

10a Jornada de
Recerca de l'ICS

11a Jornada de
Recerca de l'IDIAP



La recerca amb
grans bases de
dades clíniques
millora la salut

7 de juny de 2018

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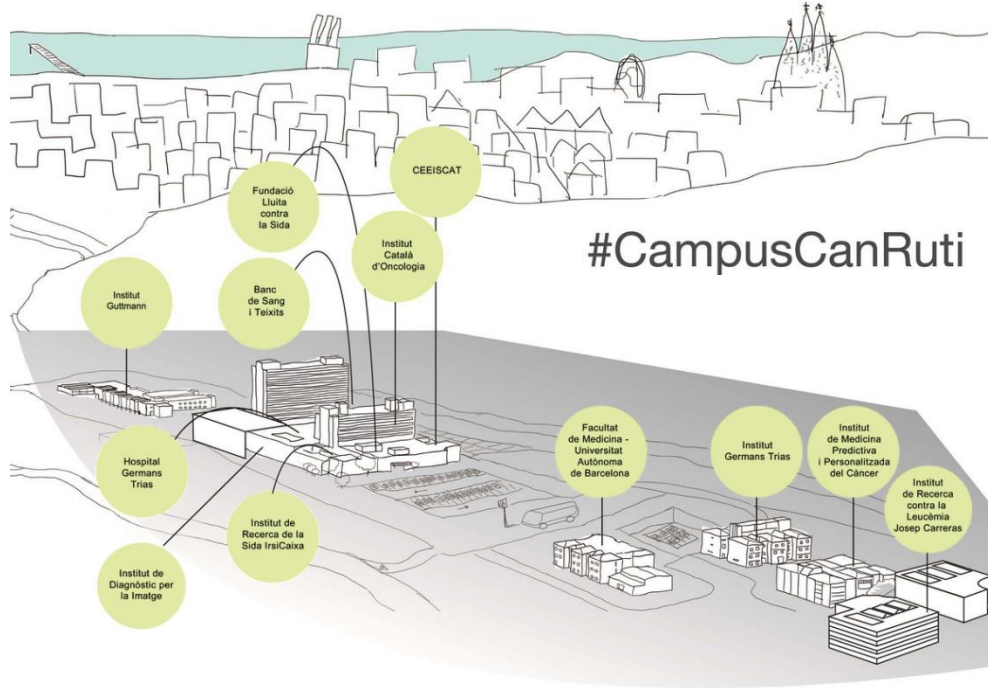
COSMOCAIXA



