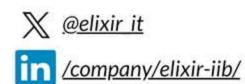


ELIXIR Infrastructure: Advancing Management, Sharing, and Analysis of Life Sciences Data across Europe



Slides: courtesy of Francesca De Leo (deputy Head of Node of Elixir-IT)

Pier Luigi Martelli Bologna Biocomputing Group



www.elixir-italy.org/

ELIXIR – who we are

ELIXIR is an intergovernmental organisation, formally established in **2016** as a **Landmark European Research Infrastructure (ESFRI Roadmap)**. The Roadmap has been updated in **2021** and consists of **22 ESFRI projects** and **41 ESFRI Landmarks**.

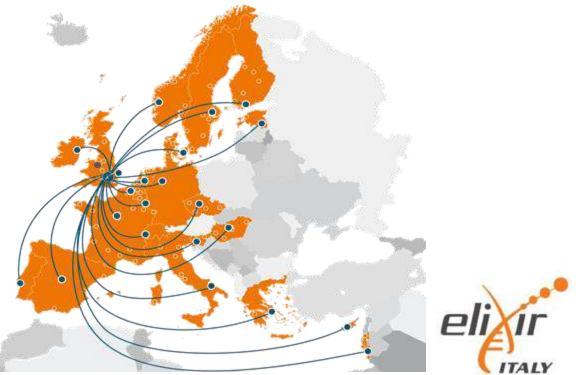
ELIXIR, brings together "bioinformatic resources" for life sciences from across Europe, including databases, software tools, training materials, best practices, cloud storage and supercomputers.

23 Nodes

ELIXIR Nodes form national networks of excellence, containing a total of 245 institutes/universities

1 Hub

The Hub, located at EMBL-EBI, Cambridge, UK, provides the secretariat and coordinating activities for ELIXIR



ELIXIR-IT

The Italian Node of ELIXIR (ELIXIR-IT) is formally established as as a Joint Research Unit (JRU) through a specific agreement among the coordinating organization (National Research Council, CNR) and the other 30 members. Apart from the CNR, the largest public research institution in Italy, the other 30 members are composed of 16 Universities, and 14 scientific and technological institutes that, in most cases are leaders in the country in their respective fields of expertise (e.g. CINECA and INFN in supercomputing, ISS in public health, GARR in IT infrastructure for research, TIGEM for rare diseases).

The **leading Institution** (**CNR**) has been designed by the Ministry of University and Research.

The **organisational layout** of the node has three layers: 1) policy and oversight; 2) coordination; and 3) operational. The General Assembly (GA), advised by an international SAB, is the main body of the Node, leaded by the HoN (designated by the President of CNR) and including delegates designated by the legal representatives of participating institutions. The coordination and operational layer, including the staff involved in all ELIXIR-IT activities includes both in-kind and full-time staff (about 20 FTE in 2023).



ELIXIR-IT – EPTRI-IT agreement

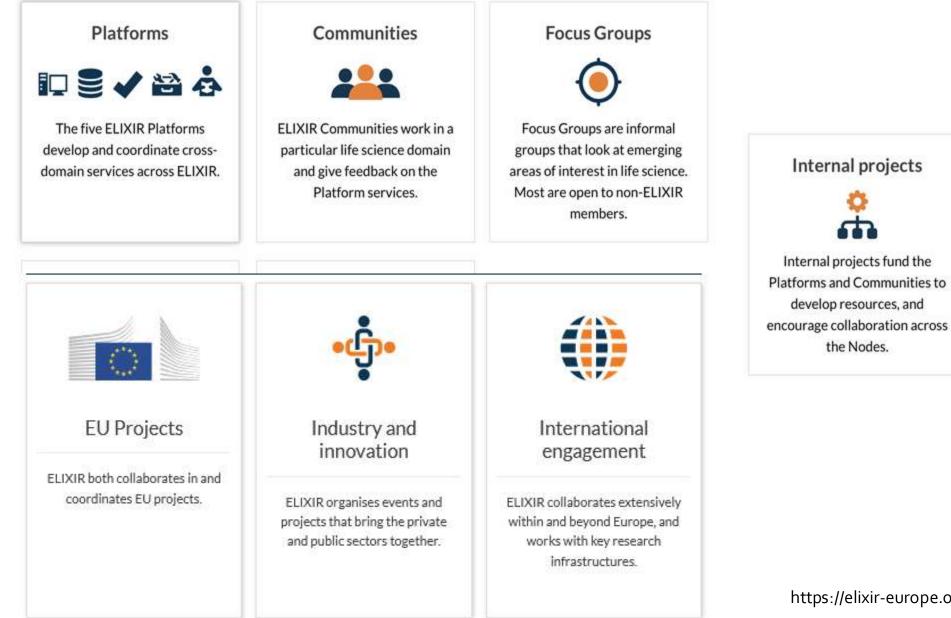
An operational agreement has been recently signed between EPTRI-IT and ELIXIR-IT

