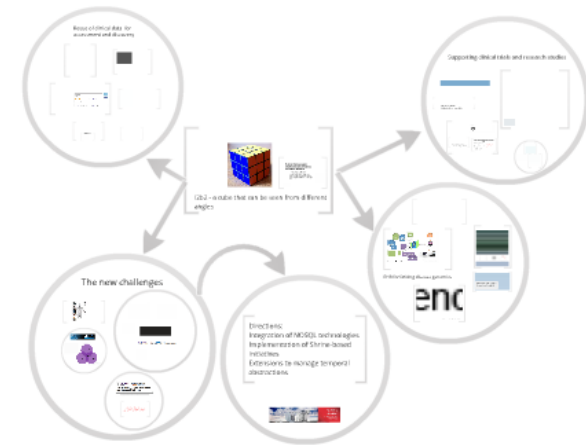


Thank you ...



Lelio Menozzi - Pavia from Porta Calcinara

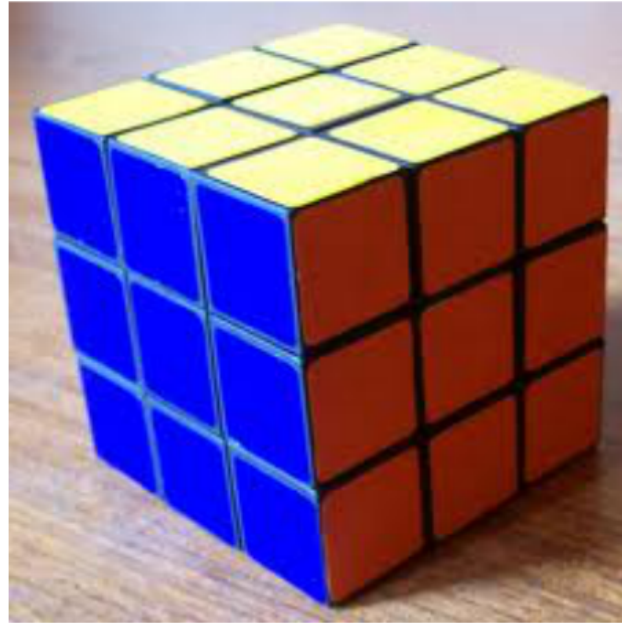


The I2b2 Pavia projects: experiences, lessons learned and future directions

Riccardo Bellazzi

Department of Electrical, Computer and
Biomedical Engineering, University of Pavia
LISRC, Maugeri Foundation Research Hospital,
Pavia





**The I2b2 Pavia projects:
experiences, lessons learned
and future directions**

Riccardo Bellazzi

Department of Electrical, Computer and
Biomedical Engineering, University of Pavia
LISRC, Maugeri Foundation Research Hospital,
Pavia



I2b2 - a cube that can be seen from different angles

Using electronic health records to drive discovery in disease genomics

Isaac S. Kohane

NATURE REVIEWS | **GENETICS**

VOLUME 12 | JUNE 2011 | 417

Dermatology Online Journal

Volume 18 Number 5

May 2012

Novel approach to utilizing electronic health records for dermatologic research: Developing a multi-institutional federated data network for clinical and translational research in psoriasis and psoriatic arthritis

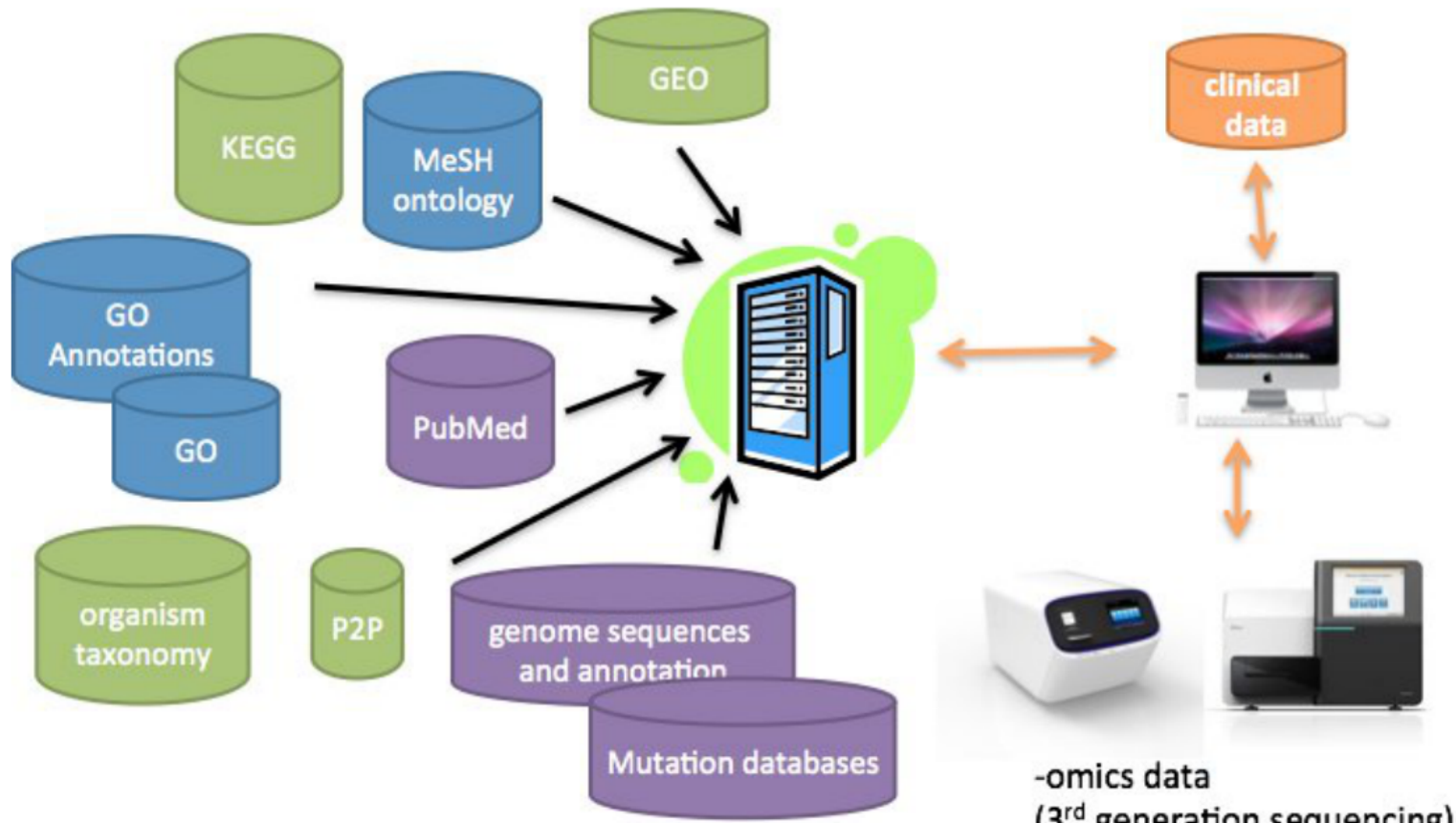
April W Armstrong¹ MD MPH, Shalini B Reddy² BA, Amit Garg³ MD