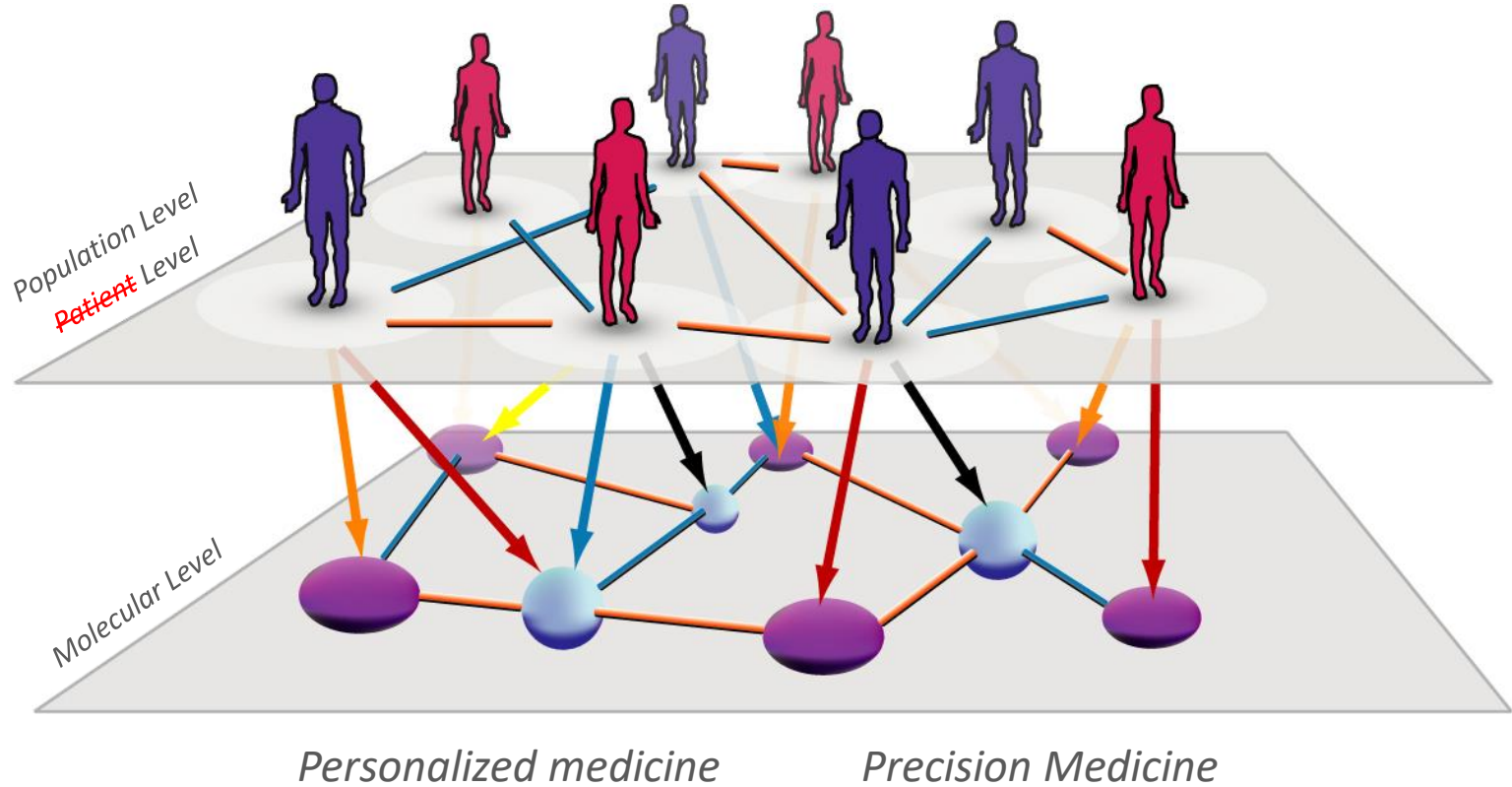
A holistic approach to common diseases

Rafael de Cid
Genomes for life-GCAT lab
PMPPC -IGTP

Personalized Medicine



Common diseases



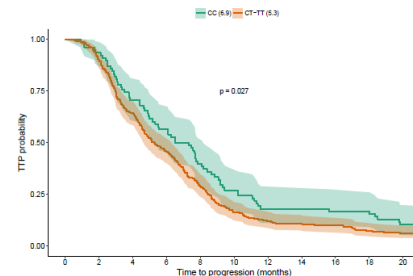
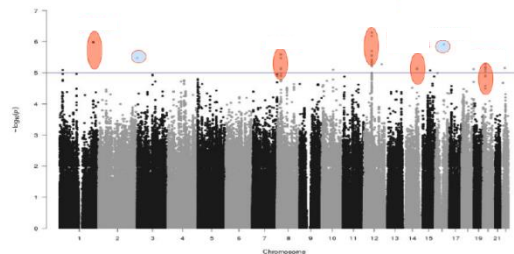


common diseases **and all human biomedical traits**

“Identifying genetic correlations between complex traits and diseases **will** **are** provide**ing** useful etiological insights and help prioritize likely causal relationships”

Genomic profiling in advanced stage non-small-cell lung cancer patients with platinum-based chemotherapy identifies germline variants with prognostic value in *SMYD2*

Iván Galván-Femenía ^a, Marta Guindo ^b, Xavier Duran ^a, Sílvia Calabuig-Fariñas ^{c, d, e}, Josep Maria Mercader ^{b, 1, 2}, Jose Luis Ramirez ^f, Rafael Rosell ^g, David Torrents ^{b, g}, Anna Carreras ^a, Takashi Kohno ^h, Eloisa Jantus-Lewintre ^{d, e, 1}, Carlos Camps ^{c, d, j, k}, Manuel Perucho ^f, Lauro Sumoy ^l, Jun Yokota ^f, Rafael de Cid ^{a, k}



