



INB/ELIXIR-ES as part of the Research Infrastructures ecosystem

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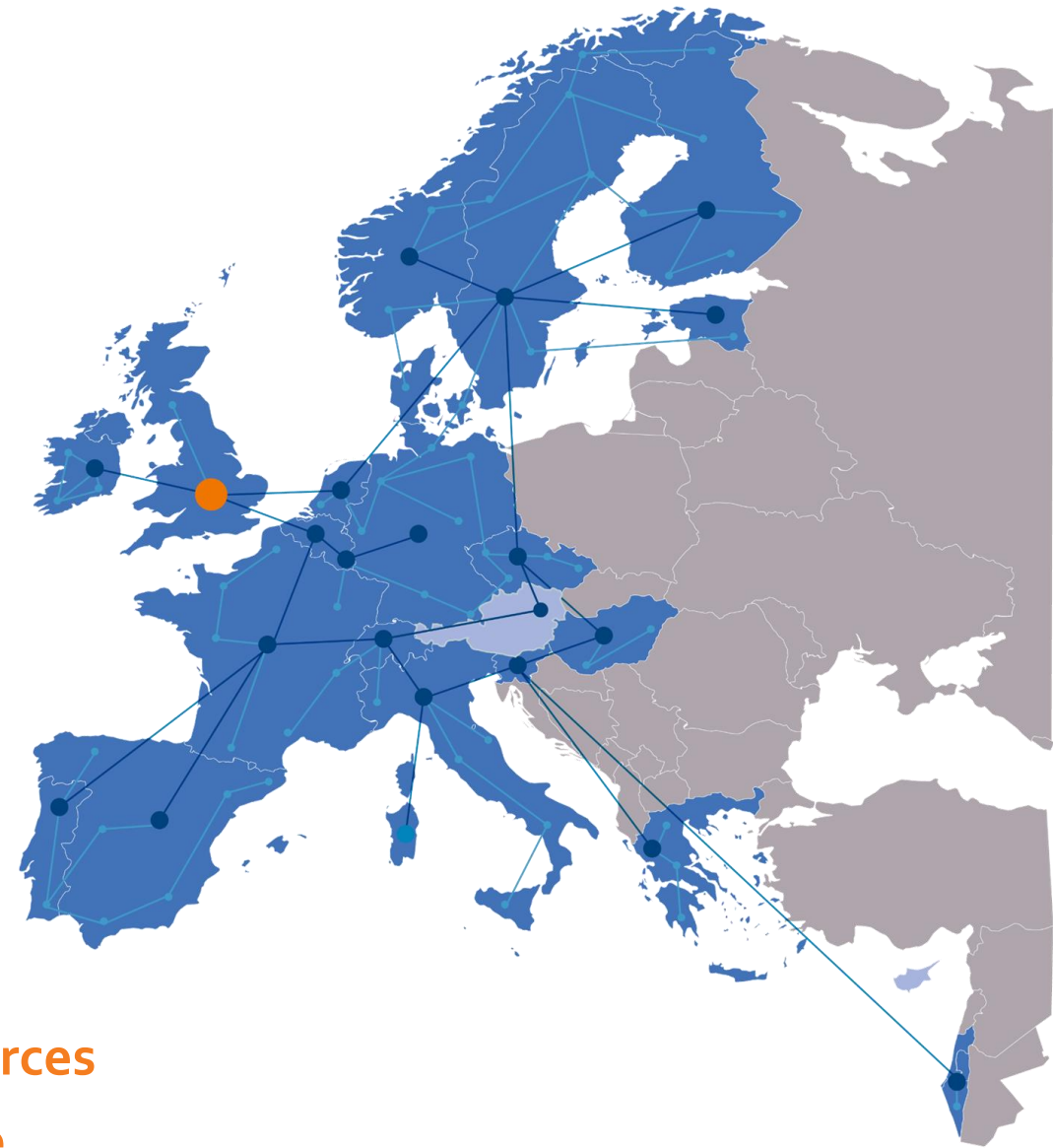


ELIXIR Europe

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

- databases
- software tools
- training materials
- interoperability resources
- compute resources
- data management support

The goal of ELIXIR is to **coordinate bioinformatics 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 13 ELIXIR **Communities** which develop standards, services, and training within their life science domains.

