Building genomics

genomDE : National and European initiatives

30th November 2020 DIGITAL EVENT



Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020

Supported by the European Union's Structural Reform Support Programme (SRSP) and implemented in cooperation with the European Commission's Directorate-General for Structural Reform Support (DG REFORM)





Federal Ministry of Health





Spain: Genome Medicine projects

Alfonso Valencia. Ph.D. ICREA Professor Dir. Life Sciences Dept. BSC Dir. Spanish Bioinformatics Institute INB-ISCIII ELIXIR-ES



Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020

Supported by the European Union's Structural Reform Support Programme (SRSP) and implemented in cooperation with the European Commission's Directorate-General for Structural Reform Support (DG REFORM)





Federal Ministry of Health





Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020



Alfonso Valencia. Ph.D. ICREA Professor Dir. Life Sciences Dept. BSC Dir. Spanish Bioinformatics Institute INB-ISCIII ELIXIR-ES



Barcelona Supercomputing Center Centro Nacional de Supercomputación



Content

- Structure of the Spanish public health system
- Organization of the National Infrastructure for Precision Medicine, IMPaCT
 - IMPaCT Projects: Predictive Medicine, Genomic Medicine and Data
 - The Spanish National Bioinformatics Institute/ELIXIR-ES and the network of bioinformaticians in Spanish hospitals (TRANSBIONET)
 - EGA, beacons and access to information from EHRs
- European COVID-19 Portal (EMBL-EBI and ELIXIR) and the Spanish branch

01

The public health system

genomDE : National and European initiatives 30TH NOVEMBER 2020

The public health system



46,6 M inhabitants

A completely decentralised system with 17 autonomous regions

Hospitals and primary care belong and are financed by the local governments

The national ministry of Health is only responsible of coordination and representation

National Health System 466 hospitals (322 public dependence) and 10.081 primary care centers.

3,1 meds for 1000 inhabitants Two times more in hospitals than in primary care

POSITIVE OPINION but it is not sustainable

67.6% of citizens favourably value the functioning of the Spanish health system, considering that it works quite well or well.

The degree of satisfaction with the operation of the public health system in Spain is rated 6.7 points out of 10.

Primary Care continues to be the best-rated level of care with 7.3 points out of 10, maintaining the trend started in 2015.

Life expectation is (was) 83,4 years. Proportion of dependents / working 53,4% growing in 7.7 since 2000. Pressure of the private systems (one of the largest in Europe in proportion)

101.721 M€ 9,1% GDP. (72.402 public - 29.319 private). The system needs 12.000 M€ - **1% of the GDP** to be in the EU average of 9,9.

8

Building genomics genomDE : National and European initiatives 30TH NOVEMBER 2020

Hetereogenous data and data models

- CANTABR ASTURIAS GALICI LA RION CATALUNA **CASTILLA Y LEÓN** ARAGÓN COMUNIDAD VALENCIANA CASTILLA-LA MANCHA EXTREMADURA ANDALUCIA ISLAS CANARI CEUTA
- Andalusia- the largest health regions in Europe- completely public

>> All EHR and drug admin integrated and operational

 - Catalunya- the oldest one privately own hospitals are part of the public health system
 > all EHR and drug admin integrated but heterogenous and difficult to operate

 Madrid- public hospitals together with privately managed hospitals with public doctors, nurses and services.
 > Very difficult of integrations

A general health database (BDPP-SNS) and tools for the interoperability and traceability projects for clinical information including electronic prescriptions (SNS-HCDSNS-) 42,129 persons - **93.2% of the population.**

The decision system based on committees



Decisions taken by an Interterritorial Council composed of: - Ministry of Health & - The responsible of the 17 health systems

decisions taken by unanimity and issued as "recommendations"

Advisory and Technical decision bodies:

- National Medicament Agency (Spanish branch of EMA)
- Committee of the Agencies for the Evaluation of the quality of the system
- Technical advisory group with the heads of Health Informatics of the 17 autonomous regions

Science in the Spanish health System Instituto de Salud Carlos III

Autonomous public Spanish *Belongs to the science Ministry s the Minister of Health is the president of the government council*

- Carries out Intramural Research with its own institutes
- Finances research projects in biomedicine in Spain
- Represents Spain in the international institutions
- Participates in Public Health decisions





02

Organization of the National Infrastructure for Precision Medicine, IMPaCT



genomDE : National and European initiatives 30TH NOVEMBER 2020

Regional Genomic Medicine Projects: the case of Catalunya

In 2016 created the General Directorate of Research and Innovation in Health and the Strategic Plan for Research and Innovation in Health (PERIS by its acronym in Catalan).