Goals

- To foster the development of innovative solutions in paediatric data management and analysis
- To promote the adoption of FAIR and Open Science principles in paediatrics
- To promote the integration of paediatric research in national and European infrastructures
- To drive digital innovation in health in paediatrics
- To promote the joint application to funding calls



ELIXIR - How we work



Technical and Training Coordinators advise the Platforms and promote collaboration between the Platforms and the Nodes.

Coordinators



https://elixir-europe.org/how-we-work

ELIXIR Platforms

Compute

Omics

(Italy only)

Platforms bring together experts from Nodes to develop ELIXIR's technical vision and coordinate activities in defined technical areas. There are five Platforms:

Builds and integrates cloud, compute, storage and access services for the life-science research community

- DataDrives the use, re-use and value of life science data by providing
robust, long-term sustainable data resources within a coordinated,
scalable and connected data ecosystem
- ToolsHelps communities find, register and benchmark software tools;
maintains information standards and produces, adopts and promotes
best practices for tool development
- Interoperability Helps people and machines to discover, access, integrate and analyse biological data; encourages the life science community to adopt standardised file formats, metadata & vocabularies
- TrainingStrengthens national training programmes; grows bioinformaticstraining capacity and competence across Europe; empowersresearchers to use ELIXIR's services and tools

Provides services for **genomics**, **transcriptomics**, **metabolomics** and **proteomics** data production





ELIXIR Communities –

connecting infrastructure & life science experts



Formed around domain experts in ELIXIR Nodes (including non-ELIXIR partners)

- Provide a mechanism for long-term collaborations with other ESFRIs and large-scale initiatives
 - Drive service developments in the ELIXIR Platforms



Provide a framework to develop and maintain community standards



The <u>ELIXIR Communities Handbook</u> tells you what a Community is, who can join, what the benefits are, and how Communities are structured.



ELIXIR Infrastructure Landscape

Communities & Focus groups

3D-BioInfo

Helps to understand the 3D structure of macromolecules like proteins and DNA.



Biodiversity

Aims to help us understand biodiversity, and how it responds to changing environmental pressures.

Food and Nutrition

Aims to help us understand the effect of food choices on human health.



Galaxy Fosters a Galaxy community in Europe, together with Galaxy resources and training.

Metabolomics

Provides the resources, analysis tools and infrastructure to help metabolite identification.



Microbial Biotechnology Helps the development of tailor-ma

Helps the development of tailor-made microbes and biological systems.

Microbiome

Promotes standards, training, and other resources for microbiome research across all biomes.

Plant Sciences Develops an infrastructure to facilitate genotype-phenotype analysis for crop and tree species.

Single-Cell Omics

cell and spatial omics.

Proteomics

Develops and maintains sustainable proteomics tools and data resources.

Research Data Management Brings together RDM experts to coordinate and promote the use of RDM resources.

Systems Biology

Aims to make systems biology modelling a central pillar of research in biology.



Toxicology Supports the integration of standards, tools and resources to aid toxicology research projects.

Intrinsically Disordered Proteins

Develops standards, tools and resources

Identifies and addresses challenges in single-

to help identify and characterise IDPs.

Human Data Communities



Federated Human Data Develops long-term strategies for managing and accessing sensitive human data.



Cancer Data Enhancing data for precision medicine by improving data management, interoperability and accessibility.

Current Focus Groups

- Biocuration
- Domestic Animals Genome and Phenome
- Environmental Impact
- EOSC
- FAIR Training
- Health Data
- Learning Paths
- Machine Learning (AI)
- Pathogen Data
- Professionalising Careers in Research Infrastructures
- Research Data Alliance (RDA) Activities

Human Copy Number Variation Aims to make it easier to detect, annotate and interpret human Copy Number Variations (hCNVs).