Dermatology Online Journal 18 (5): 2

1. University of California Davis, Department of Dermatology, Sacramento, California

2. Boston University School of Medicine, Boston, Massachusetts

3. Boston University School of Medicine, Department of Dermatology, Boston, Massachusetts

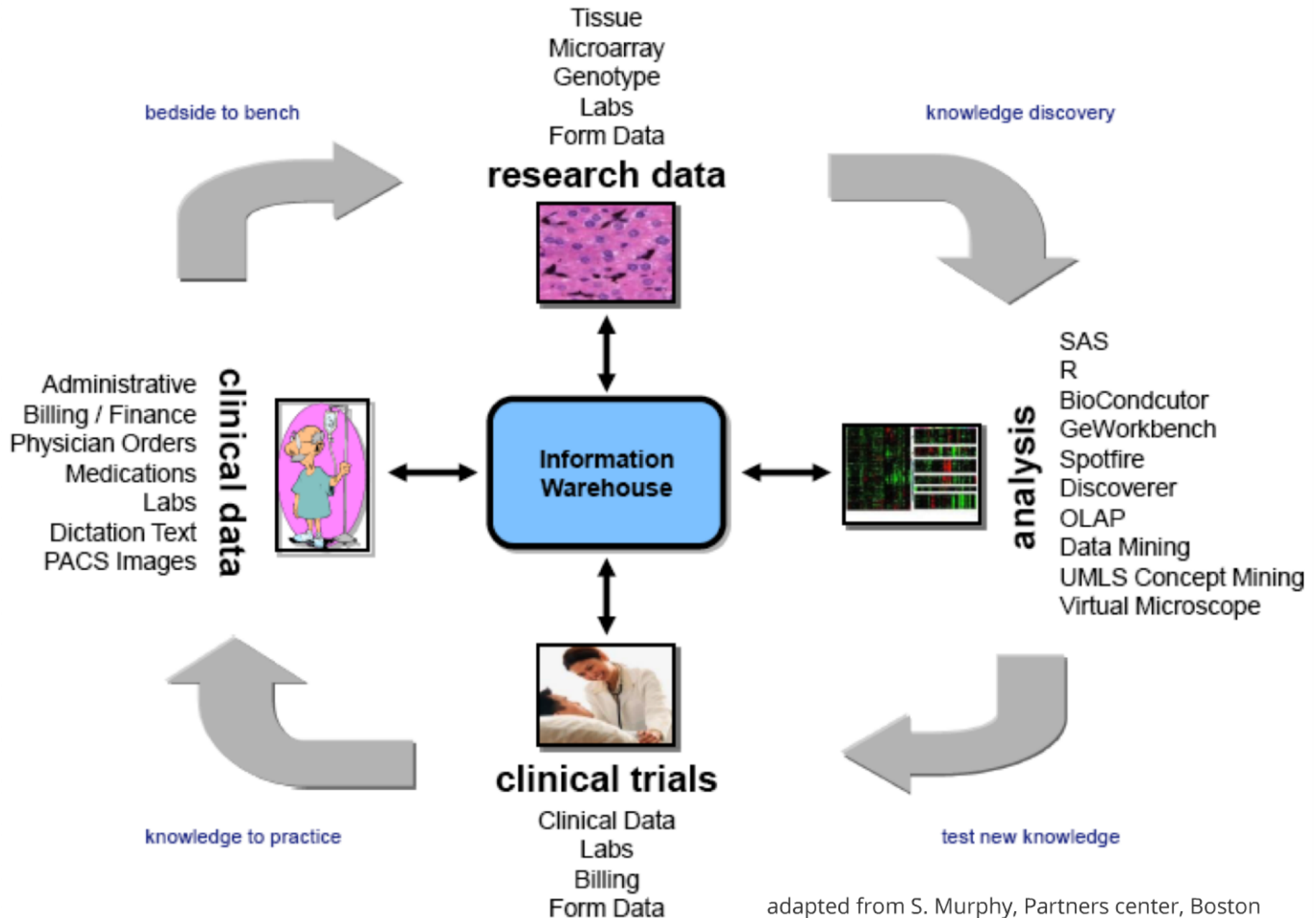


Adapted from Zupan (2009)

-omics data
(3rd generation sequencing)



clinical
data



adapted from S. Murphy, Partners center, Boston



FONDAZIONE ISTITUTO NEUROLOGICO NAZIONALE C. MONDINO

Istituto di Ricovero e Cura a Carattere Scientifico



Foundation

How to reach us

Contact

For the patient

Hospitalization and Treatment

Research

Training

English Italiano

NGS Projects - SVELA - epilepsy (36 genes over 200 patients)





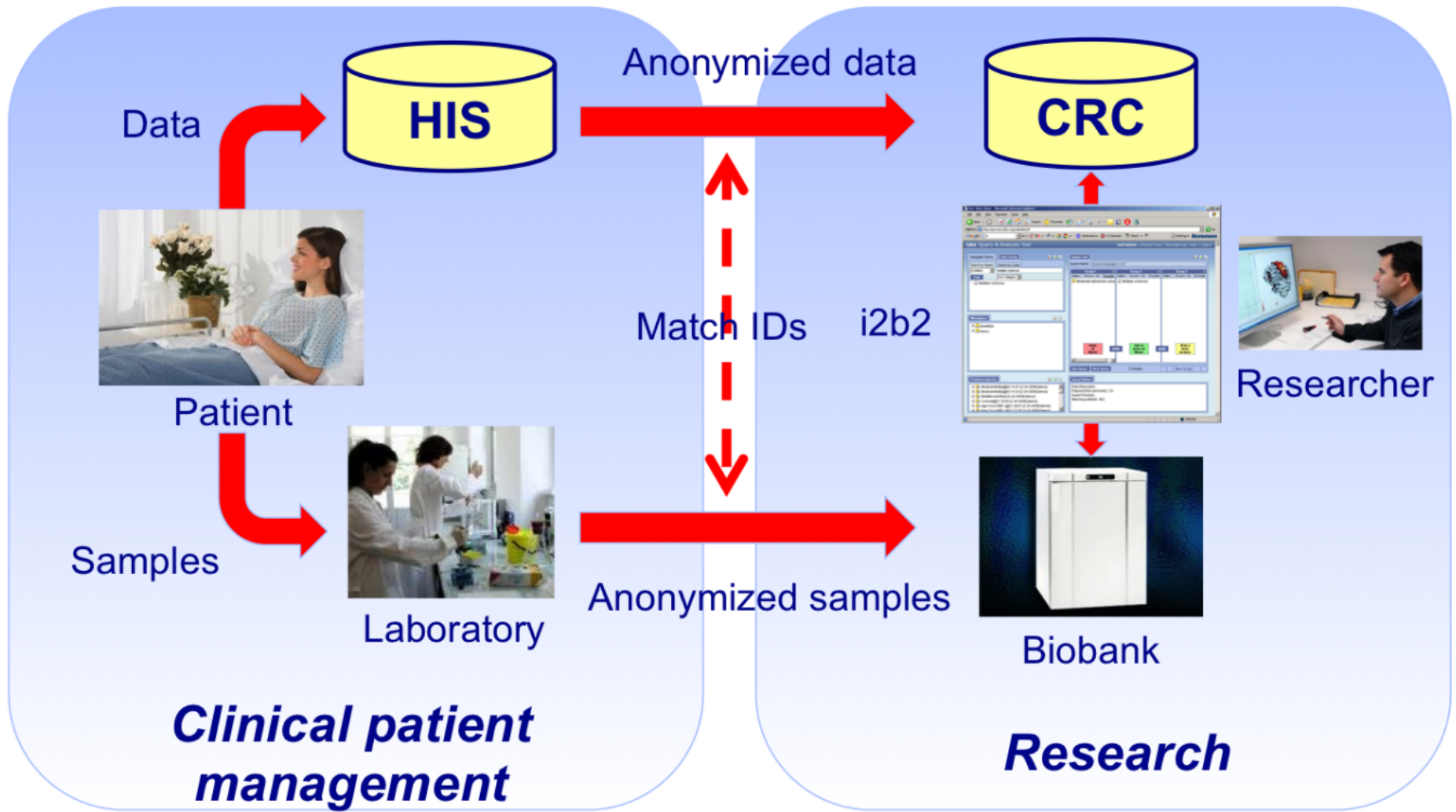
RECONFORCING WITH AI AND DATA SCIENCE AND TECHNOLOGY
 TO INTEGRATE
 CLINICAL AND BIOLOGICAL KNOWLEDGE
 SUPPORTING
 ONCOLOGY TRANSLATIONAL RESEARCH

Dimensioni (in %)	n
Breast	1343
Gastric	710
Stomach	62
Colon	54
Liver Metastasis	78
Metastatic, unspecified	25
Bladder	14
Other	149
TOT	2882

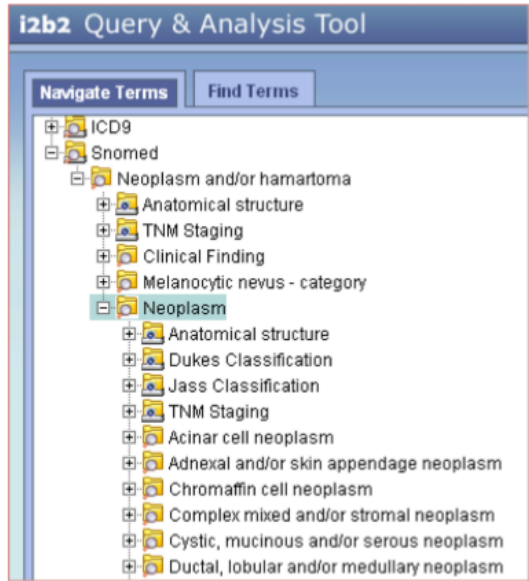
Ontologies AND NLP

FONDAZIONE SALVATORE MAUGERI
 CLINICA DEL LAVORO E DELLA RIABILITAZIONE
 I.R.C.C.S.
 ISTITUTO DI RICOVERO E CURA A CARATTERE SCIENTIFICO

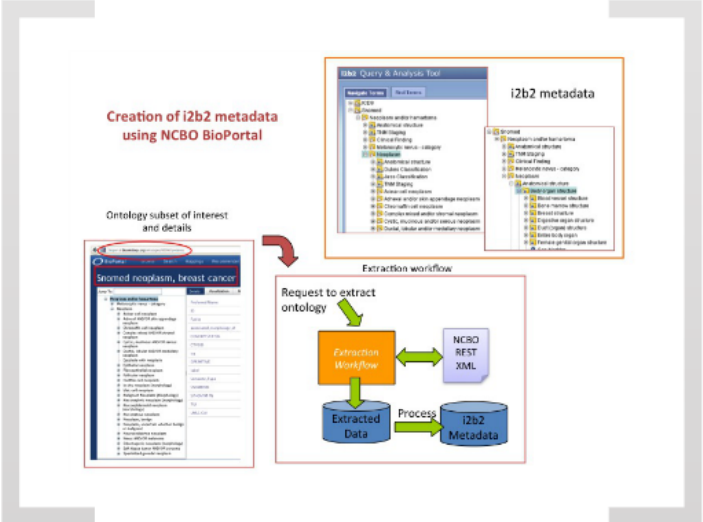




Tumoral Tissue Type	#
Breast	1548
Colon	410
Stomach	62
Thyroid	58
Liver Metastasis	58
Metastatic Lymph nodes	35
Kidney	18
Other	149
TOT	2338