This structure is completed by different **Focus Groups** including the **Cancer Research** one.



ELIXIR - How national nodes contribute to the ecosystem

- ELIXIR provides publicly available and free of charge services and resources through its National nodes (and the associated organizations to them), which are used by the entire Life Sciences community.
- Following this model, it is possible to utilize some of those services as part of the national activities, e.g. in IMPaCT-Data, and also bring in software developed and maintained by other ELIXIR nodes.
- ELIXIR nodes contribute to the overarching ELIXIR objectives with their service delivery plans, with in-kind contributions leading different structures, e.g. platforms, communities, and through commission services to connect existing services, coordinate actions across nodes and explore the use of new technologies.
- The Spanish National Bioinformatics Institute (INB) is the ELIXIR node in Spain, which was ratified in 2017.



INB/ELIXIR-ES: Activities in 2022

- Establishment of the **Spanish Federated EGA node** (FEGA-es).
- **Leadership** in the Beacon and its associated network, and the release and adoption of the Beacon V2, including text-mining extraction and partial manual curation of results by DisGeNET, according to the open-source discovery protocol GA4GH Beacon API.
- Maintained the **contribution to IMPaCT, including coordinating IMPaCT-Data**, releasing two demos on how to build a federated ecosystem enabling distributed analysis via Galaxy and using GA4GH Passports and Visas technologies.
- **European Genomic Data Infrastructure (GDI)**: CNAG-CRG Spanish representative with ISCIII, BSC leads Pillar III.
- Maintained the **TransBioNet collaborations** with hCNV community and CIBERER, providing connections with the Genomics pillar (IMPaCT-Genómica).
- Contribution to **establishing the Gaia-X Spanish Hub** and co-leadership of the Technology area within the **Health working group**.
- **Bioinfo4Women initiative** continued the gender bias projects in the BioHackathon, deepened collaboration with de.NBI WiDS initiative, and launched the **ELEAD leadership pilot** within ELIXIR.

INB/ELIXIR-ES: Activities in 2022

→ Organised several events such as:

- European Conference on Computational Biology - ECCB2022 including an outreach event to connect with the Latin American bioinformatics communities
- The collocated ELIXIR Innovation and SME Forum: Data-driven innovation in healthcare diagnostics
- Federated EGA Celebration Symposium and presentation of the inaugural FEGA nodes within the 10th GA4GH plenary in Barcelona.

The Spanish National Bioinformatics Institute (INB) is the ELIXIR Node in Spain (ELIXIR-ES). The INB was founded in 2003 as a distributed network of nodes with a central coordination hub. At present, INB counts with 26 research groups distributed across 14 institutions in Spain. Currently, the two main overarching objectives are: **deepen its alignment with ELIXIR and its communities with a strong focus on human data related activities; and increasing its translational impact within the Spanish National Health System** regarding the secondary use of health-related data for research. At the national level, the INB/ELIXIR-ES leads the Translational Bioinformatics Network (TransBioNet) and coordinates IMPaCT-Data, the Data Science pillar of the Spanish National Infrastructure for Precision Medicine.