- Promote the participation of patients and citizens in the research and innovation policies.
- Increase the quality of research in the health field to guarantee the excellence of the health system.
- Intensify the capacities to transfer the knowledge generated by researchers and technologists to healthcare
 processes in the areas of prevention, diagnosis and treatment of pathological processes and in the promotion of
 health.
- Strengthen the capacities and international leadership of the research centers of Catalonia.
- Promote the training and employability of scientists and technologists and strengthen the scientific capacities of health professionals.
- Integrate health research and innovation policies with other existing ones in Catalonia

24 research projects aimed at Primary Health Care.

16 Projects in Personalized Medicine, Regenerative Medicine, Cohort Study, and Independent Cooperative Clinical Research,

20 research projects in Mental Health.

Research contracts 65 PhDs, 89 nursing professionals and 31 specialist doctors.

IMPaCT project Building the framework for the national Personalized Medicine initiative by the Health Ministry

The Government approves the call to launch the new Precision Medicine infrastructure, IMPaCT- with an initial Budget of 25.8 million €.

It is the first step to launch the Spanish Strategy for Personalized Medicine to be announced in 2021.

It is part of the objective of strengthening the bio-sanitary and public health research system, defined in the Shock Plan for Science and Innovation

It includes three new programs to be managed by the ISCIII:

- Predictive Medicine,
- Genomic Medicine and
- Data Science

Predictive Medicine, is aimed at the design and implementation of a **large population cohort with clinical, epidemiological and biological data**, measured at the individual level, which allows representing the entire population residing in Spain, including ethnic variability and geographic and environmental diversity. The idea is to generate a record of individual and population data that is the basis for better decision-making in health; It will make it possible to build predictive models of disease, identify health inequalities, monitor key indicators, and evaluate the impact of health policies.

Genomic Medicine, will develop coordination infrastructures and protocols **to carry out genomic analyses and other 'omic' data in an effective, efficient and equitable accessible manner throughout the national territory**, taking as support large Spanish research centers that they already have state-ofthe-art sequencing technology and experience in its application to the diagnosis of human diseases. Its purpose is to guarantee a strategic state reserve of very complex diagnostic sequencing services, which will be provided at no cost to patients or regional health services; It will optimize and strengthen the available massive sequencing capacities, orienting them to the needs of genetic diagnosis - exomes, complete genomes ... – and paying special attention to the early molecular diagnosis of rare diseases.

CANTABRI **FPGMX** Navarra **Biotech CNAG** COMUNIDAD **CIBER-**VALENCIAN Madrid FPS CEUTA MELILLA

Genomic Medicine

Use cases:

1) RD; sequencing of exomes or genomes for the diagnosis of patients with **rare disease**s;

Building genomics

genomDE : National and Europear

2) **Cancer**; sequencing of exomes or genomes in several hundred patients with chronic B-cell lymphoproliferative syndrome, patients with primary tumours of unknown origin or unresolved cases of

suspected hereditary predisposition to cancer; and 3) **Pharmacogenomics**; genotyping and / or sequencing of biomarkers for drugs of high therapeutic impact.

- **standardized protocols** for sequencing and genome and exome analysis, quality management systems and laboratory information management systems in all centers.
- **Portfolio of processes** will be established diagnoses, with adequate response times, as well as a clinical report template for each group of patients pathology.
- The Group of **Clinical Experts** in rare diseases, cancer and pharmacogenomics for the analysis of the data.

Data Science Program seeks to develop a system for the collection, integration and **analysis of clinical and molecular data** aimed at improving the health of each individual patient, and which allows the secondary use of the existing information in the National Health System for the benefit of society with objectives of public health, health planning and research. Its objective is to optimize the management of the information generated, to interpret and apply it in the most effective way for the population and the health system; It will provide bioinformatics tools for the management of genomic data and Medical Informatics solutions and for the integration of clinical data, facilitating the interoperability of the clinical information systems of the different autonomous health systems.