*Ontologies
AND NLP*



Using electronic health records to drive discovery in disease genomics

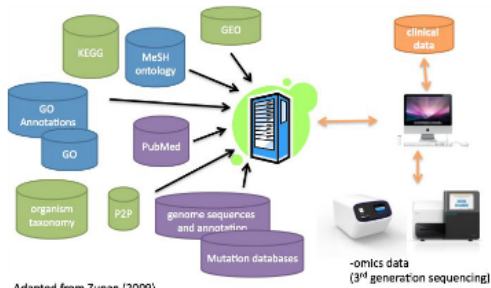
Greg S. Aronow

bioRxiv preprint doi: <https://doi.org/10.1101/000000>; this version posted January 1, 2013. The copyright holder for this preprint (which was not certified by peer review) is the author/funder, who has granted bioRxiv a license to display the preprint in perpetuity. It is made available under aCC-BY-NC-ND 4.0 International license.

Dermatology Online Journal

Volume 18 Number 1
April 2013

Next steps in using electronic health records for dermatologic research: challenges and solutions. Greg S. Aronow, MD, PhD, and
Michael J. Goldstein, MD, PhD. [doi:10.1007/s12247-013-0001-1](#)
J Am Acad Dermatol 2013;68:1-10. [http://dx.doi.org/10.1016/j.jaad.2012.12.010](#)

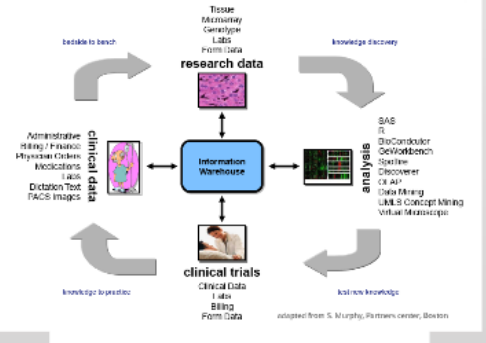


Adapted from Zupan (2009)



DEPARTMENT OF GENETICS
UNIVERSITY OF CALIFORNIA, SAN DIEGO
116D GENETICS BUILDING
SAN DIEGO, CA 92161-0601
TEL: 619-594-3800
WWW.GENETICS.UCLA.EDU

EHR for Driving disease genomics



adapted from S. M. Lathrop, Partners Center, Boston

UNIVERSITY OF CALIFORNIA, SAN DIEGO
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NGS Projects - SVELA - epilepsy
(36 genes over 200 patients)

Supporting clinical trials and research studies



SUMMIT Substrate markers for Micro- and Macro-vascular hard endpoints for Innovative diabetes Tools
 Diabetes complications
 Multicenter study > 1000 patients



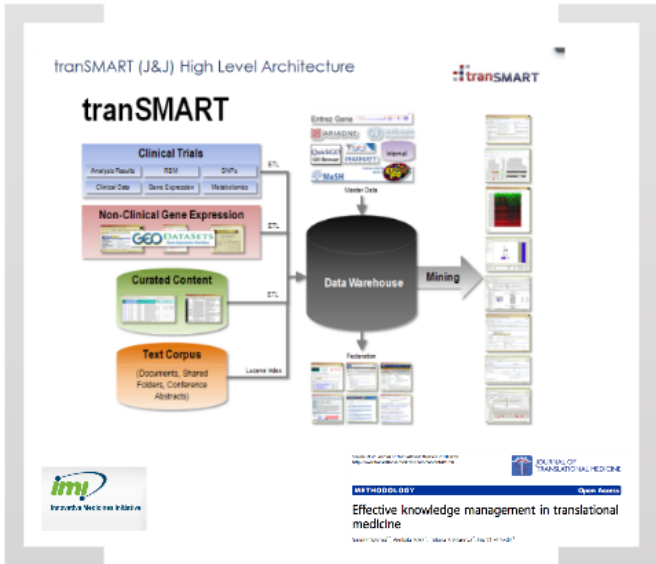
Fondazione IRCCS
Policlinico San Matteo



Dilated cardiomyopathy
 multicenter study - 360 patients

An I2b2 repository for Immuno-transplant services

Genetics: AgeGen, Cytosine, Metabac, Blood groups, HLA, HLA-C, serotype A, HLA-D, KR, Regg, T346
 Demographics: Age, gender, Race, Weight
 Administrative: Hospital departments, billing
 Total number of samples: 17007
 RNA: 16000
 Observations: 100,000
 Patients: 7000

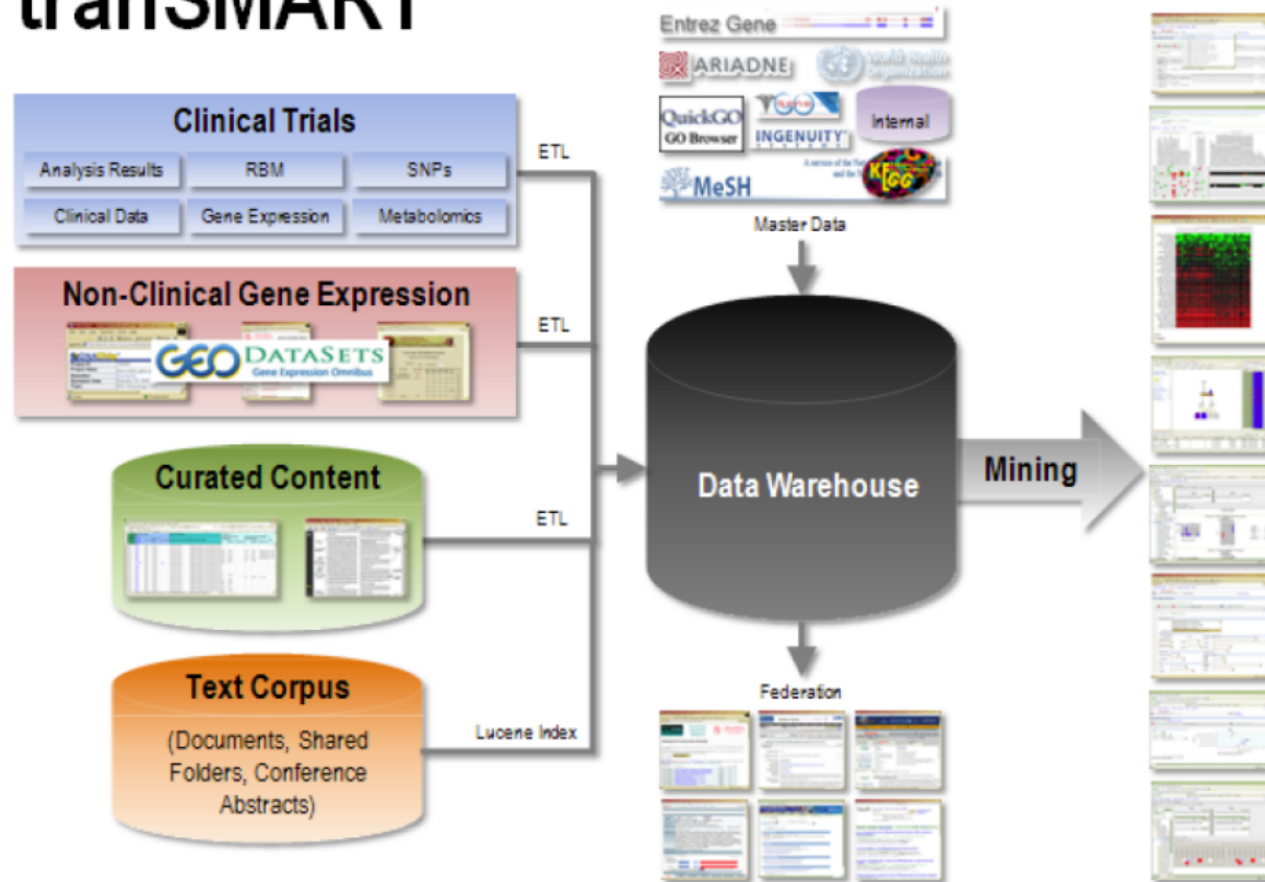


EHR4CR

*Electronic Health Records
for Clinical Research*



tranSMART



Szalma et al. *Journal of Translational Medicine* 2010, 8:68
<http://www.translational-medicine.com/content/8/1/68>

