



Federation of sensitive human genomics data

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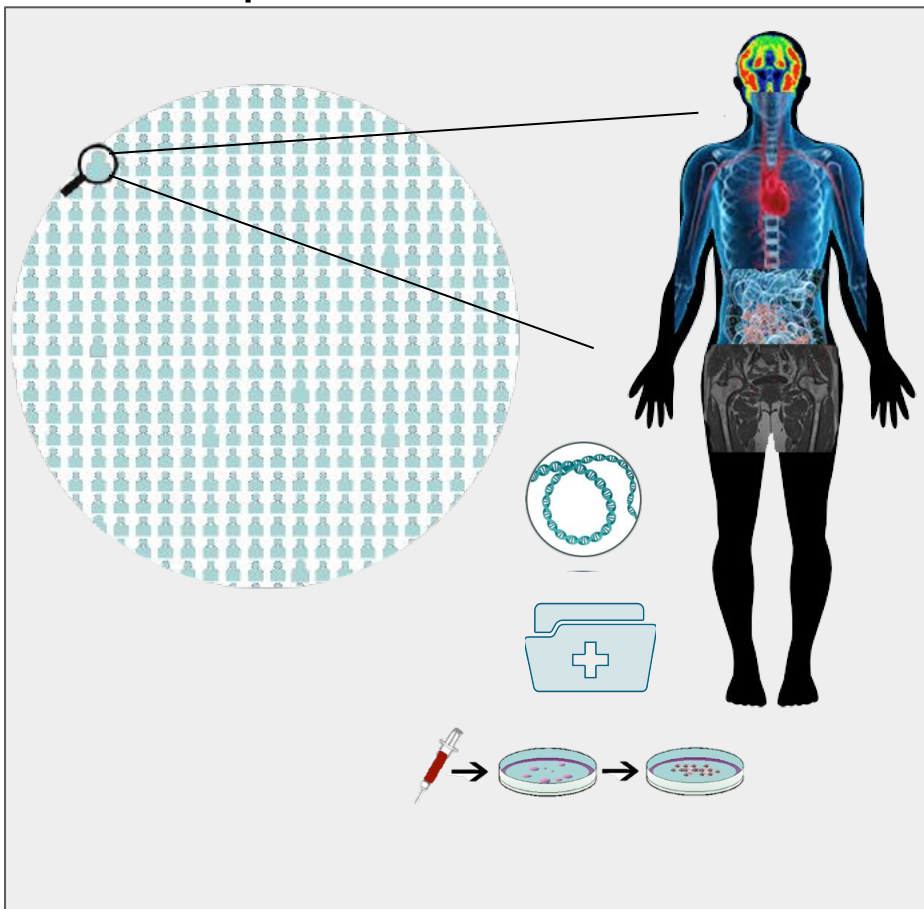
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Cross-border access to sensitive human genomics data

Unleash the potential



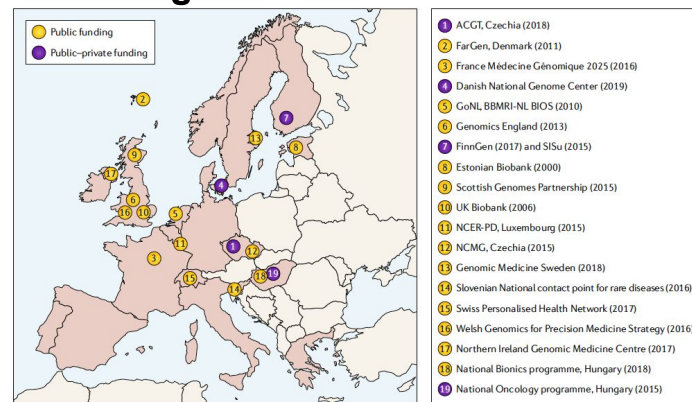
Percentage human genomes and exomes funded by the healthcare systems

1%
2012

20%
2017

80%
2022

National genomics initiatives



Human Genomics Data: changing environment

- Most sequencing of the future will be for healthcare not research
- 60M virtual cohort by 2025