Rare Diseases Supports the development of new therapies

Next presentation

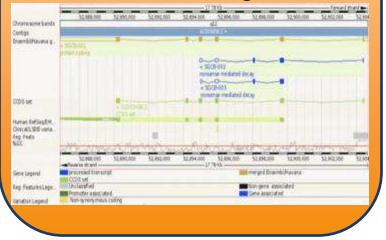


Data Platform

The ELIXIR Data Platform supports:

Core Data Resources

Databases of fundamental importance for life sciences research and involved in the long-term conservation of biological data.



Deposition Databases

Recommended repositories for the deposition of experimental data.

Dataset ID	Description	Technology
EGAD00000000001	WTCCC1 project samples from 1958 British Birth Coh	Aflymetrix 500K
EGAD0000000002	WTCCC1 project samples from UK National Blood Se	Affymetrix 500K
EGAD0000000003	WTCCC1 project Bipolar Disorder (BD) samples	
EGAD0000000004	WTCCC1 project Coronary Artery Disease (CAD) sa	
EGAD0000000005	WTCCC1 project Inflammatory Bowel Disease (IBD) s	
EGAD0000000006	WTCCC1 project Hypertension (HT) samples	
EGAD00000000007	WTCCC1 project Rheumatooid arthritis (RA) samples	
EGAD0000000008	WTCCC1 project Type 1 Diabetes (T1D) samples	
EGAD0000000009	WTCCC1 project Type 2 Diabetes (T2D) samples	
EGAD0000000010	WTCCC1 project Ankylosing Spondylitis (AS) samples	Illumina 15K
EGAD0000000011	WTCCC1 project Autoimmune Thyroid Disease (ATD)	illumina 15K
EGAD0000000012	WTCCC1 project Multiple Sclerosis (MS) samples	
EGAD0000000013	WTCCC1 project Breast cancer (BC) samples	Illumina 15K

Data Resource Services

List of services provided by different nodes.

iearch this section	Search		
Name of service		Tag	Related links*
AspicDB	0		Dis Non
Berlin RNA Toolbox	0	Genes and genomes	
снорснор	0		and the second
ENZYME	0	Enzymes, interactions and pathways	Shittens?
Glyco@Expasy	0	Proteins and proteomes	BULANCE.
HCVIVdb	0	Molecular and cellular structures	
ITSoneDB	0	Genes and genomes	(Sector)

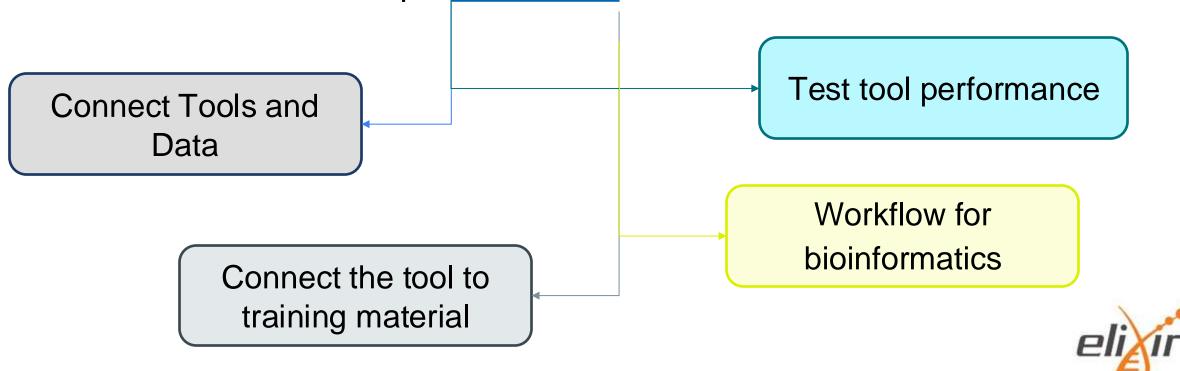
ELIXIR Data Platform facilitates the access to vast Omics datasets, adhering to **FAIR** principles and embracing **Open Science**. This approach fosters the development of innovative applications, products, and services

Data Platform – Some of the core resources



Tools Platform

- Facilitate access to and use of bioinformatics tools
- Define and promote the use of standards for tool documentation (tool descriptions) and associated metadata
- Define and promote Best Practices for the development and maintenance of developed tools



Tools Platform



<u>**Bio.tools**</u> is a registry of tools and databases (~30k), managed by the community. By using controlled ontologies, it allows users to quickly find the most appropriate tool for their needs.

<u>WorkflowHub</u> is a domain-independent registry for workflows designed based on FAIR principles.