Effective knowledge management in translational medicine

Sándor Szalma^{1*}, Venkata Koka¹, Tatiana Khasanova², Eric D Perakslis³



SUMMIT

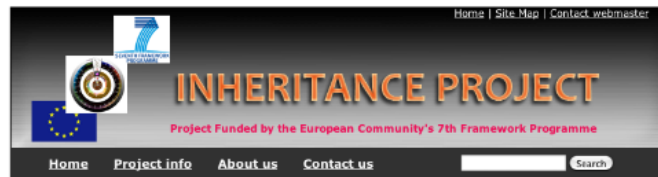
SURrogate markers for Micro- and Macro-vascular hard endpoints
for Innovative diabetes Tools

Diabetes complications

Multicenter study > 1000 patients



Fondazione IRCCS
Policlinico San Matteo



Dilated cardiomyopathy
multicenter study - 300 patients

An i2b2 repository for Immuno-transplant services

Genetics

Antigens, Citokines, Bw4Bw6,
Blood groups, HLA, HLA G,
serum HLA, Hsp70-1, Kir, Rage,
TLR4

Demographics

Age, gender, Race, Weight

Administrative

Hospital department, billing


Total number of concepts: 17501

HLA: 16014

Observations: 192363

Patients: 7396

[Home](#) | [Site Map](#) | [Contact webmaster](#)



INHERITANCE PROJECT

Project Funded by the European Community's 7th Framework Programme

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Dilated cardiomyopathy
multicenter study - 300 patients

An i2b2 repository for Immuno-transplant services

Genetics

Antigens, Citokines, Bw4Bw6,
Blood groups, HLA, HLA G,
serum HLA, Hsp70-1, Kir, Rage,
TLR4

Demographics

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Hospital department, billing

Total number of concepts: 17501

HLA: 16014

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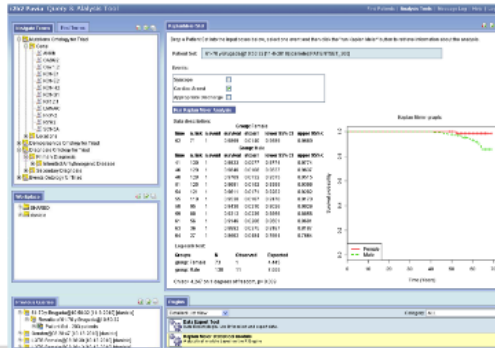
Patients: 7396



R Engine Cell: integrating R into the i2b2 software infrastructure

Daniele Segagni, Fulvia Ferrazzi, Cristiana Larizza, et al.

JAMIA published online January 24, 2011
doi: 10.1136/jamia.2010.007914



This is the home of the i2b2 Wiki ExportXLS plugin

About the ExportXLS Web Client Plugin

The plugin is a web client for the i2b2 Wiki ExportXLS plugin. It allows you to export data from the i2b2 Wiki ExportXLS plugin to a spreadsheet.

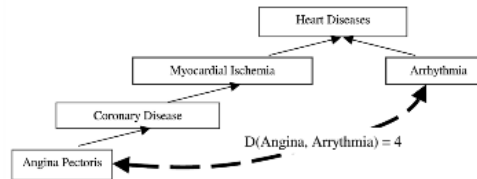
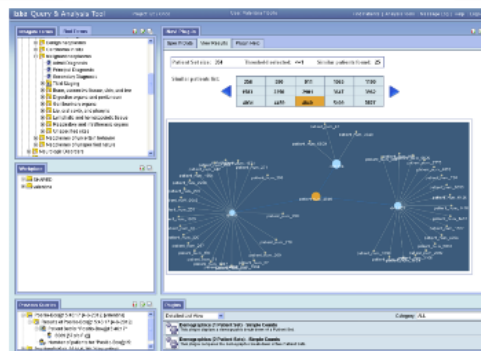
Introduction

This plugin provides the following functions:

1. Retrieve any selected Concepts (Strategy defined in export) in a set of selected subjects.
2. Export the data to a spreadsheet.
3. Export the selected data into an Excel spreadsheet.

Instructions

1. Follow the "Export Data" link. Then, drag and drop a "Patient Set" and one or more "Concepts (Strategy Term)" into the input boxes.
2. Check or uncheck the options under "Options" to select the relevant concepts/terms to be exported.
3. Finally, click the "View Results" link to view the table of the observations.





R Engine Cell: integrating R into the i2b2 software infrastructure

Daniele Segagni, Fulvia Ferrazzi, Cristiana Larizza, et al.

JAMIA published online January 24, 2011
doi: 10.1136/jamia.2010.007914

i2b2 Pavia Query & Analysis Tool Find Patients | Analysis Tools | Message Log | Help | Logout

Navigate Terms **Find Terms**

- Mutations Ontology for Triad
 - Gene
 - ANKB
 - CASQ2
 - Cav 1.2
 - KCNE1
 - KCNE2
 - KCNH2
 - KCNQ1
 - KIR 2.1
 - LMNA
 - PKP-2
 - RYR2
 - SCN5A
 - Locations
- Demographics Ontology for Triad
- Diagnosis Ontology for Triad
 - Primary Diagnosis
 - Inherited Arrhythmogenic Disease
 - Secondary Diagnosis
- Events Ontology for Triad

Workplace

- SHARED
- daniele

Previous Queries

- 61-70 y-Brugada@10:50:32 [11-8-2010] [daniele]
 - Results of 61-70 y-Brugada@10:50:32
 - Patient Set - 203 patients
- Gender@08:28:47 [10-13-2010] [daniele]
- LQTS-Female@08:28:30 [10-13-2010] [daniele]
- LQTS-Female@08:28:13 [10-13-2010] [daniele]

KaplanMeierStat

Drop a Patient Set into the input boxes below, select one event and then click the "run Kaplan Meier" button to retrieve information about the analysis.

Patient Set: 61-70 y-Brugada@10:50:32 [11-8-2010] [daniele] [PATIENTSET_360]

Events:

Syncope

Cardiac Arrest

Appropriate Discharge

Run Kaplan Meier Analysis

Data description:

Group: Female							
time	n.risk	n.event	survival	stdErr	lower 95% CI	upper 95% CI	
52	71	1	0.9859	0.0140	0.9589	0.9589	

Group: Male							
time	n.risk	n.event	survival	stdErr	lower 95% CI	upper 95% CI	
41	130	1	0.9923	0.0077	0.9774	0.9774	
46	129	1	0.9846	0.0108	0.9637	0.9637	
48	128	1	0.9769	0.0132	0.9515	0.9515	
51	125	1	0.9691	0.0152	0.9398	0.9398	
54	121	1	0.9611	0.0171	0.9282	0.9282	
55	119	1	0.9530	0.0187	0.9170	0.9170	
58	95	1	0.9430	0.0210	0.9026	0.9026	
59	80	1	0.9312	0.0239	0.8856	0.8856	
61	56	1	0.9146	0.0286	0.8601	0.8601	
63	36	1	0.8892	0.0375	0.8187	0.8187	
64	27	1	0.8562	0.0484	0.7664	0.7664	

Log-rank test:

Groups	N	Observed	Expected
group: Female	73	1	4.445
group: Male	130	11	7.555

Chisq= 4.247 on 1 degrees of freedom, p= 0.039

Kaplan Meier graph:

Plugins

Detailed List View Category: ALL

- Data Export Tool**
Data Export plugin. Use it for select and export data.
- Kaplan Meier statistical module**
A statistical module based on the R Engine

This is the home of the Related Project - ExportXLS space.

About the ExportXLS Web Client Plugin



ExportXLS

This plugin tabulates unidentified patient data, and applicable diagnoses from specified concepts, of a Patient Set; as well as provides convenient exportation of these data to an Excel spreadsheet.

Introduction

This plugin provides the following functions:

1. Tabulates any selected Concepts (Ontology terms) relevant (observed on) to a set of selected Patients.
2. Optionally displays relevant unidentified Patient data.
3. Exports aforementioned tabulated data into an Excel spreadsheet file conveniently.