Data Science Program objectives

- Federated data platform that includes medical, genomic and phenotype data.
- Implementation of a first version of a cloud computing infrastructure.
- Protocols, methods and integrated data analysis systems based on FAIRification mechanisms.
- Automatic and safe extraction of minimum standardized and pseudonymized information from Electronic Medical Record
- Extraction of quantitative information from **medical images**
- Integration of the results of the genomic and image analysis with info. from EHRs,
- Ecosystems of "demonstrators" on translational information interoperability functions, including a specific case on COVID-19.
- Concerted execution in different locations of the demonstrators by members of TransBioNet.
- Coordination of protocols, standards of analysis and procedures with **international initiatives**: ELIXIR, GA4GH, 1+ Million Genomes, ICGC, IRDiRC, EOSC and EHDS.
- **Report to the regulatory bodies**: CTI-SEIS, RedETS and AEMPS.
- Collaborate with existing **training activities** at national and international level both in medical informatics and bioinformatics.

Spanish National Bioinformatics Institute (INB) Spanish Node of ELIXIR (ELIXIR-ES)

Core Facilities at medical Institutes roups at Health Research stitutes certified by the ISCII ouns at Health Research **EUROPEAN GENOME-PHENOME** ARCHIVE **ELIXIR:** European infrastructure for biological information

TransBioNet

network of Bioinformaticians in Research Institutes of Spanish Hospitals (INB hosted)









Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020

Spanish National Bioinformatics Institute (INB) Spanish Node of ELIXIR (ELIXIR-ES)

TransBioNet

Network of Translational Bioinformatics Groups in Research Institutes of Spanish Hospitals. (INB hosted)





TransBioNet (mainly Institutes in Research Hospitals)

		Hospitals	Regional			
Acronym	TransBioNet memb		Health Systems			
GENYO	Centre for Genomics and Oncological Research			nearth systems		
IBIMA	Instituto de Investigación Biomédica de Málaga					
IBiS	Instituto de Biomedicina de Sevilla	H V del Rocio		SAS		
IMIBIC	Instituto Maimónides de Investigación Biomédica de Cordoba	Sovilla				
IACS	Instituto Aragonés de Ciencias de la Salud	Sevilla	_			
IIS Aragón	Instituto de Investigación Sanitaria Aragón		_			
ISPA	Instituto de Investigación Sanitaria del Principado de Asturias					
IBSAL	Instituto de Investigación Biomédica de Salamanca					
CRG	Centre for Genomic Regulation			CatSalut		
IDIBAPS	Institut d'Investigacions Biomèdiques August Pi y Sunyer	H Clinic		CatSalut		
IDIBELL	Instituto de Investigaciones Biomédicas de Bellvitge	Barcelona	BARCELONA Hospital Universitari	(AOUAS)		
IGTP	Germans Trias i Pujol Research Institute			(, (20, (3))		
IRB Barcelona	Institute for Research in Biomedicine Barcelona					
IRBLleida	Institut de Recerca Biomèdica de Lleida					
IRSJD	Institut de Recerca Sant Joan de Déu					
PSMAR-IMIM	Parque de Salud MAR (PSMAR) / Instituto Hospital del Mar de Investigad	iones Médicas (IMIM)				
VHIR	Vall d'Hebron Institut de Recerca					
CIPF	Centro de Investigación Príncipe Felipe					
INCLIVA	Institut d'Investigació Sanitària INCLIVA					
IDIS / FPGMX	Instituto de Investigación Sanitaria de Santiago de Compostela / Fundacio	Instituto de Investigación Sanitaria de Santiago de Compostela / Fundación Pública Galega de Medicina Xenómica				
IdISBa	Instituto de Investigación Sanitaria Islas Baleares	H 12Octubro				
i+12	Instituto de Investigación Hospital 12 de Octubre	II izottubie		SERMAS		
IdiPaz	Instituto de Investigación Hospital Universitario La Paz	Madrid	Hospital Universitario	JERIVIAS		
IDIPHIM	IIS Instituto de Investigación Sanitaria Puerta de Hierro		12 de Octubre			
IdISSC	Instituto de Investigación Sanitaria Hospital Clínico San Carlos					
IIS-FJD	IIS-Fundación Jiménez Díaz					
liSGM	Gregorio Marañón Health Research Institute					
IISPrincesa	Instituto de Investigación Sanitaria Hospital de la Princesa					
IMDEA	Instituto Madrileño de Estudios Avanzados					
IRYCIS	Instituto Ramón y Cajal de Investigación Sanitaria			7		
ISCIII	Instituto de Salud Carlos III		BSC			
IMIB	Instituto Murciano de Investigación Biosanitaria	Research contro	Research contracts as part of the			
IDISNA	Instituto de Investigación Sanitaria de Navarra					
IISBiodonostia	Instituto de Investigación Sanitaria BioDonostia		National NLP P	lan		
ibs.GRANADA	Instituto de Investigación Biosanitaria de Granada			Parch Impediate Set		
IDIVAL	Instituto de Investigación Sanitaria Marqués de Valdecillas			and Transford Trappe . Settle		
CNIC	Centro Nacional de Investigaciones Cardiovasculares			Plan de Impulso de la		
IIB Sant Pau	Institut d'Investigació Biomèdica de Sant Pau			Tecnologías del		