CHALLENGES

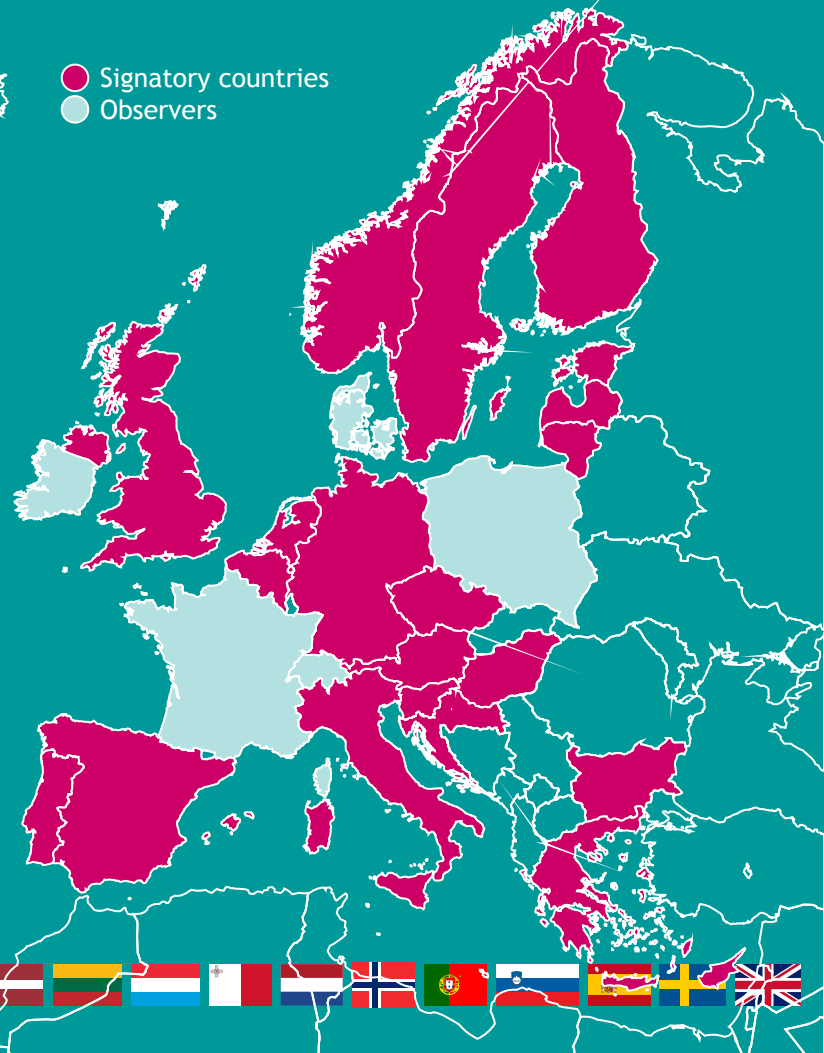
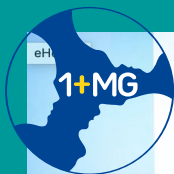
- Data geographically distributed
- Healthcare data is a national competence and often not able to leave jurisdiction(s)
 - Huge volumes of data testing limits of centralised model
 - Clinical data are of variable quality and not interoperable
- Healthcare is not used to this type and amount of data: terabyte to exabyte
 - Technical knowhow is in the research community
- Attitudes and action towards open data need to progress
 - Secure access and governance

1+MG Declaration of cooperation: 2018



● Signatory countries
● Observers

23 countries and 5 observers



1+MG Roadmap

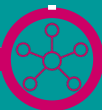
Timeline & Objectives

Adopted in February 2020

2020

ENGAGE

Governance,
cooperation and
collaboration



2021

TRANSLATE

Infrastructure,
guidelines and
pilots



2022

DRIVE

Sharing, scaling
and sustaining



Drive adoption
and support
long-term
operations via
a maturity
model and a
methodology
for economic
evaluation

**1+ Million
accessible
genomes**

The B1MG project

Coordination and support action of the 1+MG

Started 1 June 2020

29

Partners

€4 M

Budget

3

Years

25

EU countries

B1MG objectives

Aligned to the 1+MG roadmap



2020

Engaging local, regional, national and European stakeholders

... to define requirements for accessing genomics and personalised medicine data.

2021

Translating requirements for data quality, standards, technical infrastructure, and ELSI

... into technical specifications and implementation guidelines.

2022

Driving adoption and supporting long-term operations

... via the B1MG maturity level model and a methodology for economic evaluation.