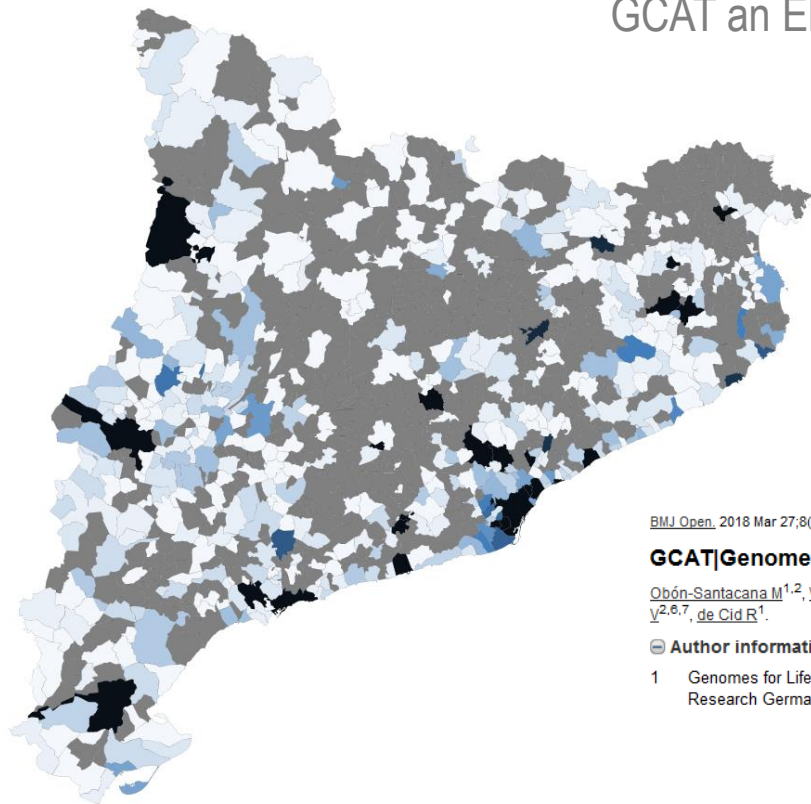


“The limiting factor in progress towards a more complete understanding of the relationship between genome and phenome ~~will be~~ **was** the availability of high-dimensional phenotype data”

Gratten et Vischer. Genome Medicine, 2016.



GCAT an Electronic Health Record-linked cohort (2014-2017)



[BMJ Open](#), 2018 Mar 27;8(3):e018324. doi: 10.1136/bmjopen-2017-018324.

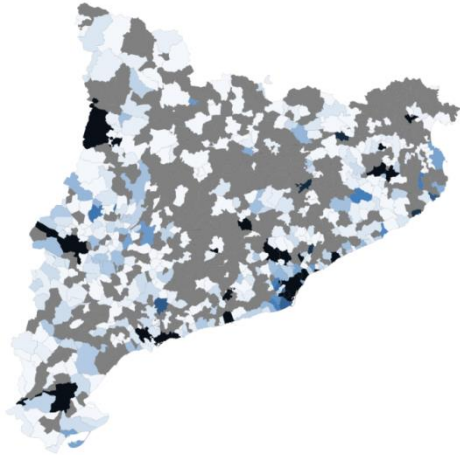
GCAT|Genomes for life: a prospective cohort study of the genomes of Catalonia.

[Obón-Santacana M](#)^{1,2}, [Vilardell M](#)¹, [Carreras A](#)¹, [Duran X](#)¹, [Velasco J](#)¹, [Galván-Femenía I](#)¹, [Alonso T](#)¹, [Pujol L](#)³, [Sumoy L](#)⁴, [Duell EJ](#)⁵, [Perucho M](#)⁴, [Moreno V](#)^{2,6,7}, [de Cid R](#)¹.

Author information

- 1 Genomes for Life -GCAT lab Group, Program of Predictive and Personalized Medicine of Cancer (PMPPC), Institute for Health Science Research Germans Trias i Pujol (IGTP), Badalona, Spain.