Renewed Service Delivery Plan

40 services	24 groups	12 institutions	
19 tools	9 web-services	4 platforms	8 databases

Alignment and involvement in ELIXIR

- Co-leadership of the ELIXIR Tools and Training platforms; the Federated Human Data, Rare Diseases and Biodiversity Communities; and the Cancer Data Focus Group.
- Secured controlled access to human data through the European Genome-phenome Archive (EGA), jointly managed by EMBL-EBI and CRG with the technological support of BSC. EGA is part of the ELIXIR Core Data Resources (CDR) and the ELIXIR Recommended Deposition Databases (EDD).
- Establishment of the Spanish Federated EGA node (FEGA-ES).
- Development of the ELIXIR platform for supporting technical monitoring and community-driven benchmarking efforts OpenEBench.
- Designation of ELIXIR-ES resources 3D-Bionotes and FAIRtracks (in collaboration with EMBL-EBI and ELIXIR-NO) as part of the ELIXIR Recommended Interoperability Resources (RIR).
- Leadership of the GA4GH Beacon and ELIXIR Beacon Network, including recent extensions of the Beacon V2 standard, e.g. clinical, medical imaging.

Moreover, groups from INB/ELIXIR-ES have intense activity across the ELIXIR structure:

- 5 Platforms: Tools, Data, Compute, Training, Interoperability.
- 13 Communities: Federated Human Data, Human Copy Number Variation, Rare Diseases, Galaxy, Proteomics, 3D-BioInfo, Intrinsically Disordered Proteins, Toxicology, Plants Sciences, Single Cell Omics, Systems Biology and Biodiversity, and the emerging Research Data Management community.
- 6 Focus Groups: Machine Learning, Cancer Data, Health Data, EOSC, Impact, Innovation and Industry.
- 11 Implementation studies, including strategic ones like Federated EGA framework, ELIXIR Beacon, ELIXIR Rare Disease services; and Making container services, sustainable and widely adopted.

INB/ELIXIR-ES are also active in relevant activities for ELIXIR and the whole community:

- Promotion of FAIR data principles and scientific software development best practices, and training.
- Organised the ECCB2022, including an ELIXIR outreach event with Latin American bioinformatics communities, and the co-located ELIXIR Innovation and SME Forum on Data-driven innovation in Healthcare Diagnostics.
- Organised the Federated EGA Celebration Symposium and presentation of the inaugural FEGA nodes in the framework of the 10th GA4GH plenary in Barcelona.
- Gender equity in Bioinformatics: ELIXIR has launched the ELIXIR Leadership And Diversity mentoring programme (LEAD Pilot), which builds from previous experiences of the BioInfo4Women programme.

BSC leads the Spanish participation in ELIXIR-CONVERGE, with other members of the node as beneficiaries. In this context, the UPF is responsible for the FAIR encoding and access to Toxicology data use-case while the IRB Barcelona leads the FAIR organisation of biomolecular simulation information one, which triggered particular efforts on organising biomolecular simulation data through dedicated infrastructures. The CRG co-leads WP7 related to the Federated EGA for translational access of COVID-19 host data, which served to the development and initial testing of the Federated EGA Maturity Model. This maturity model is nowadays the reference for the onboarding of new nodes into the federation. It has also served as reference for the establishment of the maturity model for the Federated One Health Surveillance Platforms. Beyond ELIXIR-CONVERGE, Spanish institutions have an active role in other ELIXIR-led projects like EOSC-Life, B1MG, GDI, and BY-COVID.

Translational Bioinformatics

- Creation and coordination of TransBioNet, Translational Bioinformatics Network, an open experts' forum bringing together 38 Bioinformatics groups at Health Research Institutions across Spain.
- Share best-practices and experiences, early access to standards, protocols and best practices by ELIXIR and GA4GH, benefit from access to the computational infrastructures provided by the INB and ELIXIR, and training activities, altogether have a direct impact on researchers working closely to clinicians.