Develop Europe's future health data landscape

3 years



Maturity Model

ELSI toolkits

Technical recommendations and guidelines

European Health Data Space



Long-term strategy — Use Cases working groups

Cancer, infectious diseases, rare diseases, common complex disease

Coordinating action vs implementation

3 years



Coordination, Guidance and Recommendations

Adoption and implementation

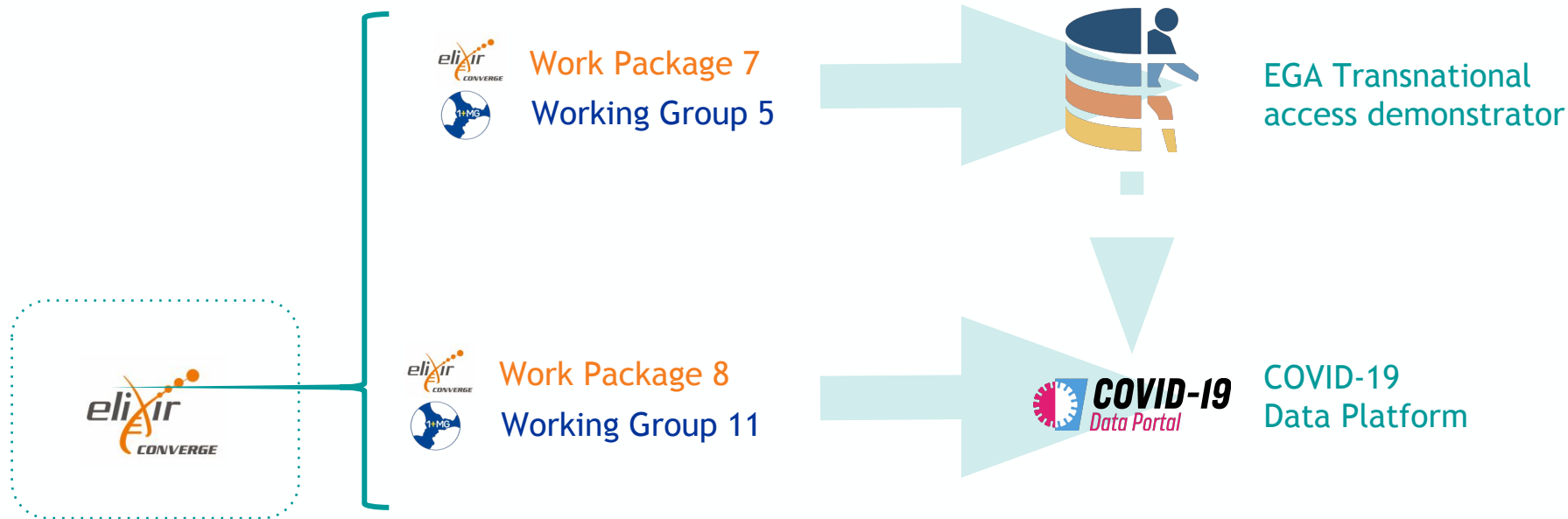


HEALTHYCLOUD
Health Research & Innovation Cloud



Adoption and Implementation

ELIXIR-CONVERGE Case



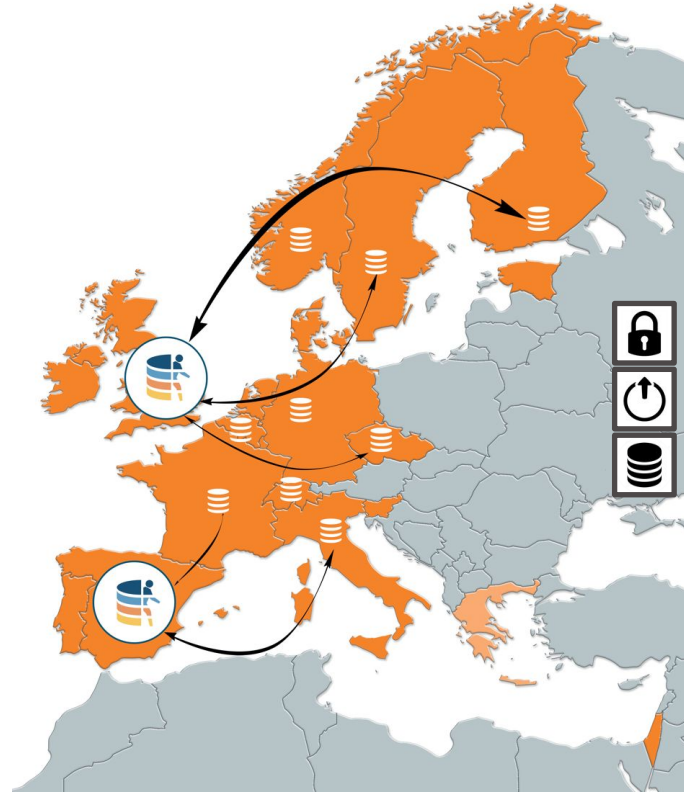
Federation of sensitive human data

- 1+ Million Genomes Initiative driving federated sensitive human framework from the EU + EEA Member States
- Beyond One Million Genomes is the Coordination and Support Action to support the 1+MG Initiative
- The technical and policy developments are resourced and coordinated from many projects
- The Federated EGA framework is enabling sensitive data archival at the National and regional levels, connected to the pan-Europe framework



