

INB/ELIXIR-ES: El papel de las RIs en medicina personalizada

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Jornada Informativa RIs en Ciencias Biomédicas: El papel de las

RIs en medicina personalizada.

ELIXIR connects national bioinformatics centres and EMBL-EBI into a sustainable European infrastructure for biological research data



Agriculture

Bioindustries

Environment

ELIXIR underpins life science research – across academia and industry

ELIXIR - Data for life

ELIXIR is an intergovernmental organisation that brings together life science resources such as

- → databases
- → software tools
- training materials
- → data standards
- compute resources

The goal of ELIXIR is to coordinate life science resources from across Europe so they form a single infrastructure.

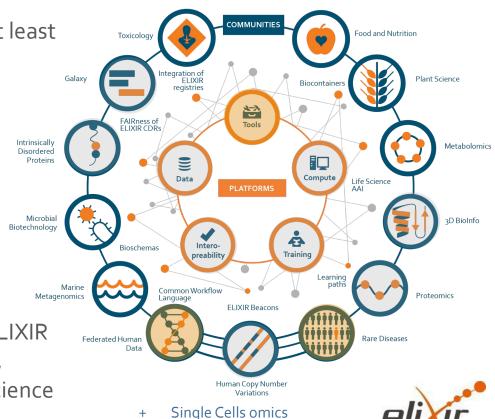


ELIXIR - Platforms and Communities

ELIXIR coordinates activities through at least one of the five 'areas of activity' called Platforms:

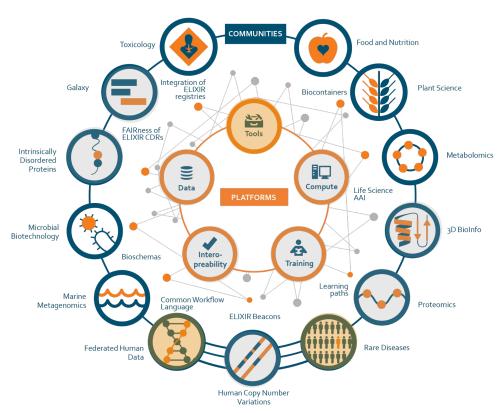
- Compute
- → Data
- Interoperability
- → Tools
- Training

These Platforms are driven by eleven ELIXIR Communities which develop standards, services, and training within their life science domains.



Systems Biology

ELIXIR - Platforms, Communities and Focus Groups



- + Single Cells omics
- + Systems Biology

Emerging Communities

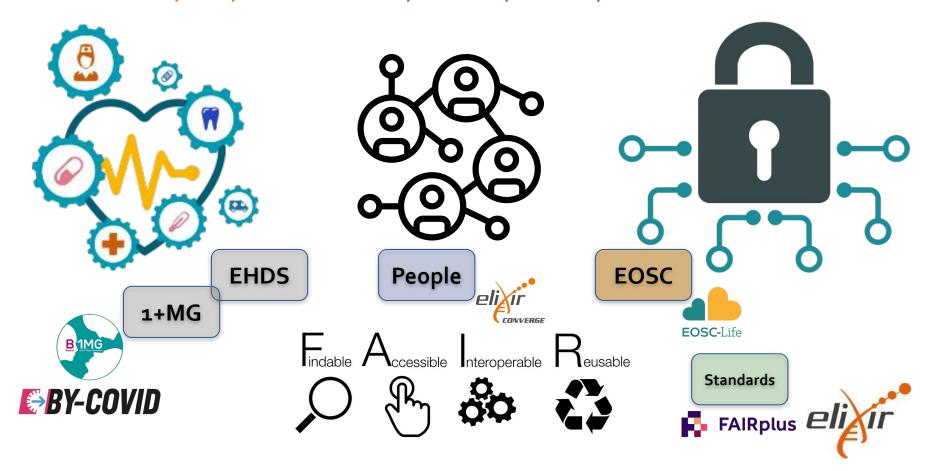
- → Biodiversity
- → Microbiome (emerging from the Marine Metagenomics Community)