Precision Medicine

- IMPaCT-Data is the Data Science pillar of the Spanish National Infrastructure for Precision Medicine associated with Science and Technology (IMPaCT), created by the Institute of Health Carlos III in 2021.
- INB/ELIXIR-ES nodes and TransBioNet members are integrated into IMPaCT-Data, structured around 60 research groups of 47 institutions across Spain, and coordinated by BSC.
- IMPaCT-Data aims to develop the first iteration of a Biomedical Cloud to establish the mechanisms for working with genomics, clinical and medical imaging data for research purposes in a distributed manner.
- The project has delivered 1) two demonstrators on how to build a federated ecosystem enabling distributed analysis via Galaxy and using GA4GH Passports and Visas technologies; 2) recommendations and guidelines regarding the standardization of biomedical data to facilitate its interoperability, access, and governance.
- The reference implementation of the Biomedical Cloud under development will include several components as Beacon and Beacon network, Local EGA, Galaxy, openVIRE, AAI, CVMFS, OMDP, OHDSI, cBioPortal, and other central services.
- IMPaCT-Data aligns with many European projects and initiatives and implements standards pushed forward by community-driven initiatives, e.g. ELIXIR, GA4GH, 1+MG/B1MG, EUCAIM.

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Logos: INB, BSC, ELIXIR SPAIN, FEDER, Institute of Health Carlos III, European Union, European Regional Development Fund (ERDF), "a way to make Europe".

Alloza E, INB/ELIXIR-ES members, Valencia A and Capella-Gutierrez S. The Spanish National Bioinformatics Institute (INB/ELIXIR-ES). Renovating its portfolio in its 20th anniversary. *F1000Research* 2023, 12(ELIXIR):579

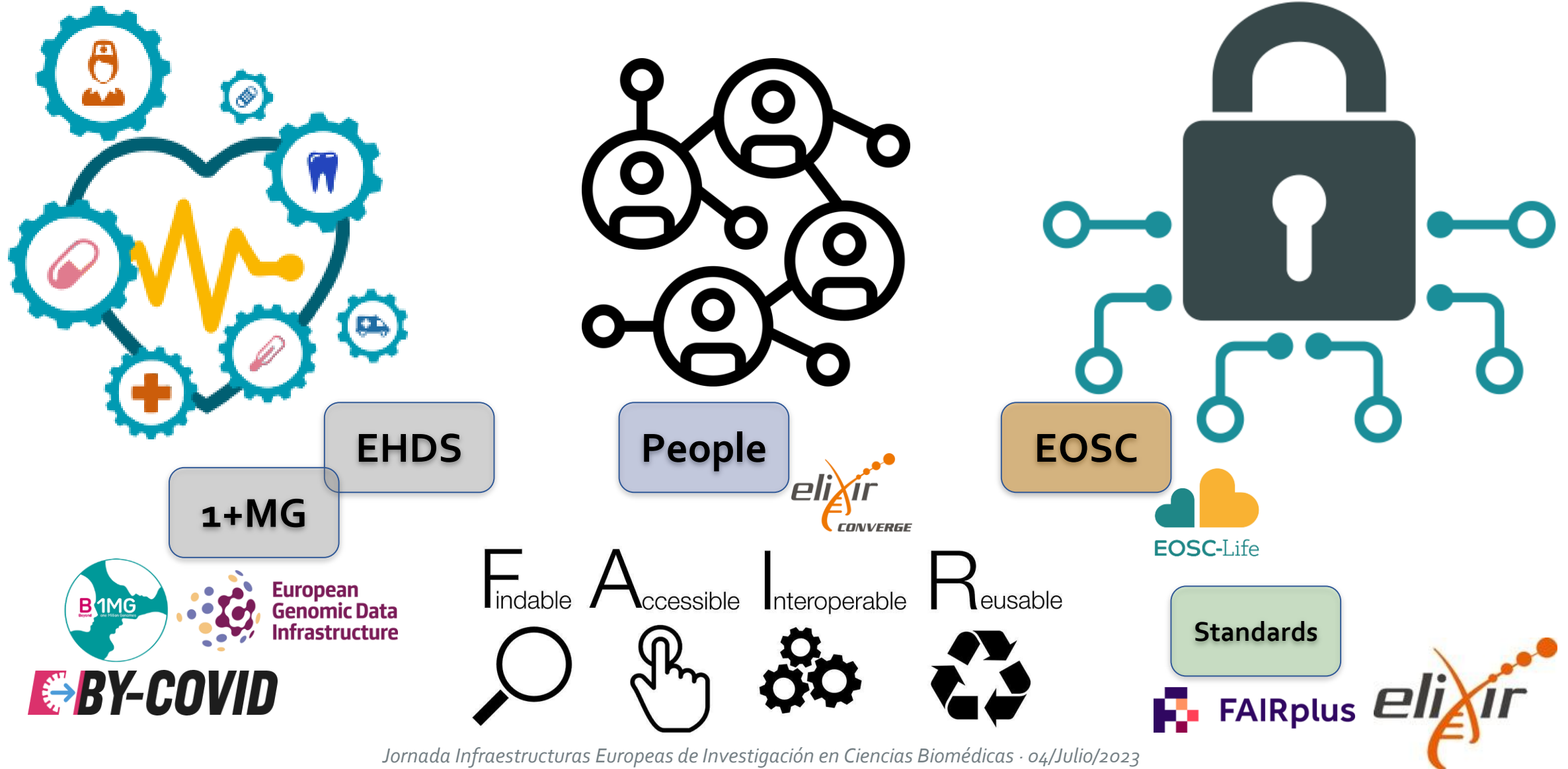
(poster) (doi: [10.7490/f1000research.1119435.1](https://doi.org/10.7490/f1000research.1119435.1))



