ELIXIR is building a sustainable and long-term infrastructure for Human Genomics and Translational data in Europe. This will support life science research and its translation to medicine globally.

Genomics technologies have advanced rapidly. This has resulted in a shift in the challenges within human genomics from data generation to data management and infrastructure. Between 100 million and 2 billion human genomes could be sequenced by 2025.¹ To realise its potential, genotype data need to be linked to a range of phenotype data e.g. –omics, imaging, wellness, electronic health records, lifestyle and family history. For the scientific and medical community to use these data effectively, we need an infrastructure that enables researchers to find, manage, store, share, access, link and reuse data, whilst respecting data privacy regulations. This cannot be achieved by one single institute or project. ELIXIR addresses this need by providing a federation of resources connected through a robust technical infrastructure.

Examples of ELIXIR Human Data activities

Collaborating with global initiatives

Global Alliance for Genomics and Health (GA4GH) beacons provide a discovery service for genetic data. The service enables researchers to share data. Users are able to identify organisations that might have data for a variant they are interested in. ELIXIR is bringing European data forward for international data discovery by implementing beacons in the EGA and in national flagship projects at a number of ELIXIR Nodes, including Finland, Sweden, the Netherlands, France, Belgium and Switzerland. In addition, ELIXIR Beacons are developing a three-tier access model (public – registered – controlled) to support the provision of data with varying levels of openness.

European Genome-phenome Archive (EGA)

The EGA forms the backbone for many ELIXIR projects, which aim to extend the access, authorisation and secure data transfer developed in the EGA, and make it available to clinical and translational researchers across the ELIXIR Nodes and beyond. The EGA provides a service for the permanent archiving and access-controlled distribution of personally identifiable genetic and phenotypic data. EMBL-EBI launched the EGA in 2008, in 2014 EGA expanded to the Centre for Genome Regulation (part of ELIXIR Spain) as a joint venture.

Collaboration with Industry

Collaborating with OncoTrack (Innovative Medicine Initiative project pioneering the use of large-scale genomics to improve the early diagnosis of colon cancer), ELIXIR set out to understand and document the long term data storage and metadata mapping requirements for heterogeneous data (clinical, genomic, animal study) using the EGA.

Infrastructure for data analysis

A scoping study with the TraIT project (initiated by the Dutch Center for Translational Molecular Medicine) served as a use case to develop IT infrastructure for translational research. The collaboration with ELIXIR connected TraIT’s data analysis platform (Galaxy) and the data portal (tranSMART) with the EGA and enabled Dutch researchers to use EGA as the long-term storage solution for raw data.

Local EGA

Local instances of the EGA model established at a number of ELIXIR nodes will enable the storage of sensitive, access-controlled data that may – for a variety of reasons – not leave its country of origin. Integration of the associated metadata in EGA will ensure discoverability of such data at an international level.

Services to rare disease research community

An ELIXIR registry of data sources and analysis tools critical for the development of rare disease research is currently being built. It will enable researchers to discover, access and analyse different rare disease repositories across Europe.

www.elixir-europe.org/seat-cases/rare-diseases

ELIXIR is an intergovernmental organisation that provides long-term perspective and has legal agreements in place to operate as a unified distributed infrastructure.

ELIXIR unites Europe’s leading life science organisations in managing and safeguarding the increasing volume of data being generated by publicly funded research. It coordinates, integrates and sustains bioinformatics resources across its member states and enables users in academia and industry to access vital services for their research.

Contact us

If you generate or use human data as part of your research, ELIXIR can help you to store, discover, access, analyse and share data of multiple types and origins. ELIXIR also arranges courses, workshops and hackathons to train researchers to use ELIXIR services.

For more information on existing project or to discuss proposals for new initiatives, contact Serena Scollen, ELIXIR Head of Human Genomics and Translational Data.

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Learn more at:
www.elixir-europe.org/use-cases/human-data

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