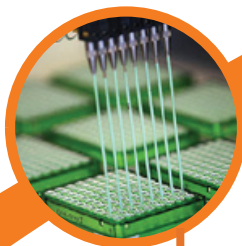


ELIXIR's mission

To build a sustainable European infrastructure for biological information, supporting life science research and its translation to:



society



bioindustries



environment



medicine

Thank you!

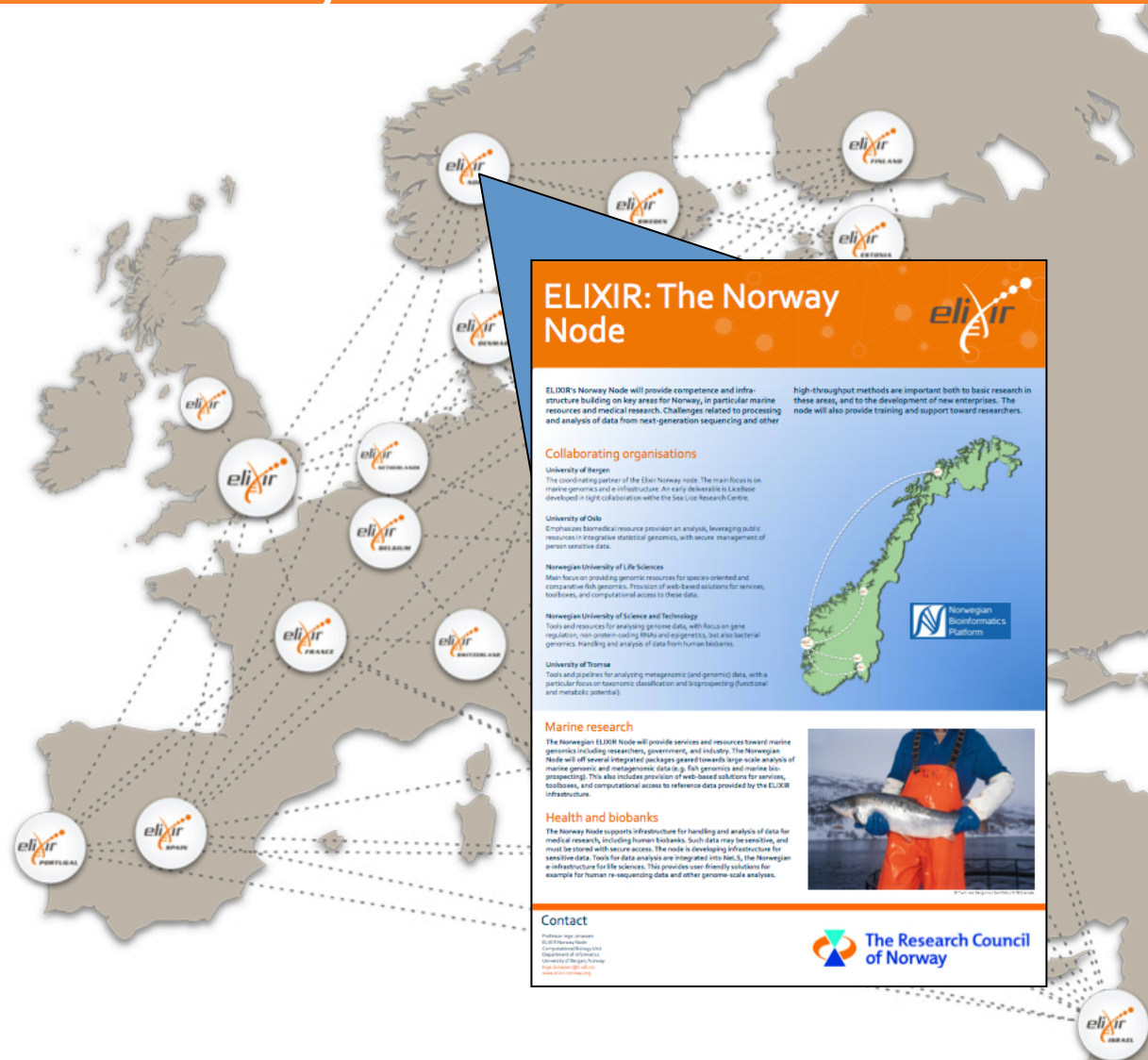
ELIXIR

- Elixir Consortium Agreement (ECA) entered into legal force Jan 2014
- 11 members signed to date
 - Czech Republic, EMBL, Denmark, Estonia, Netherlands, Norway, Israel, Portugal, Sweden, Switzerland and the UK
- Further 8 countries have signed MoU and are working towards national signatures
- Discussions on-going with additional prospective member states



A distributed infrastructure to scale with the challenges

- **ELIXIR** deliver services through national ELIXIR Nodes
- **ELIXIR** Nodes build local bioinformatics capacity throughout Europe
- **ELIXIR** Nodes build on national strengths and priorities



ELIXIR: The Norway Node

ELIXIR's Norway Node will provide competence and infrastructure building on key areas for Norway, in particular marine resources and medical research. Challenges related to processing and analysis of data from next-generation sequencing and other high-throughput methods are important both to basic research in these areas, and to the development of new enterprises. The node will also provide training and support toward researchers.