Current Focus Groups

- Biocuration
- Biodiversity
- Cancer Data
- EC Research Funding
- EOSC
- FAIR Training
- Health Data
- Impact
- Innovation and Industry
- Machine Learning (AI)
- Registries
- Research Data Alliance (RDA) Activities
- Systems Biology



An ELIXIR perspective on key concepts for personalized medicine



ELIXIR Communities and Focus Groups

Human data Communities:

- Federated Human Data
- Rare Diseases
- Human Copy Number Variation (hCNV)

Biomedical Focus Groups:

- Health Data
- Cancer Data



Estructura de INB/ELIXIR-ES (Spanish Node of ELIXIR)

https://inb-elixir.es



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INB Mission

INB Nodes

INB Publications

The National Spanish Bioinformatics Institute serves the wider research community by pursuing the following goals:

- To provide world-class core bioinformatics resources do to the national and international life science research community in key fields such as functional genomics, transcriptomics with RNA-Seq, genotyping, genomic medicine, and molecular dynamics simulations.
- To provide a platform of integrated core bioinformatics services to the research community. The INB groups have developed over the years an extensive platform of web services that integrates a wide range of bioinformatics methods. This platform is evolving to take advantage of cloud computing to scale up to large data processing workflows that are required for example in the analysis of data produced with next generation sequencing platforms.
- To facilitate coordinated participation of bioinformatics groups in large-scale national and international projects that require and benefit from closely collaborating teams that know each other and are used to working together.

Call for Expressions of Interest

for being part of the

INB/ELIXIR-ES resources portfolio









Personalized Medicine in Spain



IMPaCT (Infrastructure for Personalized Medicine associated to Science and Technology) aims to set the foundations of the national genome medicine in Spain.

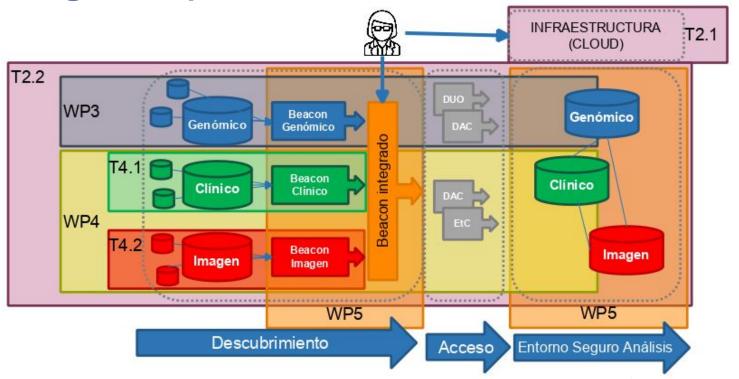
- Divided into three complementary programs: Preventive Medicine, Genomics Medicine and **Data Science**.
- BSC coordinates the Data Science programme (46 partners + 20 other organizations), which aims to focus in the interception of clinical information, genomics data and biomedical imaging.
- The Data Science program will strongly rely in existing standards and mechanisms to favour **interoperability**, e.g. **OMOP**, **FHIR**, **openEHR**.
- Connects with other efforts like the **National Plan for Natural Processing Language** (among others domains from electronic health records).





Working concept











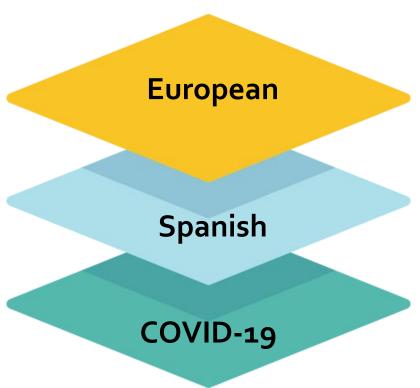








Perspective



- Personalized medicine strongly relies on diverse data types which are diverse and broad.
- Strong focus on interoperability and existing standards to lower the barrier for adoption and promote sustainability.
- Existing solutions for federated access to genomics, e.g. European Genome-phenome Archive (EGA), beacon, can be potentially extended to other domains.
- Strong connection with the 1+ Million Genomes Initiative (1+MG) by 23 members states.









https://inb-elixir.es http://elixir-europe.org