Open Active resource



GCAT STARTS THE FIRST HEALTH FOLLOW-UP

- LIFESTYLE
- FAMILY HEALTH
- ADDITIONAL RESEARCH

GCAT SUMMARY DATA

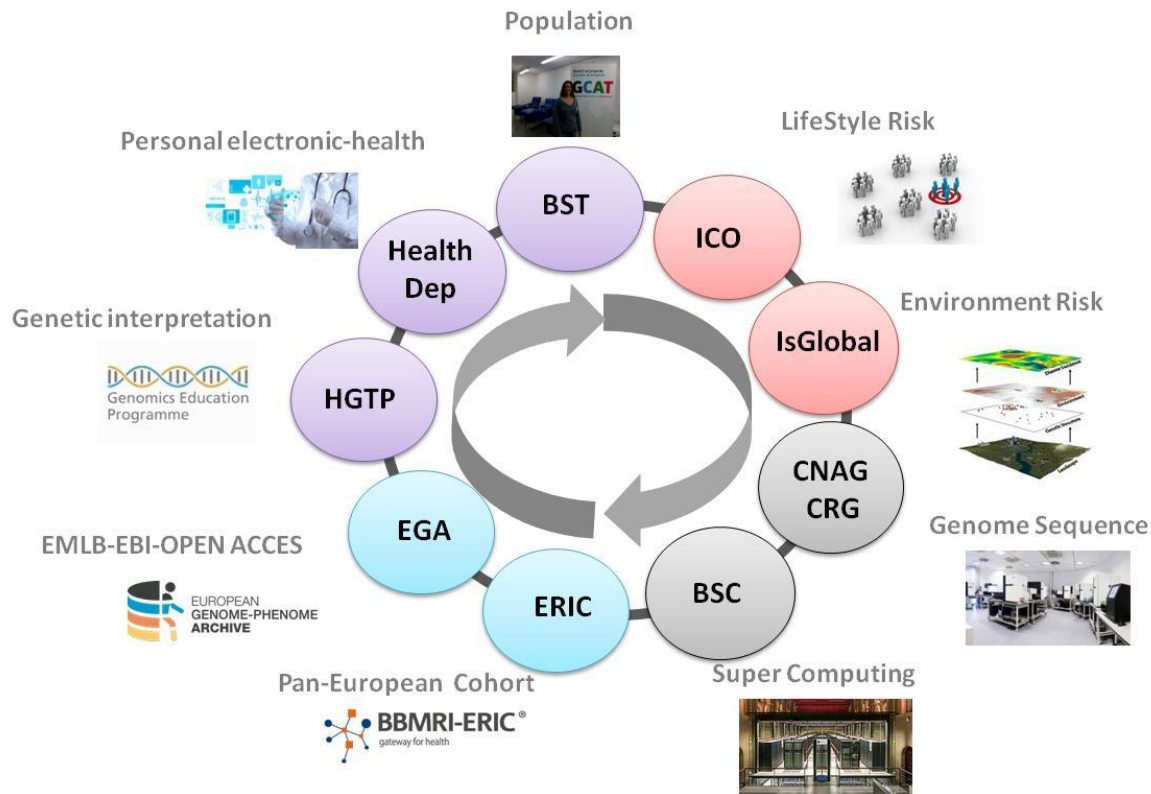
The GCAT data variables definition as well as summary data can be browsed on the [GCAT variable catalogue](#) to help identify variables of interest to support your collaborative research projects.

A short video tutorial showing how to use the variable browser is available below (use CC button for video captions).

GCAT variable search from GCAT ' Genomes for Life, on Vimeo.