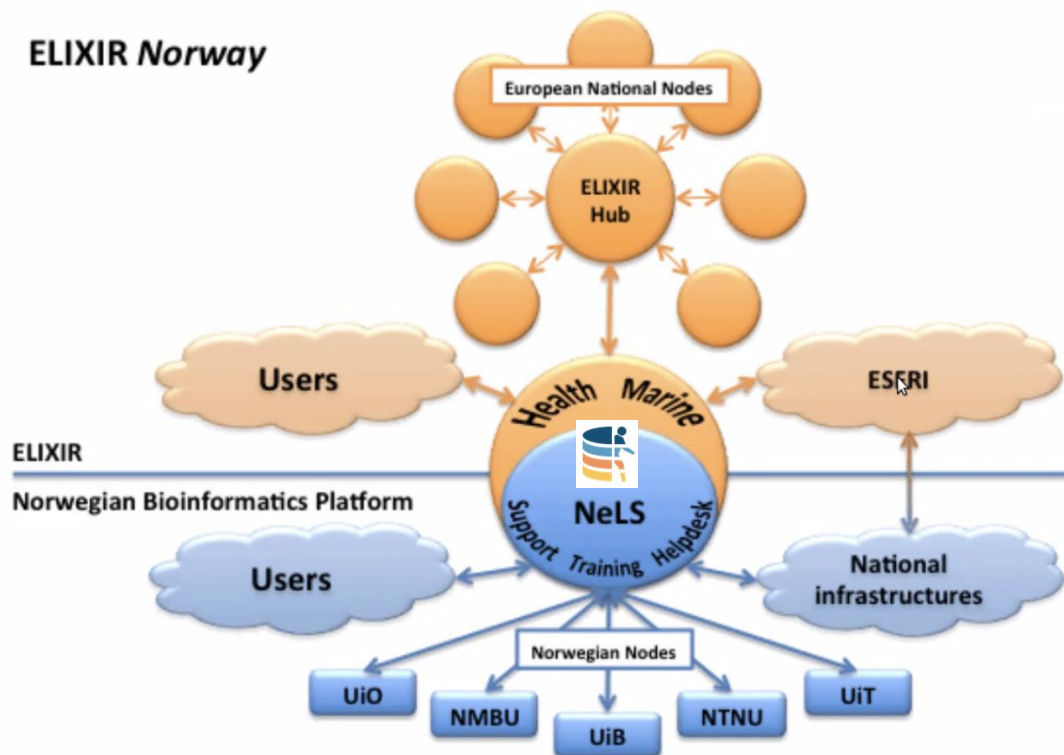
Federation of human genomic data

- Many national datasets from human research participants needs to be **stored locally** (European Genome phenome Archive – EGA)
- **Federated EGA** is a framework that can be joined:
 - ELIXIR developing a federation with shared metadata (FAIR) and local data store (secure)
 - ELSI framework
 - Technically this is based on suite of interoperable, reusable, adopted, and fit-for-purpose **standards**
- 17/23 ELIXIR Nodes are funded in the **ELIXIR FHD Community**, where Federated EGA is the driving product
- Use cases: Rare Diseases, Cancer, Common Complex Diseases, Infectious Diseases



Federation in regions, an example

ELIXIR Norway



- Norway has 5 regions (4 health regions)
- Norwegian Nodes across 4 of the 5 regions in the country
- Share sensitive human research data from the Nodes to a central Norwegian Node
- Agreements allow the central Node to hold the data and act as Data Controllers (based on appropriate Data Processing Agreements)



Thank you