Instructions

1. Navigate to the "Specify Data" tab. Then, drag and drop a Patient Set and one or more Concepts (Ontology Term) onto the input boxes.
2. Check or uncheck the *Include Patient Data* box if you want to include the relevant non-identifying information for each patient.
3. Finally, click the "View Results" tab to view the table of the observations.

izb2 Query & Analysis Tool Project: izb2 Onco User: Valentina Tibollo Find Patients | Analysis Tools | Message Log | Help | Logout

Navigate Terms Find Terms

- Benign neoplasms
- Carcinoma in situ
- Malignant neoplasms
 - Admit Diagnosis
 - Principal Diagnosis
 - Secondary Diagnosis
 - TNM Staging
 - Bone, connective tissue, skin, and bre
 - Digestive organs and peritoneum
 - Genitourinary organs
 - Lip, oral cavity, and pharynx
 - Lymphatic and hematopoietic tissue
 - Respiratory and intrathoracic organs
 - Unspecified sites
 - Neoplasms of uncertain behavior
 - Neoplasms of unspecified nature
 - Neurologic Disorders

Workplace

- SHARED
- valentina

Previous Queries

- Positio-Box@15.46.17 [4-6-2012] [valentina]
- Results of Positio-Box@15.46.17 [4-6-2012]
- Patient Set for "Positio-Box@15.46.17"
 - 6069 [57 y/o F]
 - Number of patients for "Positio-Box@15.46.17" [4-6-2012]
- Positio-Box@15.46.17 [4-6-2012] [valentina]

iNavi Plug-in Specify Data View Results Plug-in Help

Patient Set size: 354 Threshold selected: <=1 Similar patients found: 25

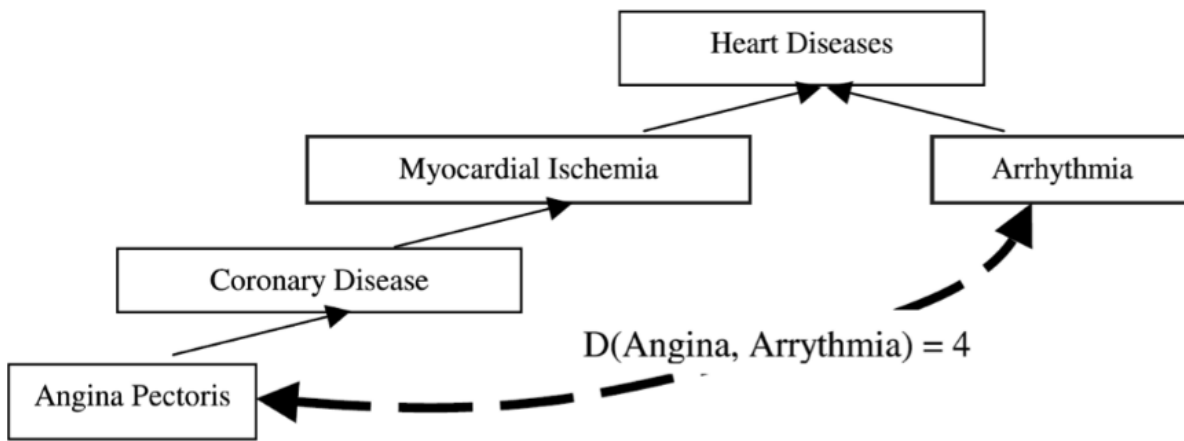
Similar patients list:

256	590	911	1065	1190
1583	2250	2901	3647	3962
4031	4459	4840	5109	5827

Plugins

- Detailed List View
- Demographics (1 Patient Set) - Simple Counts
- Demographics (2 Patient Sets) - Simple Counts

gy Term) onto the input boxes.
tion for each patient.



Reuse of clinical data for assessment and discovery

NEWS UK POLITICS

Everyone 'to be research patient', says David Cameron

BY JONATHAN LEWIS

EVERYONE IN THE UK WILL BE ASKED TO become a research patient, says David Cameron. The Prime Minister has announced that he will ask the public to give up their personal health data to help researchers find better ways to treat and prevent disease.

The PM said the move would be a "historic moment" for the NHS, and would help to speed up the development of new drugs and treatments. He said the data would be used to help researchers understand how the disease works, and how to prevent it.

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Reckoning health in Europe for 2010

EUROPEAN COMMISSION

REPORT

Healthcare in Europe: A report on the state of the sector in 2010

The report highlights the challenges facing the healthcare system in Europe, including the need for more resources, the need for better coordination, and the need for more research and innovation.

2012 European Summit on Trustworthy Reuse of Health Data

EUROPEAN COMMISSION

REPORT

Trustworthy reuse of health data: A transnational perspective

A. Grönlund¹, C. Selten², L. Becker³, B. Rufus⁴, S. Lohse⁵, K. Drenth⁶, A. Cassi⁷, C. Richardson⁸, J. Mannix⁹, F. Murray¹⁰, G. De Moor¹¹

The report discusses the challenges of trustworthy reuse of health data and provides a transnational perspective on the issue.

Identifying Drug Interactions From Adverse-Event Reports: Interaction Between Phenothiazine and Paracetamol Increases Blood Glucose Levels

Journal of Clinical Pharmacy and Therapeutics

The study identifies a potential drug interaction between phenothiazine and paracetamol, which may lead to increased blood glucose levels.

Country	2008	2009	2010	2011
Germany	1000	1100	1200	1300
France	800	900	1000	1100
Italy	600	700	800	900
Spain	400	500	600	700
UK	200	300	400	500

Trustworthy reuse of health data: A transnational perspective

Health Data

Health data is a valuable resource for researchers and clinicians. It can be used to identify trends, assess risk, and improve patient care.

Health data can be used in a variety of ways, including:

- Identifying trends and patterns in disease incidence
- Assessing the effectiveness of treatments and interventions
- Improving patient care and outcomes

Everyone 'to be research patient', says David Cameron

COMMENTS (654)

Every NHS patient should be a "research patient" with their medical details "opened up" to private healthcare firms, says David Cameron.

The PM says it will mean all those who use the NHS in England will be helping in the fight against disease.

He hopes the result will be that patients get faster access to new treatments and Britain's life sciences sector will become a world leader.

But critics say commercial interests are being put ahead of patient privacy.

In a speech in London Mr Cameron said he would consult on changing the NHS constitution, which governs how the the health service is run, so that all patients' data is used for medical research unless they want to opt out.

'Anonymous data'



David Cameron has said he wants to make huge numbers of patient records in England available to the drugs industry

Related Stories

[NHS-life sciences partnership](#)

[Private firms 'to share NHS data'](#)

[Challenges ahead over free data](#)

eHealth Task Force Report

Redesigning health in Europe for 2020



Lever for change #2:
Liberate the data



Data can be compared to oil. In the ground it is unusable and worthless. Extracted and refined, it has huge value. Large amounts of data currently sit in different silos within health and social care systems. If this data is released in an appropriate manner and used effectively it could transform the way that care is provided.



2012 European Summit on Trustworthy Reuse of Health Data

May 14-15, 2012 | Brussels, Belgium | <http://euhealthdata2012.imia.info>



INTERNATIONAL JOURNAL OF MEDICAL INFORMATICS 82 (2013) 1-9



ELSEVIER

journal homepage: www.ijmijournal.com



Trustworthy reuse of health data: A transnational perspective

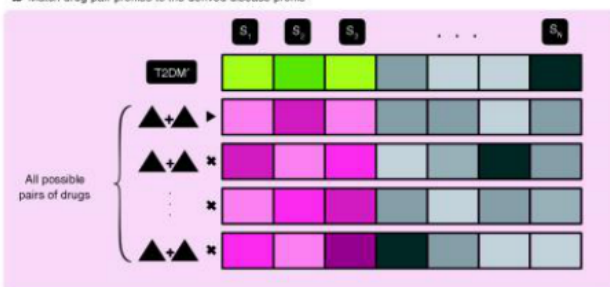
A. Geissbuhler^{a,b,*}, C. Safran^c, I. Buchan^d, R. Bellazzi^{a,e}, S. Labkoff^f, K. Eilenberg^g, A. Leese^h, C. Richardsonⁱ, J. Mantas^{a,j}, P. Murray^a, G. De Moor^k



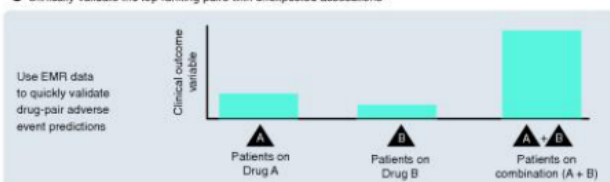
a Learn a disease "symptom" profile that uniquely identifies the given disease



b Match drug-pair profiles to the derived disease profile



c Clinically validate the top ranking pairs with unexpected associations



Clin Pharmacol Ther. 2011 July ; 90(1): 133–142. doi:10.1038/clpt.2011.83.