https://inb-elixir.es/transbionet

Plan TL



Global Alliance

for Genomics & Health





ELIXIR **Core Data Resources are of fundamental importance** to the wider life-science community and the long-term preservation of biological data.



Controlled access for human genomic data





The selected EGA a **Driver Projects** of the Global Alliance for Genomics and Health Definition of **global human data sharing standards** together with national projects

ELIXIR - GA4GH Beacon project

- ELIXIR Beacon as driver project lead by the EGA Team
- GA4GH Beacons are a discovery service
- ELIXIR pilot project with partners from the Netherlands, Sweden, Finland and Spain.
- Complies to the GA4GH Beacon project standard and security working group policies.

EGA Beaco	on						EMBL-EBI	CR			
By use of this Beacon Ser authorized by law or by a	ervice, l'agree a written prior	n to forego any r permission fro	attempt to re-iden on the respective	offy individuals DAC. (more de	represented in Beacon : tails)	Service Replies, except where expre	saly	1 Jordi Rambia -			
Query available datasets	an ann an a' s				100	Note: The EGA archives a lar datasets. To access Common description page under "Who	ge number of datasets, some of which are swallable. If you have an account on this website, y distasets can be done by contacting the relevant Data Access Committee (DAC), whose datais are controls access to this dataset' (cick on the Dataset D to go to the Dataset page). Once you have acc	ou can access to isplayed on the Dataset ass to a dataset, you will t			
Select a d	enome:	all (204632	363 variants)		>	EGA ID	 Short title 	~ Access type			
Chrome	iosome	18			~	EGAD00001000433	This sample set comprises cases of schizophrenia with additional cognitive measurements, college	te CONTROLLED		lianaa	
Po	osition	2656142				EGAD00001000814 EGAD00001000443	This sample set of UK origin consists of clinically identified subjects with Autism Spectrum Daord The sample selection consists of subjects with schicophrenia (SZ), autism, or other psycholes a	CONTROLLED	🤹 Global Al	llance	nlivir
	Allele:	T				EGAD00001000740 EGAD00001000613	Low-coverage whole genome sequencing; variant calling, genotype calling and phasing The MGAS (Molecular Genetics of Autism Study) samples are from a claical sample seen by spe-	PUBLIC ISE CONTROLLED	for Genom	nics & Health	EIAII
		This Beaco keep in mini	s is based on the I I that indel queries	GA4GH Beaco s are not suppo	n API 0.3. Please, inted yet.	EGAD00001000430 EGAD00001000434	Two groups of samples with diagnosis of schizophrenia or schizoaffective disorder in the UK ca The BioNED (Biomarkers for Childhood onset neuropsychiatric disorders) study has been carrying	CONTROLLED			F ²
		Find				EGAD00001000437 EGAD00001000439 EGAD00001000442	The Tampere Autiam sample set consists of samples from Finnish subjects with ASD (autiam spe The entire sample collection consists of 2756 individuals from 456 families of whom 931 are deap Samples from three sources: the Genetics and Psychosis (GAP) set consists of samples from su	df CONTROLLED OS CONTROLLED	Collaborate. Inr	novate. Accelerate.	
						EGAD000000028	Procardia study for coronary artery disease. GWAS study 3352 cases - 3145 control	Percent Roman			
Beacon search history					Clear results	EGA000010000000	Summary statistics from travengen RBC GWAS (Anema) Apprendix results from a case-control study on simile and ischemic stroke 19002 samples	Contractor (Contractor)		_	
Dateset Ref	r	Chr	Pos	Var	Exists	EGA20000000115	WNT-signaling and Dupuytren's Disease. GWAS analysis. 856 cases - 2836 control.	CONTROLLED		BV	I Ramhla FGA
el CP	201637	18	2658142	т.	Palao	EGAD00001000435	These samples are a subset of a nationwide collection of Finnish autism spectrum disorder (ASD	B CONTROLLED		Dy .	
-1 00		10	1000114	*	(1000)	EGAD00001000615	These Finnish schizophrenia samples have been collected from a population cohort using national	re			
al GR	ech37	16	2000141		Talse	EGAD00001000440	These affected schizophrenia families have been diagnosed using the SADS-L clinical instrument	AL. CONTROLLED			
al GR	RCh37	18	2656141	A	fatse	EGAD00001000436	This is an Irlah sample set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50% with comorbid intellectual on the set of individuals with ASD (approximately 50\% with comorbid intellectual on the set of individuals with ASD (approximately 50\% with comorbid intellectual on t	BB. CONTROLLED			
al GR	RCh37	18	2656145	G	false	EGA200001000438	The MOSAC data set consess or subjects with scriptophrenia recruited from psychiatric m-patient and The MOSAC data set represents an international collection of families containing children ascerta	DUL. CONTROLLED			
al GR	RCh37	18	2656145	A	false	< Total tens: 22		and the second			