www.genomesforlife.com

Big Data / Complex Data



Genomic correlation



Scenario 1

Trait A



Allele a

Trait B



Allele b

Trait C



Allele c

Trait D



Allele d

Trait N



Allele n

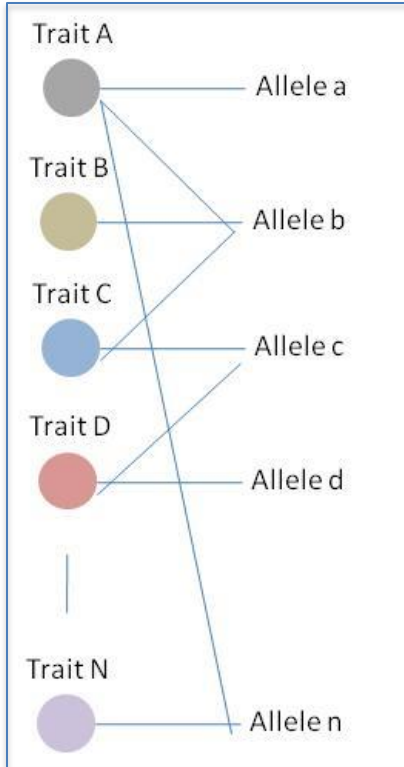


GWAS Hits

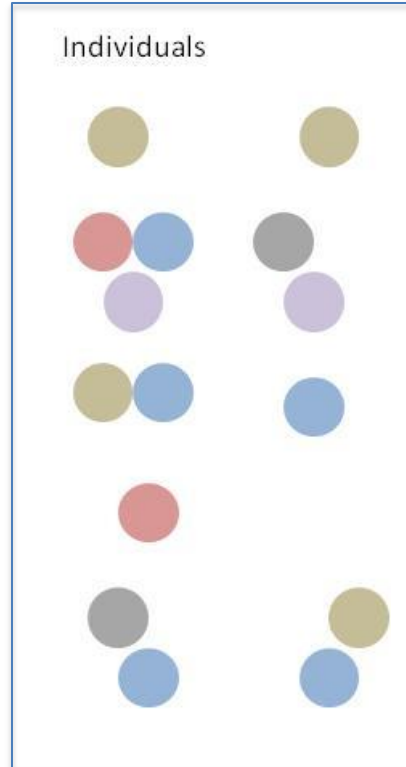
Pleiotropy Multimorbidity Trajectories



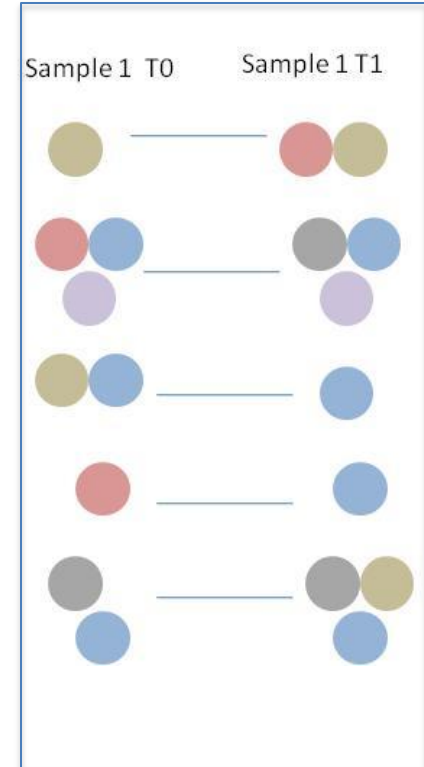