Detecting Drug Interactions From Adverse-Event Reports: Interaction Between Paroxetine and Pravastatin Increases Blood Glucose Levels

NP Tatonetti^{1,2,3}, JC Denny^{4,5}, SN Murphy^{6,7}, GH Fernald^{1,2,3}, G Krishnan⁸, V Castro⁶, P Yue⁹, PS Tsau⁹, I Kohane^{7,10,11}, DM Roden⁵, and RB Altman^{2,3}

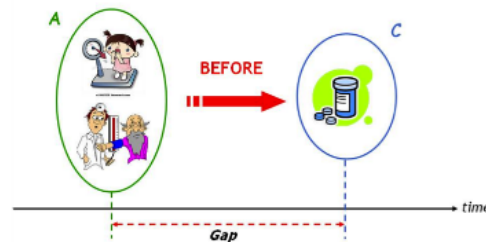
I2b2	Patients	Visits	Observations
Onco-i2b2	28838	171302	623214
Cardio-i2b2	6334	15094	272310



Biobank	Blood	Plasma	Tumoral Tissue	Normal Tissue
Samples	1710	855	2338	1618
Patients	847	847	556	428

Temporal data mining on 8969 breast cancer patients

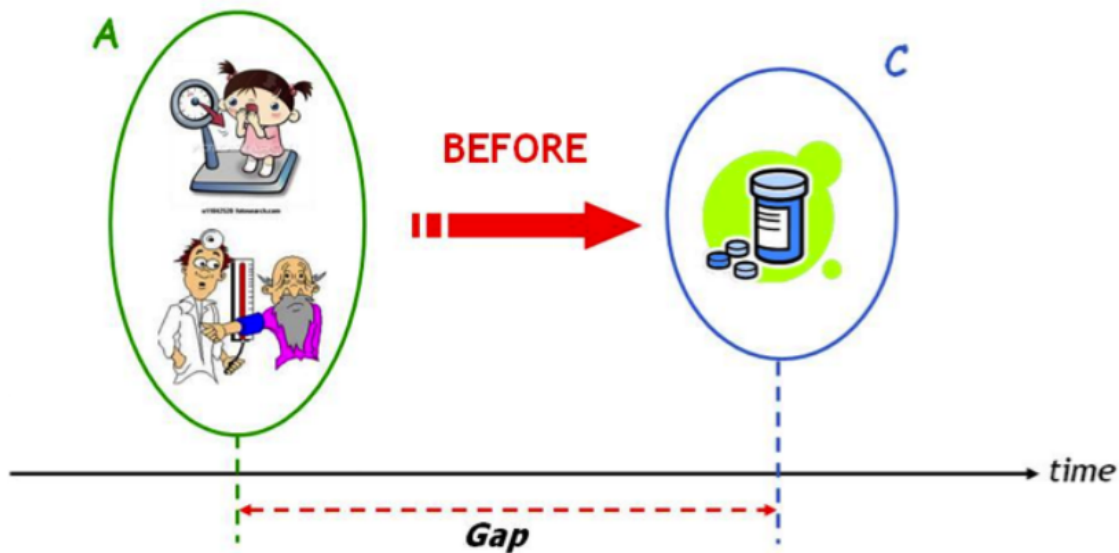
BMI 25-30 (overweight) AND SBP >=180 (hypertension)
BEFORE ACE inhibitors



- 11 careflow patterns
- Highlight a high level of guidelines implementation
- 43% of hospitalized patients - surgery only
- 65% of non hospitalized patients had regular chemotherapy cycles
- 71% of them underwent an ECG before performing a chemotherapy in the same hospitalization

Temporal data mining on 8969 breast cancer patients

BMI 25-30 (overweight) AND SBP \geq 180 (hypertension)
BEFORE ACE inhibitors



11 careflow patterns

Highlight a high level of guidelines implementation

43% of hospitalized patients - surgery only

65% of non hospitalized patients had regular chemotherapy cycles

71% of them underwent an ECG before performing a chemotherapy in the same hospitalization



About Version 1.6.1

Eureka! Clinical Analytics is a project of the [Center for Comprehensive Informatics](#) and [Department of Biomedical Informatics](#) at [Emory University](#). It was conceived as part of the vision of the [CardioVascular Research Grid \(CVRG\)](#) to create tools that enable researchers to analyze and manipulate their biomedical research data in the cloud.

What does it do?

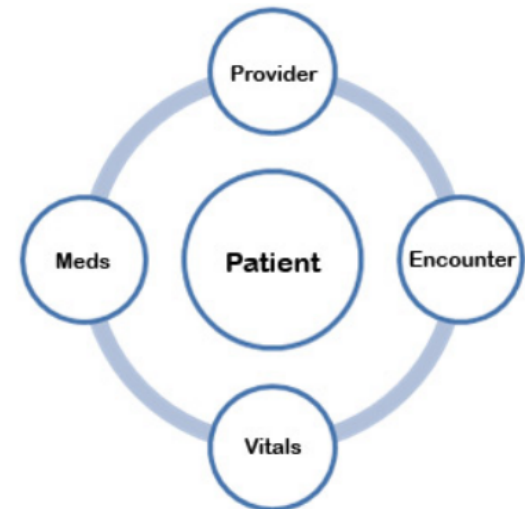
Eureka! Clinical Analytics provides you with an easy way to phenotype a patient population based on their clinical and administrative data, and load those phenotypes and data into your own instance of [i2b2](#). Once in i2b2, you and your colleagues can ask questions about the data, summarize data and download subsets of your data for your research. You provide your data in an Excel spreadsheet meeting certain criteria, and the software takes care of the rest. While its functionality is targeted to clinical datasets in cardiovascular disease research, it supports loading a wide variety of clinical data types into i2b2.

Can I load data with identifiers in it?

No, this is a demonstration website only. Please see the End-user agreement when you register for an account on the website for details.

What do you mean I can't load data with identifiers on it?

If you need to load real patient data into Eureka!, you must deploy your own instance of the software in a [HIPAA-compliant](#) environment. See the [Get the Software page](#) on the Eureka! website for details. We provide a [distribution of Eureka! for local installation](#) and an [Amazon Machine Instance](#) for you to clone on the [Amazon Elastic Compute \(EC2\) Cloud](#).



The new challenges



Redis, MongoDB, Cassandra, Riak, CouchDB, Neo4j, Membase



Scalable machine learning and data mining
mahout

Big Data Analytics Stories: Genomic Sequencing
The challenge of Genomic Sequencing at Oracle BI is using big data to feed better treatments for patients through genetic sequencing technology.



Omar Tawakol
CEO, Oracle

The Death of the Data Warehouse and the Age of Activation

Posted 2/12/2012 4:41 PM

Filed In: Insights, Oracle, Typical, Query, Challenges, Analytics, Innovation, Information

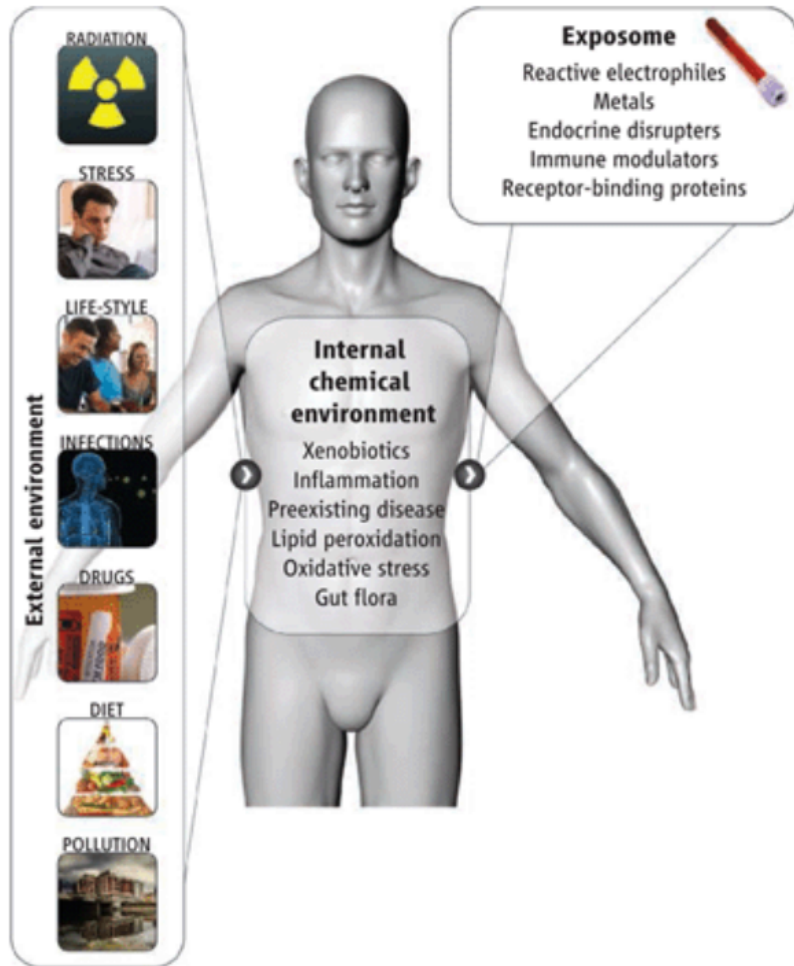
Follow: Database, Big Data, Data, Data Activation, Data Warehouse, Business News

An open-source community begins to gain traction in the NoSQL developer's marketplace.

March 8, 2012: The solution of future code measurements encompasses data storage redesign, including moving large part of the experiment data to a NoSQL DB for high performance and scalable management.

NoSQL vs SQL, Why Not Both?