Building genomics genomDE : National and European initiatives 30TH NOVEMBER 2020

European COVID-19 Portal (EMBL-EBI and ELIXIR) and the Spanish branch

()



genomDE : National and European initiatives 30TH NOVEMBER 2020

Building genomics

genomDE : National and European initiatives **30TH NOVEMBER 2020**

(i)

Unmute

You are viewing cochrane's screen

View Options ~

Instituto de Salud Carlos III

Sł

JB

Spanish National

SPAIN

👑 Speaker View













COVID-19 Data Layers

~

Stop Video



51

Participants



Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020



Spanish National Registry for COVID-19

The **Spanish National Repository for COVID-19 related data** is a data platform designed to handle data of diverse type, e.g. viral and **host sequencing, imaging, clinical, population-based results from Spanish COVID-19 related projects**, honouring all ethical, legal and societal aspects

The platform can host securely submitted data as well as to facilitate the automated submission to existing repositories, e.g. ENA, EGA, retaining associated metadata for a later retrieval. Depending on the nature of the data, e.g. electronic health records (EHRs), it is possible that only a specific/minimal set of metadata is maintained at the repository, while including information about where the data is available and the required mechanisms to gain access.



30TH NOVEMBER 2020



Expected data flow across the Spanish system





The COVID-19 CORE Case Report Form (CRF)





CLÍNIC BARCELONA

Hospital Universitari

Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020

Correspondences



Predicting Disease Outcome with Recurrent Neural Networks and Attention mechanisms

K M hm hospitales

TOWARDS PREDICTING THE EVOLUTION OF COVID-19 MORTALITY RISK: A RECURRENT NEURAL NETWORK APPROACH CP Carrino, A Gonzalez-Agirre, J Armengol-Estapé, A Gutiérrez-Fandiño, D Pérez Fernández, M Villegas. A Valencia





Digital Twin for Future Medicine

Anna is going to

When will it be possible? For what medical problems?



Building genomics

genomDE : National and European initiatives 30TH NOVEMBER 2020

Thank You



Supported by the European Union's Structural Reform Support Programme (SRSP) and implemented in cooperation with the European Commission's Directorate-General for Structural Reform Support (DG REFORM)





Federal Ministry of Health