Scenario 2



Scenario 3



Scenario 4

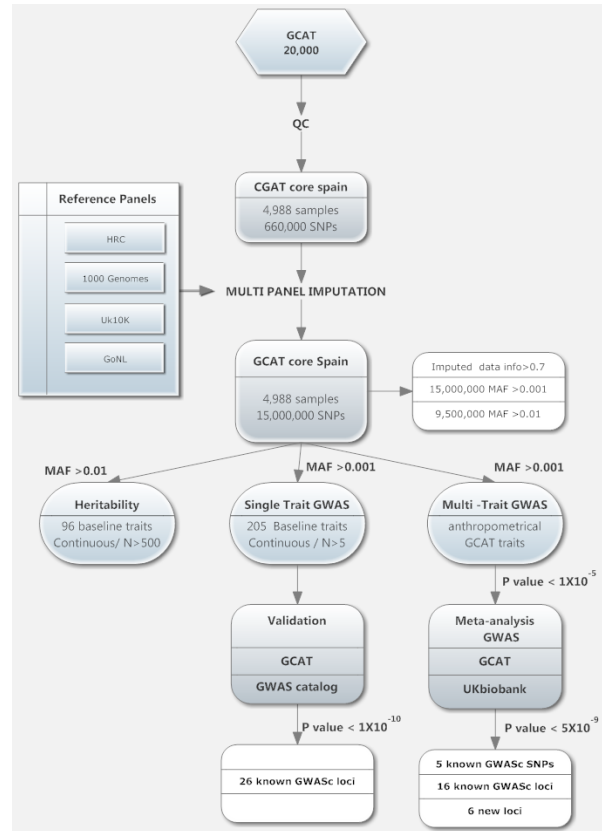


Shared Genomic Variants_population wide disease



GCAT core*

15.000.000 Variants
205 traits (baseline)
5.000 individuals

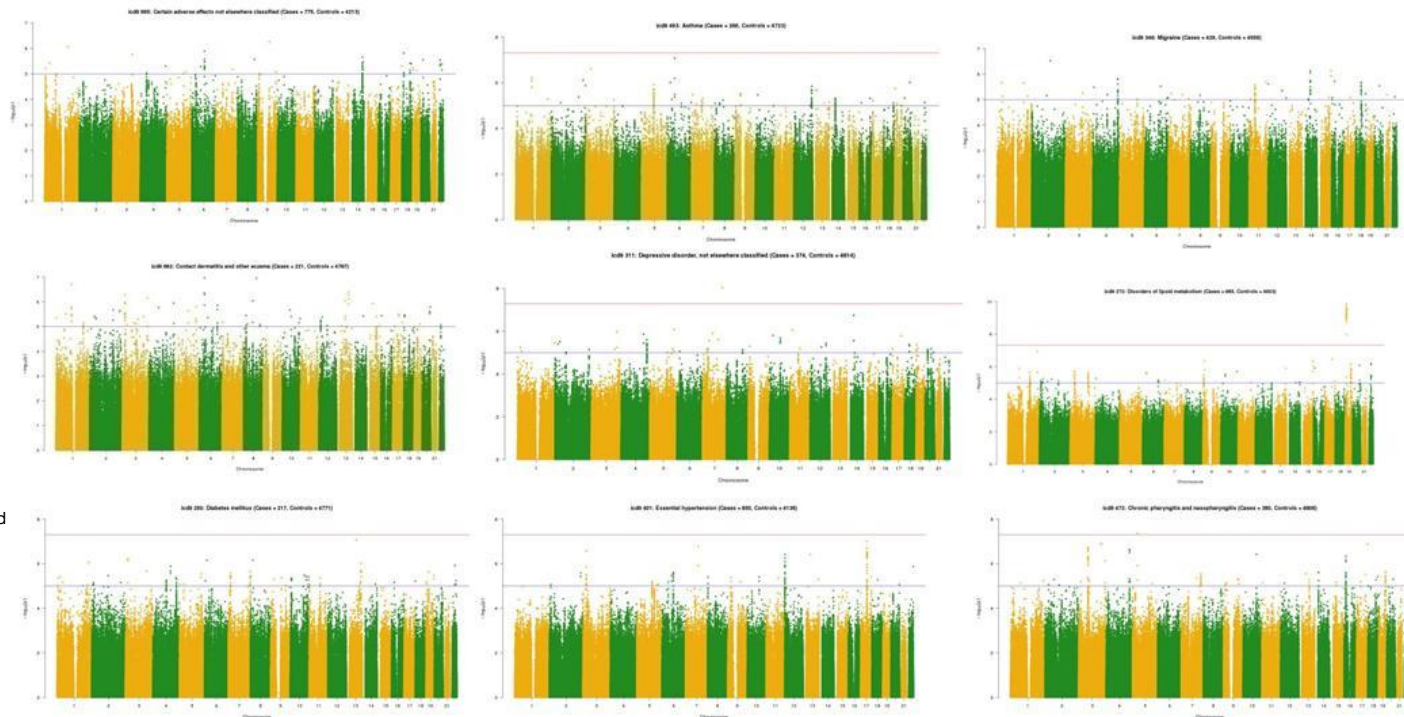


*Multi-trait-genome association analysis identifies new susceptibility genes for human anthropometric variation in the GCAT cohort.