By: Steve Hurler



Science 22 October 2010:
 Vol. 330 no. 6003 pp. 460–461
 DOI: 10.1126/science.1192603

PERSPECTIVE

EPIDEMIOLOGY

Environment and Disease Risks

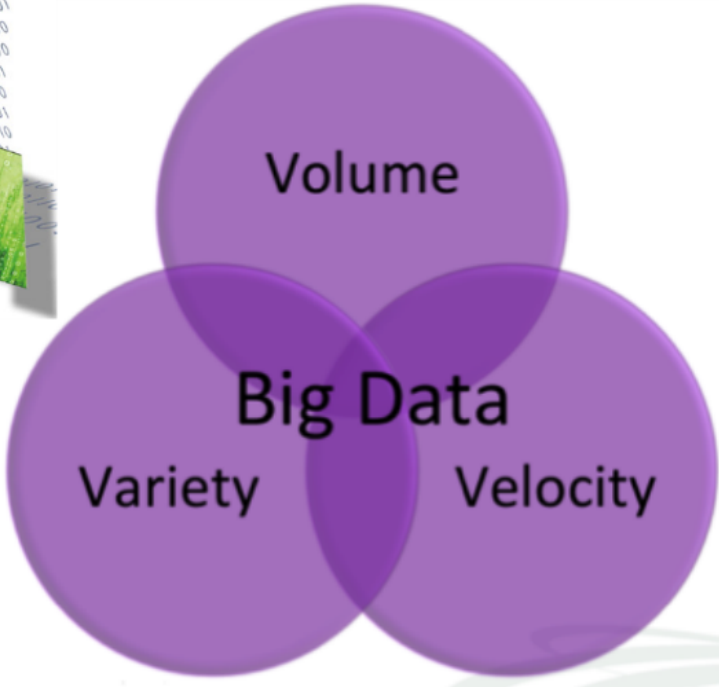
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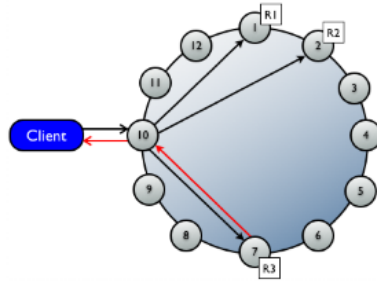
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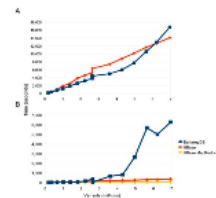


Scalable machine learning and data mining

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Big Data Analytics Stories: Genomic Sequencing

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Relax with CouchDB — Into the non-relational DBMS era of bioinformatics
 Gauri G. Matsum¹, Mihir A. Patil^{2,3}, Jack A. Smith¹, James W. Blanton², Kevin R. Coombes^{4*}
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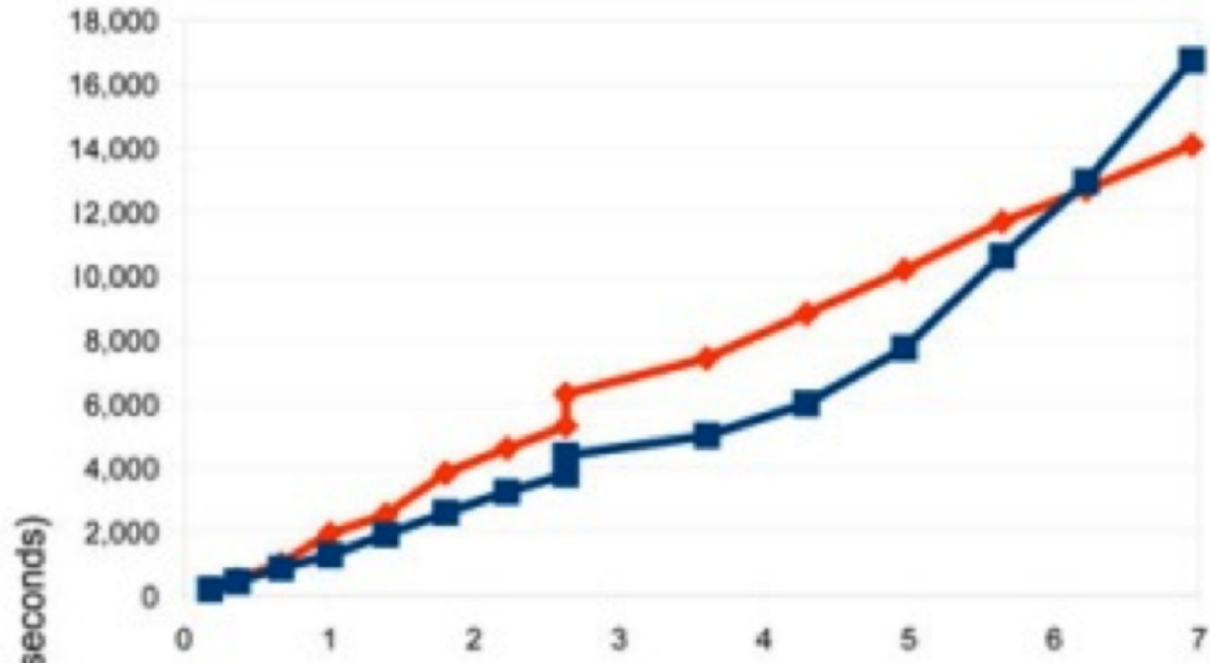
Ganiraju Manyam ^{a,1}, Michelle A. Payton ^{a,b,1}, Jack A. Roth ^c, Lynne V. Abruzzo ^b, Kevin R. Coombes ^{a,*}

^a Department of Bioinformatics and Computational Biology, The University of Texas MD Anderson Cancer Center, Houston, TX 77030, USA

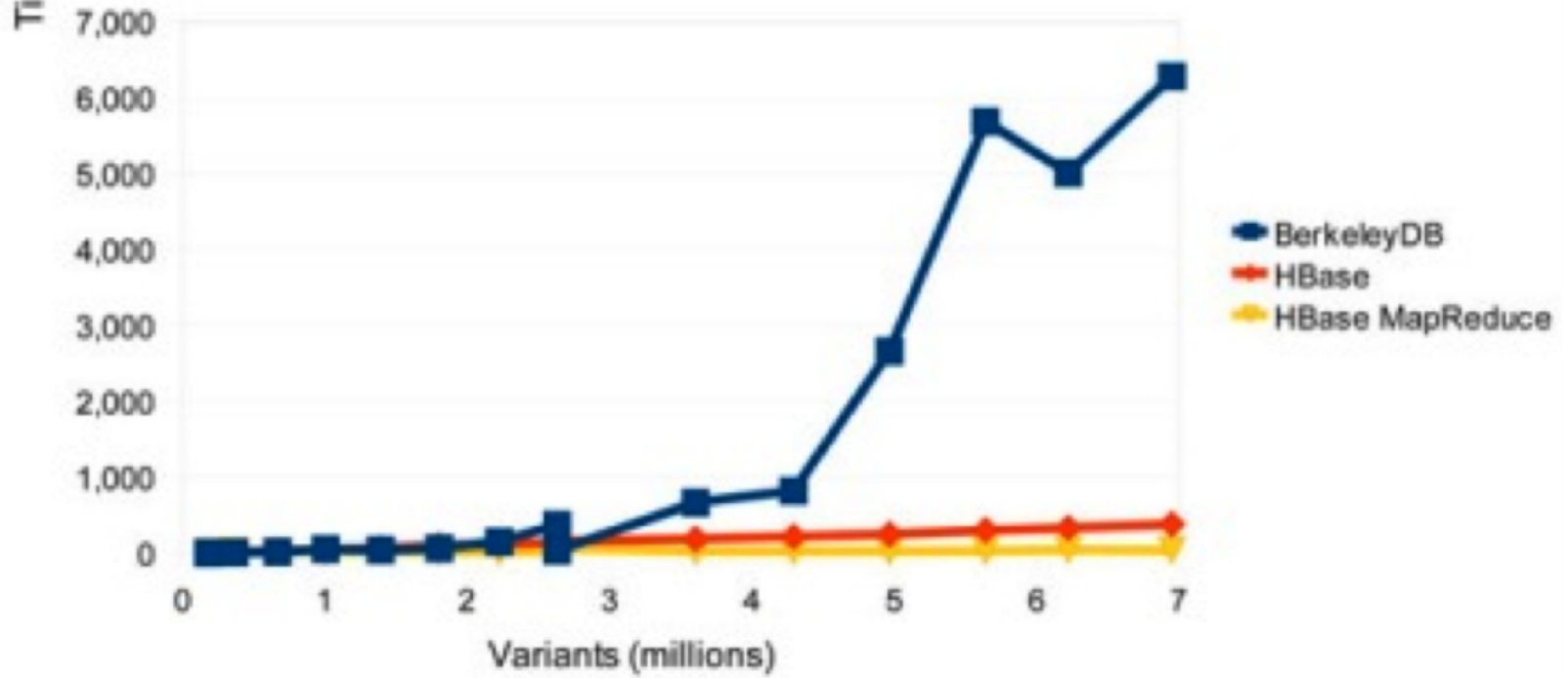
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By Alaric Snell-Pym

Directions:

