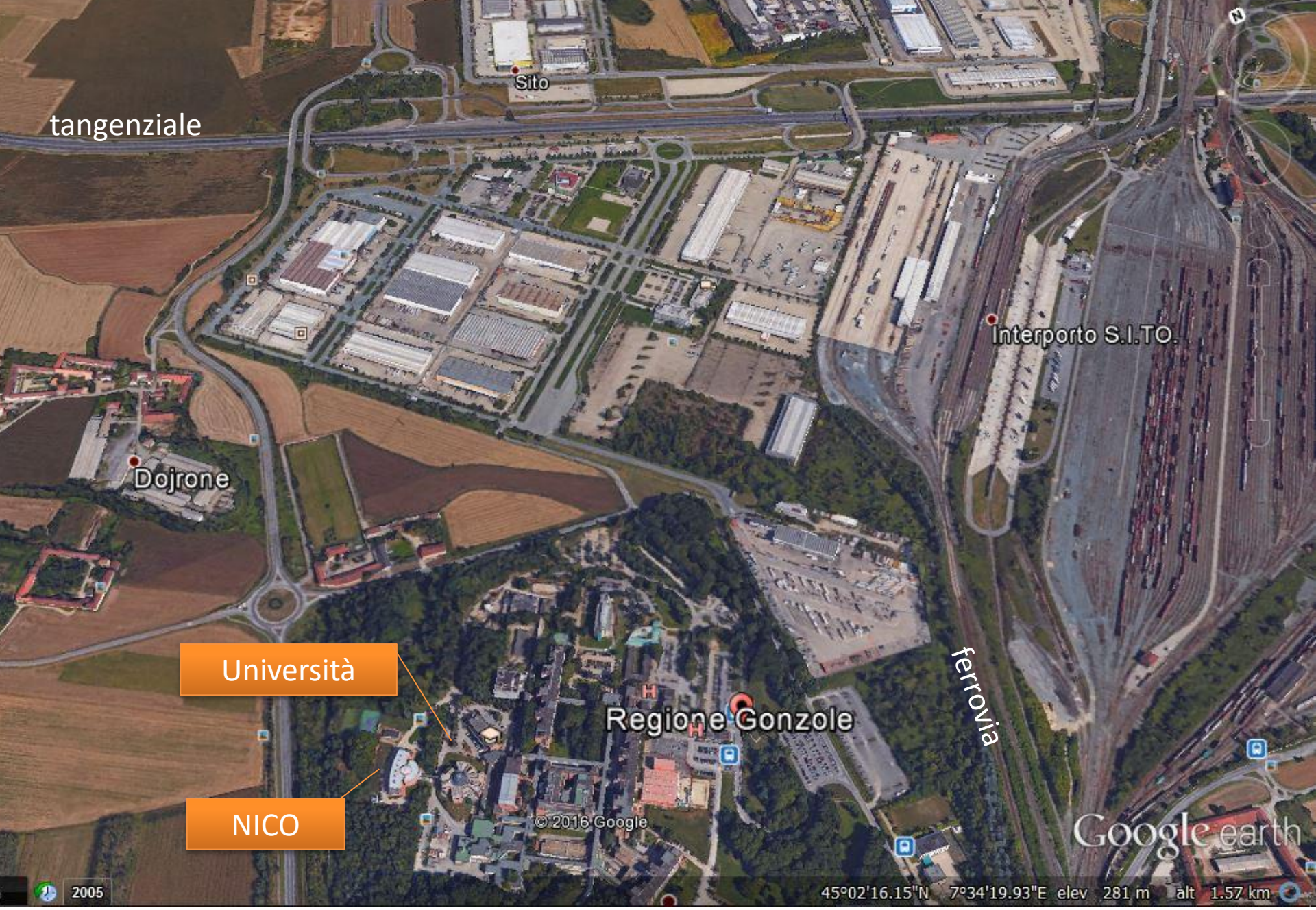
mapping the evolution of hypothetically any general topic over time

Machine Learning algorithms Super Learner solutions

active learning algorithms for unsupervised pattern

Current test: a bulk of 2,655,365 abstracts related to Machine Learning Techniques retrieved from bibliographic databases.

Dr. Paola Berchialla - Unit of Biostatistics
Department of Clinical and Biological Sciences, University of Torino


tangenziale
Sito
Interporto S.I.TO.
Dojrone
Università
Regione Gonzole
ferrovia
NICO
© 2016 Google
Google earth
2005
45°02'16.15"N 7°34'19.93"E elev 281 m alt 1.57 km

Grazie !