INB/ELIXIR-ES: A new service delivery plan

3Did 3D interacting domains <i>IRB</i>	Beyondcell <i>CNIO</i>	DREIMT <i>CNIO</i>	FAIR Cookbook <i>SAS, BSC</i>	Hipathia suite <i>FPS</i>	dSysMap Mapping human disease- mutations at systemic level <i>IRB</i>	Nextflow <i>CRG</i>	rbbt Ruby bioinformatics toolkit <i>BSC</i>
3D BIONOTES-WS and COVID19 Structural Hub  <i>CNB (CSIC)</i>	BioBB BioExcel Building Blocks <i>IRB, BSC</i>	EGA Beacon <i>CRG</i>	FAIRTracks Validation Service  <i>BSC</i>	FlexPortal INB Integrated platform for macromolecular flexibility <i>IRB, BSC</i>	Master Of Pores <i>CRG</i>	PanDrugs <i>CNIO</i>	SQANTI <i>IzSysBio (CSIC)</i>
PMut Annotations of Pathological Mutations on Proteins <i>BSC, IRB</i>	BSC Text Mining Unit tools <i>BSC</i>	eggNOG <i>CBGP (CSIC)</i>	footprintDB <i>EEAD (CSIC)</i>	Interactome3D <i>IRB</i>	MBROLE Metabolites Biological Role <i>CNB (CSIC)</i>	PhylomeDB <i>BSC, IRB</i>	Sqtlseeker2-nf <i>CRG</i>
APID Interactomes <i>IBMCC (CSIC)</i>	CANTEMIST <i>BSC</i>	ETE Toolkit <i>CBGP (CSIC)</i>	GCAT Genomes for Life <i>IGTP</i>	IntOGen <i>IRB</i>	MetaPhors <i>BSC, IRB</i>	Redundans <i>BSC, IRB</i>	T-Coffee <i>CRG</i>
APPRIS <i>CNIO</i>	CSVS Collaborative Spanish Variant Server <i>FPS</i>	EGA European Genome-phenome Archive  <i>CRG</i>	GET_HOMOLOGU ES <i>EEAD (CSIC)</i>	manta/mvgwas-nf <i>CRG</i>	OpenEBench <i>BSC</i>	RSAT Plants <i>EEAD (CSIC)</i>	WfExS-backend Workflow Execution Service  <i>BSC</i>

An **ELIXIR perspective** on key concepts for personalized medicine





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