Galván-Femenía et al, J. Med Gen. 2018 under review

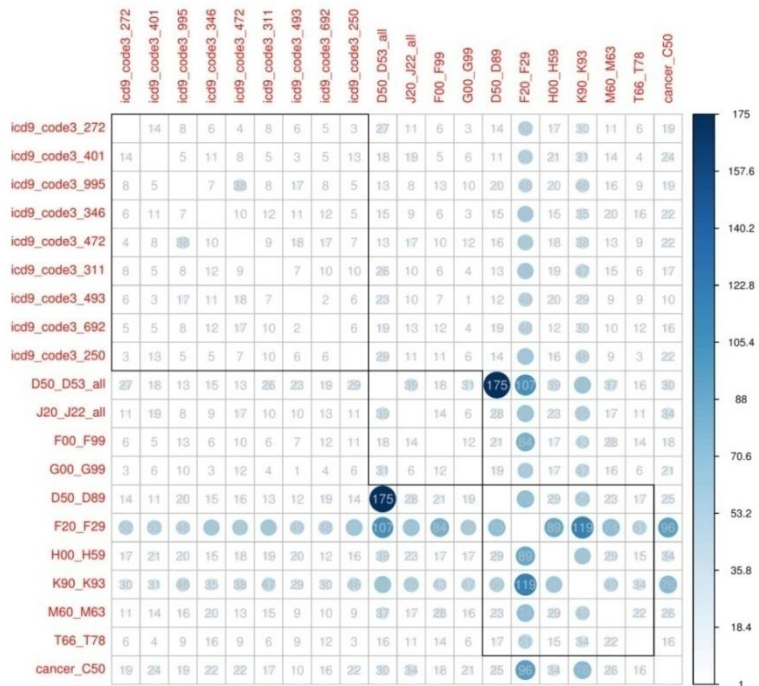


Identification of Shared Common Variants (ICD-9 coded diseases)



icd9 272: Disorders of lipid metabolism
icd9 401: Essential hypertension
icd9 995: Certain adverse effects not elsewhere classified
icd9 346: Migraine
icd9 472: Chronic pharyngitis and nasopharyngitis
icd9 311: Depressive disorder, not elsewhere classified
icd9 493: Asthma
icd9 692: Contact dermatitis and other eczema
icd9 250: Diabetes mellitus

Pair-wise GWAs_disease linkages



- icd9 272: Disorders of lipid metabolism
- icd9 401: Essential hypertension
- icd9 995: Certain adverse effects not elsewhere classified
- icd9 346: Migraine
- icd9 472: Chronic pharyngitis and nasopharyngitis
- icd9 311: Depressive disorder, not elsewhere classified
- icd9 493: Asthma
- icd9 692: Contact dermatitis and other eczema
- icd9 250: Diabetes mellitus
- D50-D53: Nutritional anaemias
- F00-F99: Mental and behavioural disorders
- G00-G99: Diseases of the nervous system
- D50-D89: Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- F20-F29: Schizophrenia, schizotypal and delusional disorders
- H00-H59: Diseases of the eye and adnexa
- K90-K93: Other diseases of the digestive system
- M60-M63: Disorders of muscles
- T66-T78: Other and unspecified effects of external causes
- cancer C50: Breast cancer

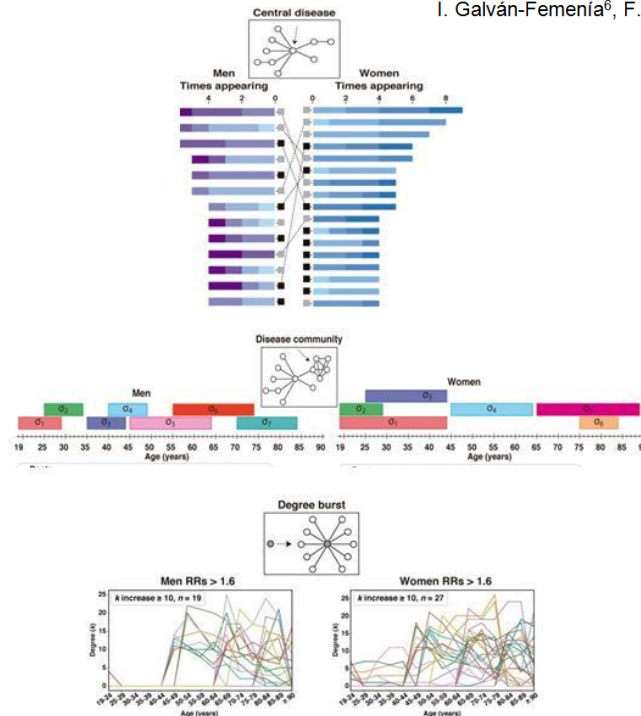
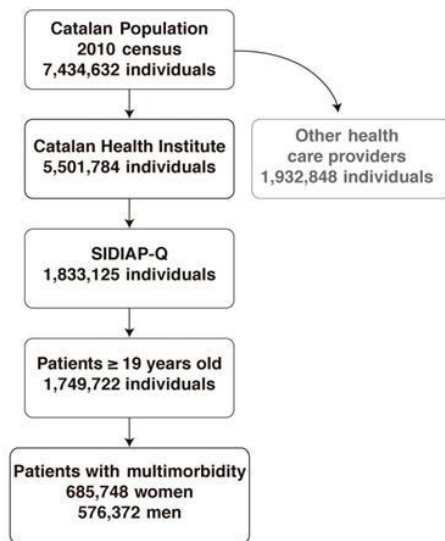
profiling of multimorbidity_ Key diseases