<u>**OpenEBench**</u> represents a platform for benchmarking tools and FAIRification of reference datasets. It is also designed to support the principles of Open Data, Open Source, and Open Science.





- 1. <u>Biocontainer</u> for gathering individual efforts for tool containerization and platform federation for sustainability.
- 2. Central repository for managing containers and their distribution.
- 3. User-driven development to identify the most suitable solutions for users.

Compute Platform

- It is responsible for **providing high-level Computing and Storage services** specialized in supporting the applications of the Elixir community.
- It is composed of several technological partners whose know-how and infrastructure represent the backbone of the node's ICT infrastructure.

Current Technological Landscape

The latest HW and SW technologies are used to implement, through intense **codesign activities with the bioinformatics community**, specialized services that are as closely aligned as possible with the requirements of researchers.



Compute Platform – ELIXIR-IT



(Mirror) CNR Napoli: 5 PB storage (Backup)

UniMi

5 PB storage

CNR Milano:

~ 5000 CPU cores

LineDitxel elixir

INDACO Update UniBo IT infrastructure update

UniPd IT infrastructure update







4.192CPU cores

2,1 PB storage; 10Gb network CNR:

12.320CPU cores, 10 GPUs 7,2 PB storage; 25Gb network

UniBa (Physics Dept.) 4000 CPU cores and GPUs 5.5 PB Cloud Storage 2 PB Posix Storage 20 PB Tape Library



NextGeniT

elivir

BiOmici

Uniba (Physics Dept.) 5 PB Storage HPC/HTC 250 CPU cores HPC with GPUs 1.5 PB Storage Cloud

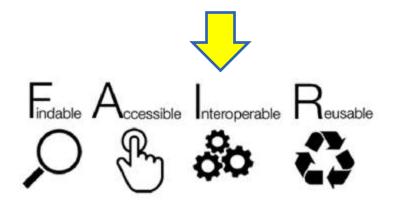


CNR and University of Bari:

Interoperability Platform

What is needed to efficiently connect the Data, Tools and Computing facilities?

- **Standards**: formats reporting, guidelines, vocabularies, ontologies
- Metadata services: ontologies, annotations, validation, indexing, search
- **Registers**: services and datasets
- Search engines for tools and datasets
- Identifiers: resolution and management
- Harmonisation and sharing of tools, workflows and pipelines
- Common programmable interfaces
- Best practices





Interoperability Platform - ELIXIR-IT Services

https://elixir-europe.org/platforms/interoperability/rirs



Catalog of standards and usage and management policies for data and metadata <u>https://fairsharing.org</u>



Practices, recipes, and guidelines for making data FAIR

https://rdmkit.elixir-europe.org/

https://faircookbook.elixir-europe.org



Markup models to improve the findability of resources on the Web (datasets, software, training) <u>https://bioschemas.org</u>



Web resource resolution service that provides consistent and persistent access to data with compact identifiers

http://identifiers.org/

Interoperability Platform - ELIXIR-IT Services

https://elixir-europe.org/platforms/interoperability/rirs



Benchmarking system for methods, tools, and bioinformatics web services

https://openebench.bsc.es



Repository of biomedical ontologies providing a single access point

https://openebench.bsc.es



Interoperable open-source platform for scientific data analysis and sharing <u>https://usegalaxy.eu/</u>