Integration of NOSQL technologies

Implementation of Shrine-based initiatives

Extensions to manage temporal abstractions

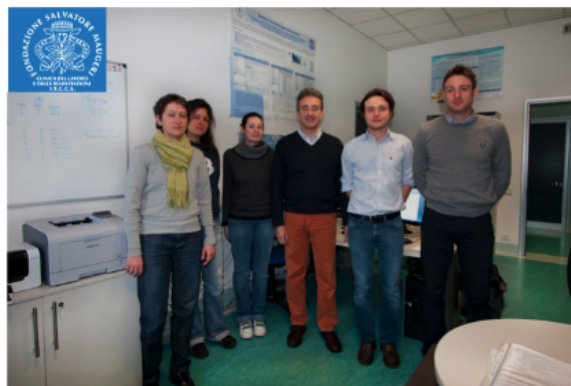


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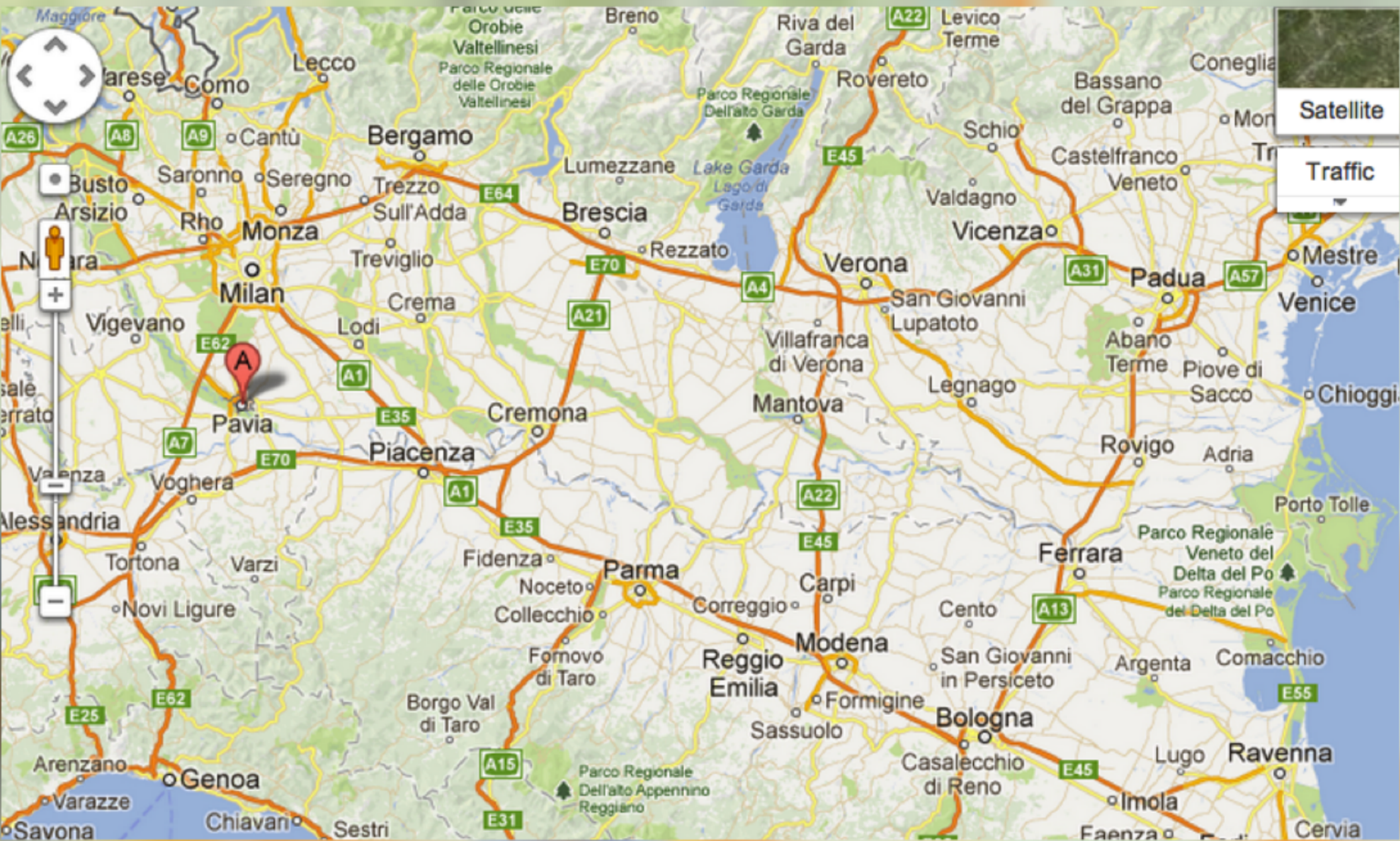
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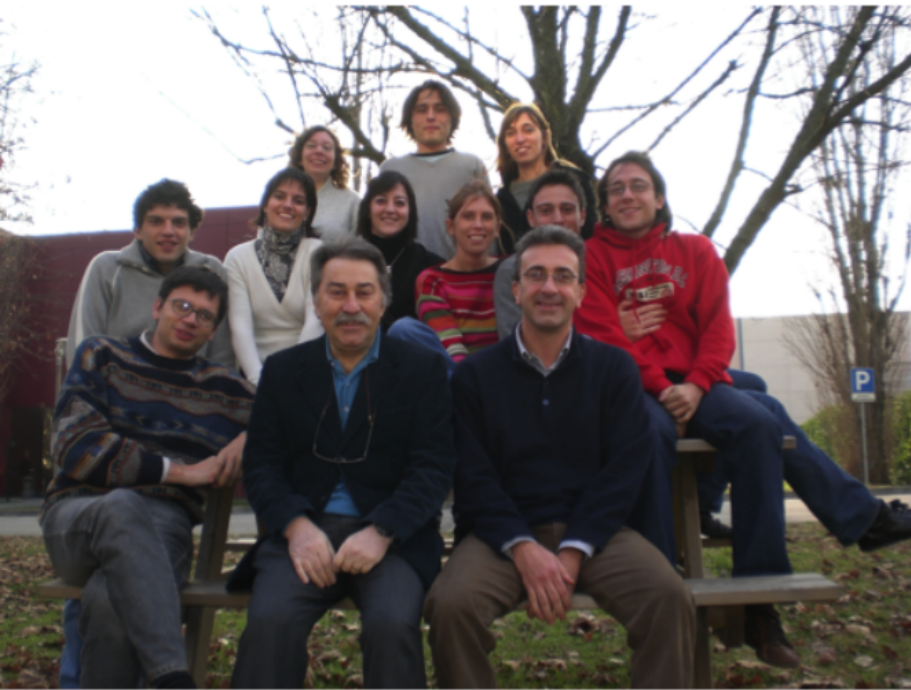




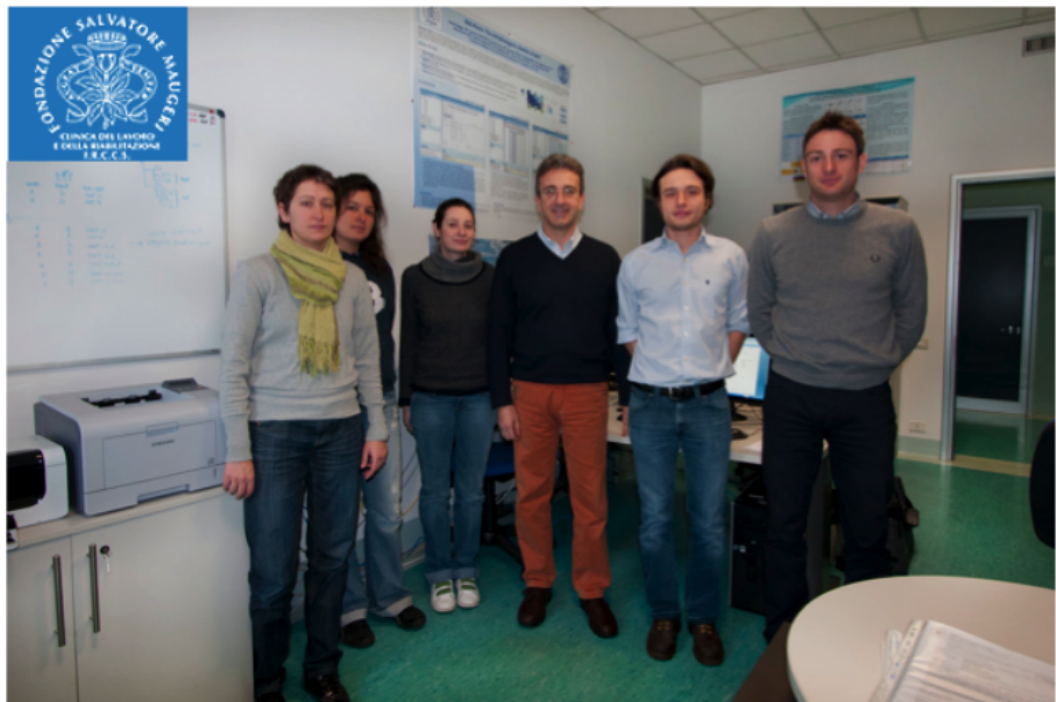








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Thank you ...



Lelio Menozzi - Pavia from Porta Calcinara