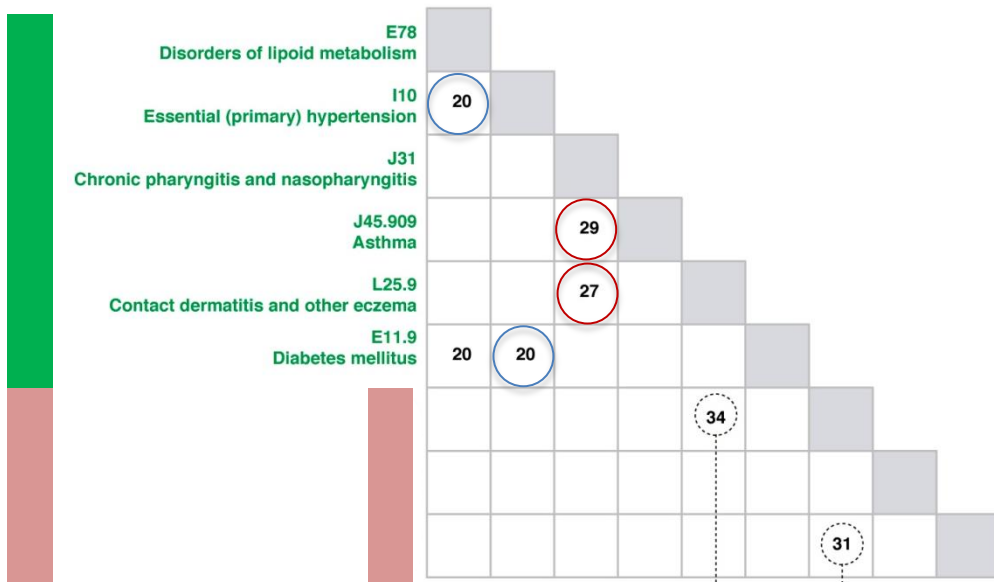
Topological analysis of disease networks in SIDIAP database

A. Amell¹, A. Roso-Llorach^{2,3}, L. Palomero⁴, D. Cuadras⁵, J. Serra-Musach⁴,
I. Galván-Femenía⁶, F. Comellas¹, R. de Cid^{6,*}, M.A. Pujana^{4,*} & C. Violán^{2,3,*}

(Under review , Plos Genetics, 2018)



Genetic profiling_key diseases prediction



Common conditions

Known relationships



E78-I10-E11.9

HTA-DM-TGD



J45-J31

Asthma- Chronic Pharyngitis

Key Diseases

Unexpected correlations

Nutrition-Nervous system

Men 65-6 RR = 2.91
Women 75-79 RR = 2.35

Men 65-69 RR = 1.52
Women 65-69 RR = 2.02



- GCAT | Genomes for Life is a big-data project
 - Population based
 - Personal Omic Data
 - Personal Clinical data
 - Sensible data

- GCAT is a Resource Recovery and Safe Reuse (RRR)
 - Reuse information
 - Reduce inversions

- GCAT will represent a powerful tool for holistic approaches to disease
 - Stratification by disease trajectories and key diseases
 - Knowledge of hidden disease linkage association



Colaborators



Funding



www.genomesforlife.com



Generalitat de Catalunya
Departament de Salut



Institut Català
de la Salut



IDIAP
Jordi Gol

ics.gencat.cat 