INB/ELIXIR-ES
The Spanish National Bioinformatics Institute

E. Alloza[1,2], J. Rambla[1,3], S. Beltrán[1,4,5,6], J.R. Macías[1,8], J.M Fernández[1,2], J. Piñero[1,5,7], INB/ELIXIR-ES members, J.M. Carazo[1,8], L.I. Furlong[1,5,7], F. Sanz[1,5,7], I. Gut[1,4,5], J.L. Gelpí[1,2,6], A. Navarro[1,5,9], A. Valencia[1,2,9] & S. Capella-Gutierrez[1,2]

[1] Spanish National Bioinformatics Institute (INB/ELIXIR-ES) [2] Barcelona Supercomputer Center (BSC) [3] Centre for Genomic Regulation (CRG) [4] Center for Genomic Regulation (CRG), CNAG-CRG, Barcelona Institute of Science and Technology (BIST) [5] Universitat Pompeu Fabra (UPF) [6] University of Barcelona (UB) [7] Research Group on Integrative Biomedical Informatics (GRIB), Institut Hospital del Mar d'Investigacions Mèdiques (IMIM) [8] Spanish National Centre of Biotechnology (CNB-CSIC)/Instruct Image Processing Centre [9] Catalan Institution for Research and Advanced Studies (ICREA)

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, with 17 research groups distributed across 10 institutions in Spain at the moment. 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 medicine impact within the Spanish National Health System** (SNS). At the national level, it leads the Translational Bioinformatics Network (TransBioNet) and coordinates IMPaCT-Data, the Data Science pillar of the Spanish National Infrastructure for Precision Medicine.

Core Resources

Strong focus on Biomedical activities, including the 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** and the **ELIXIR Recommended Deposition Databases**.

Alignment and involvement in ELIXIR

- Co-leadership of the **ELIXIR Tools platform**.
- Co-leadership of the **ELIXIR Federated Human Data**, **Rare-diseases** and **Biodiversity communities**.
- Co-leadership of the **Cancer Data focus group**.
- Resources as key components of different European efforts and projects, e.g. **RD-Connect platform Genome-Phenome Analysis Platform** (GPAP) is part of the European Joint Programme on Rare Diseases (EJP-RD) and the Beyond 1 Million Genome (B1MG) project.
- Development of the benchmarking platform **OpenEBench**.
- Designation of ELIXIR-ES resources 3D-Bionotes, DisGeNET and FAIRtracks (in collaboration with EMBL-EBI and ELIXIR-NO) as part of the ELIXIR **Recommended Interoperability Resources** (RIR).
- Development of the **COVID-19 Data Portal Spain**.

Moreover, groups from INB/ELIXIR-ES have intense activity across different **platforms, communities, focus groups** and **implementation studies.**
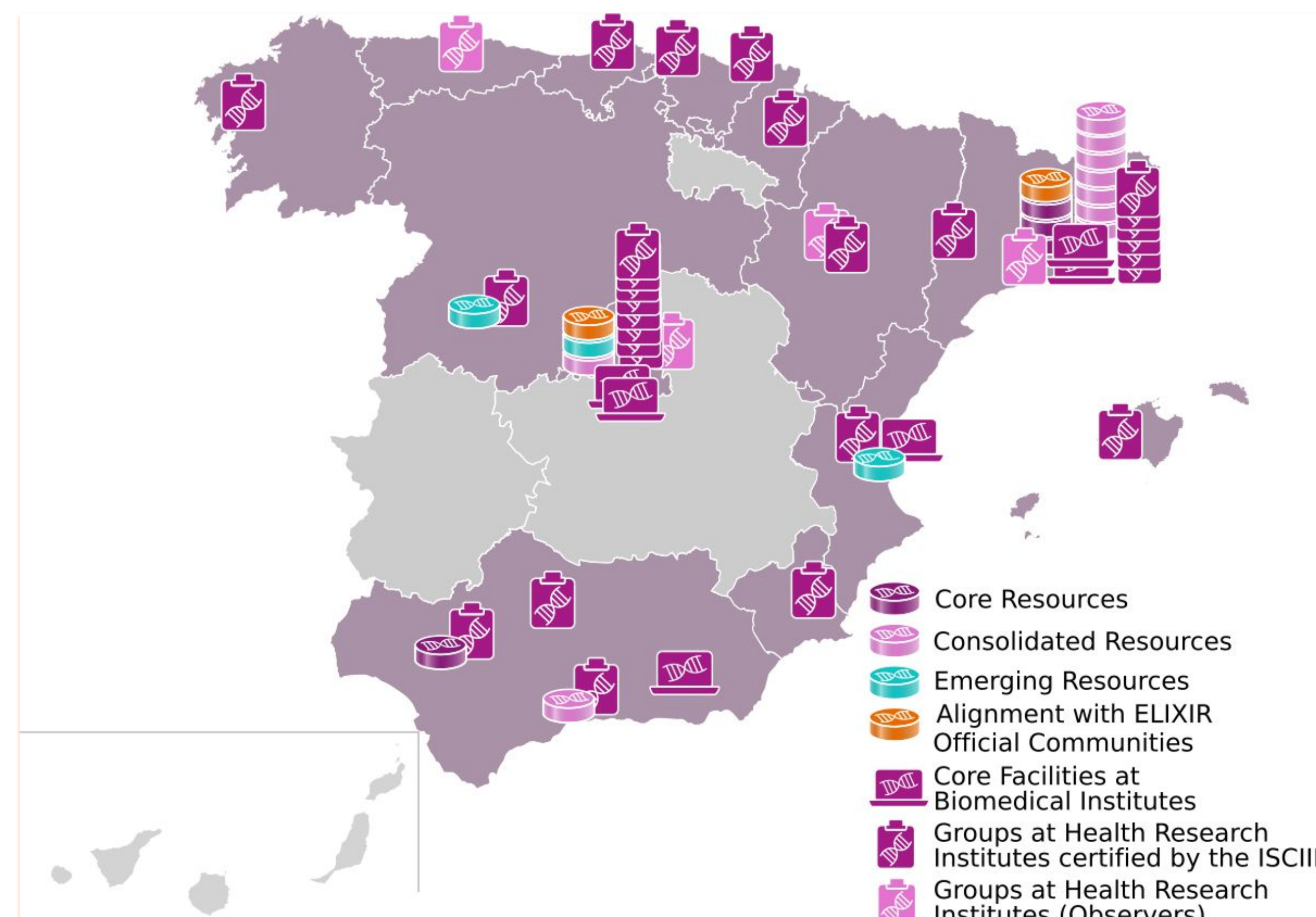- **5 Platforms**: Tools, Data, Compute, Training, Interoperability.
- **11 Communities**: Federated Human Data, Human Copy Number Variation, Rare Diseases, Galaxy, Proteomics, 3D-BioInfo, Intrinsically Disordered Proteins, Toxicology, including the emerging communities Single Cell Omics, Systems Biology and Biodiversity.
- **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.
- Organization of **training activities** e.g. EMBO course for bioinformatics core facility managers.
- Local organisers of the **BioHackathon Europe 2021** and **Industry and SME Forum 2022**.
- Involved in defining **impact assessment** actions at the Node level.
- **Gender equity** in bioinformatics commitment within the **Bioinfo4Women programme**.