Open standard for the description of workflows and analysis tools

https://www.commonwl.org



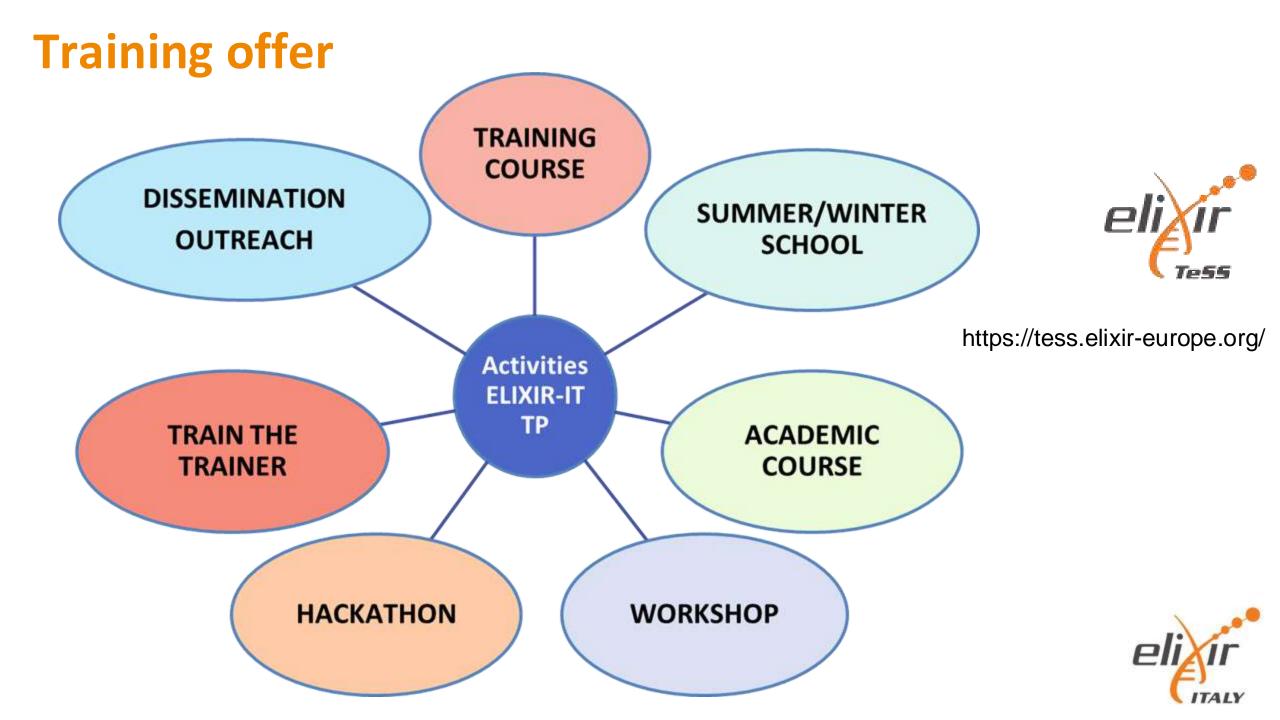
Data warehouse system integrating biological data sources and facilitating data interrogation and analysis

http://intermine.org

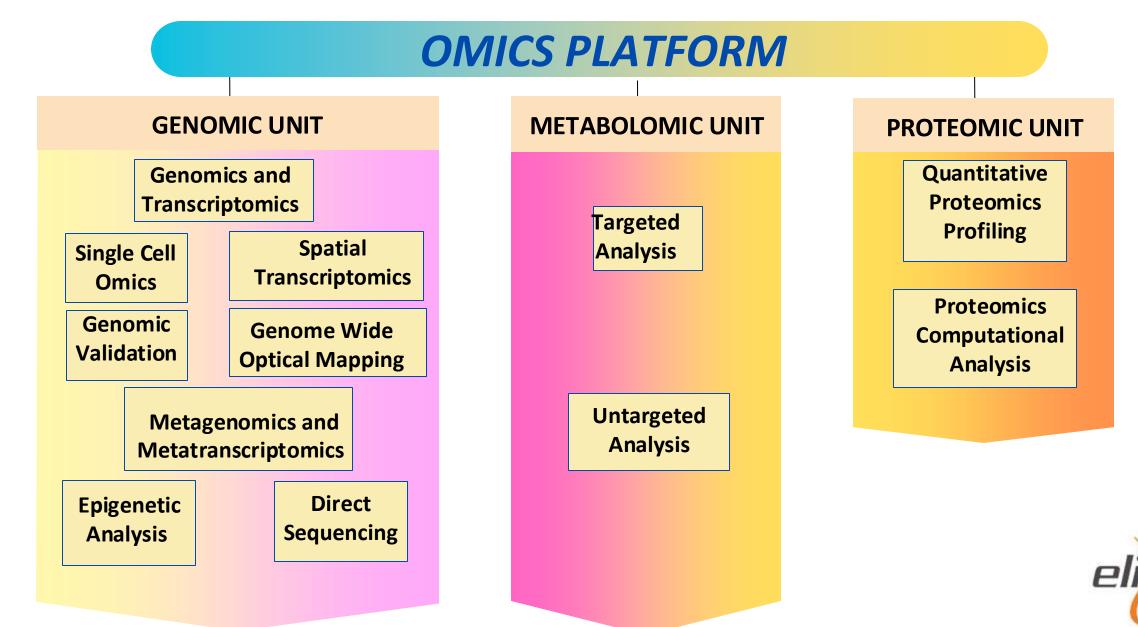
ELIXIR-IT Training Platform Mission

Training in bioinformatics is essential to achieve excellence in scientific research			
Support researchers and professionals in acquiring bioinformatics skills	Promote effective teaching and learning practices		
Establish national and international collaborations	Create a community of instructors and educators		





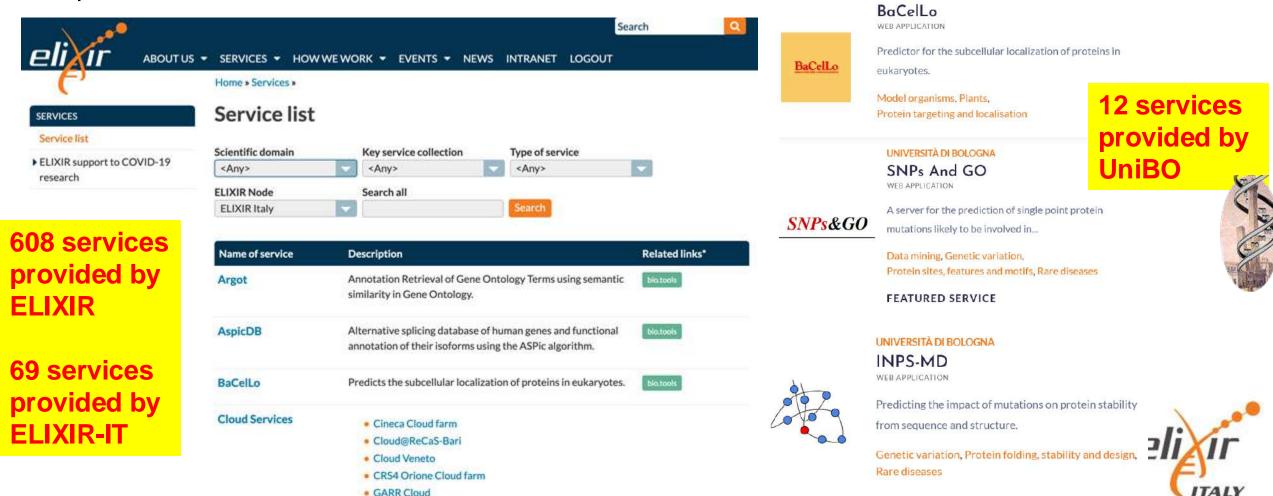
The Omics Platform



ITALY

ELIXIR Services

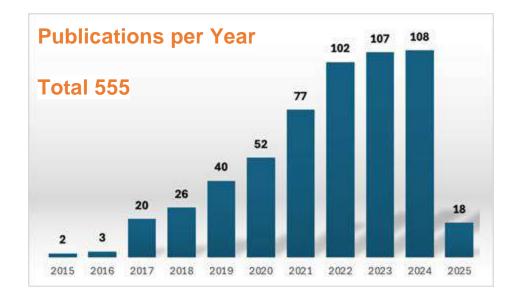
The Italian node of ELIXIR offers a vast portfolio of high quality bioinformatic services including data, tools, compute, standards, compute and training provision as well as "Omics" data production.

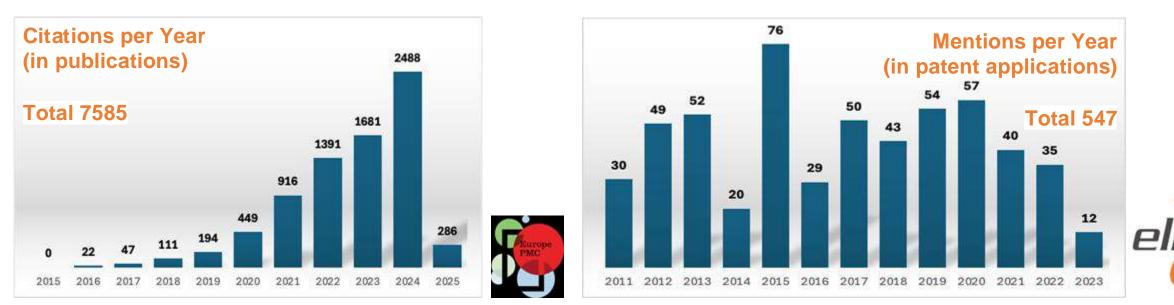


ELIXIR-IT Impact

Last update February 2025

ITALY





National and international projects empowered by ELIXIR-IT







Thanks for your attention





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