BSC leads the Spanish participation in **ELIXIR-CONVERGE** across the whole project, with other members of the node as Beneficiary. BSC co-leads WP5 on Demonstrators Projects with the relevant participation of CRG (Federated access of sensitive human data), UPF (FAIR encoding and access to Toxicology data) and IRB Barcelona (FAIR organisation of biomolecular simulation information). CRG co-leads WP7 related to the Federated EGA for transnational access of COVID-19 host data. Moreover, ISCIII, FPS and FISABIO-CSIC incorporate to mobilise viral genomes data from national sequencing efforts. Spanish nodes participate in other ELIXIR-led projects like **B1MG**, **BY-COVID**, **IMI FAIRplus**.

INB/ELIXIR-ES groups actively participate in relevant projects for advancing on the use and reuse of biomedical data for research purposes, including EOSC Life, EJP-RD, euCanSHare, CINECA, **and led projects** like PerMedCoE, HealthyCloud, EOSC4Cancer, EUCANCan, EASI-Genomics. INB/ELIXIR-ES groups are deeply involved in different European and international efforts and initiatives, e.g. leading 1+MG initiative working groups and mirror groups, IRDiRC and GA4GH. The latter includes leading efforts in the Beacon and its associated network, and the Beacon specification v2 includes the use of text-mining extraction and partial manual curation of results by DisGeNET.

INB Nodes and TransBioNet Members as of November 2020, including the alignment with ELIXIR. As of June 2022, there is an ongoing process to renovate the node composition.

Legend:
- Core Resources
- Consolidated Resources
- Emerging Resources
- Alignment with ELIXIR Official Communities
- Core Facilities at Biomedical Institutes
- Groups at Health Research Institutes certified by the ISCIII
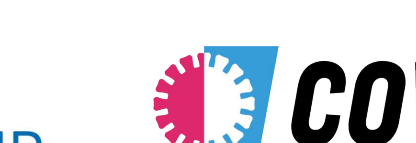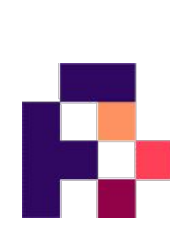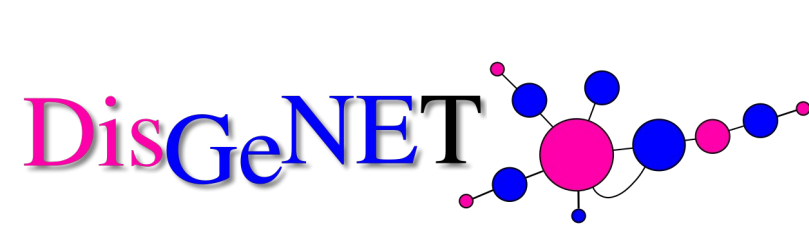- Groups at Health Research Institutes (Observers)

Precision Medicine ᴵᴹᴾᵃᶜᵀ|Data

- **IMPaCT-Data** is the Data Science pillar of the Spanish National Infrastructure for Precision Medicine associated with Science and Technology, IMPaCT, launched by 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. The combined expertise of all its members are the cornerstones of this endeavour, as it would allow to evaluate the maturity of any technological development before entering into daily healthcare routines.
- 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**.
- 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.

Translational Bioinformatics TRANSBIONET

- Creation and coordination of **TransBioNet**, Translational Bioinformatics Network, an open experts forum formed by 38 members across Spain to bring together bioinformatics groups at Health Research Institutions of the SNS.
- Share best-practices and experiences, have 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 can have a **direct impact on researchers working closely to clinicians**.