Collaborating organisations

University of Bergen
The coordinating partner of the Elixir Norway node. The main focus is on marine genomics and infrastructure. An extra deliverable is to be developed in tight collaboration with the Sea Life Research Centre.

University of Oslo
Emphasizes biomedical resource provision analysis, leveraging public resources in integrative statistical genomics, with secure management of personal sensitive data.

Norwegian University of Life Sciences
Main focus on providing genomic resources for species-oriented and comparative fish genomics. Provision of web-based solutions for services, toolboxes, and computational access to these data.

Norwegian University of Science and Technology
Tools and resources for analysing genomic data, with focus on gene regulation, non-coding RNAs and epigenetics, but also bacterial genomics, handling and analysis of data from human biobanks.

University of Tromsø
Tools and pipelines for analysing metagenomes (and genomic) data, with a particular focus on taxonomic classification and bioprospecting (functional and metabolic potential).



Marine research
The Norwegian ELIXIR Node will provide services and resources toward marine genomics including researchers, government, and industry. The Norwegian Node will offer several integrated packages geared towards large-scale analysis of marine genomics and metagenomic data (e.g. fish genomics and marine bioprospecting). This also includes provision of web-based solutions for services, toolboxes, and computational access to reference data provided by the ELIXIR infrastructure.

Health and biobanks
The Norway Node supports infrastructure for handling and analysis of data for medical research, including human biobanks. Such data may be sensitive, and must be stored with secure access. The node is developing infrastructure for sensitive data. Tools for data analysis are integrated into HPL5, the Norwegian e-infrastructure for life sciences. This provides user friendly solutions for example for human re-sequencing data and other genome-scale analyses.

Contact

Professor Espen Sævi
Bioinformatics Center
Department of Informatics
University of Bergen, Norway
esp@iuh.uib.no

The Research Council of Norway



<http://www.elixir-europe.org/about/elixir-nodes>

ELIXIR Infrastructure

- **Data**

Sustain core data resources

- **Tools**

Services & connectors to drive access and exploitation

- **Compute**

Access, Exchange & Compute on sensitive data

- **Standards**

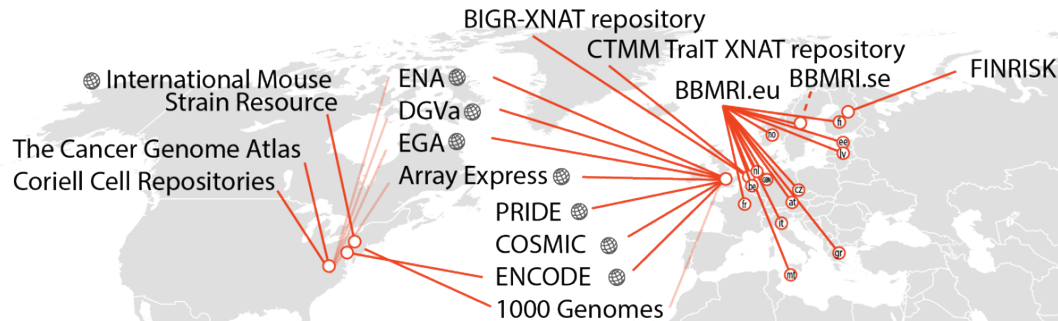
Integration and interoperability of data and services.

- **Training**

Professional skills for managing and exploiting data



Standards – and the tools to make use of them...



Consistent identification of a biosample:
BioSD finds & links 2.8M samples from 32 sources
worldwide

Example query: "Cardiac Arrhythmia" yields over 450 samples from 10 groups (via BBMRI, Array Express, ENA)

Search results

BBMRI.eu : Atrial Fibrillation Network Munich - M4-Cluster-Biobank

BBMRI.eu : Atrial Fibrillation Network Munich

BBMRI.eu : Atrial Fibrillation Network Munich

12 Homo sapiens samples from ENA SRA

Transcription profiling of mouse rapidly stimulated atrial myocytes: Conserv

Transcription profiling of mouse model of cardiac failure - particulate matter

Transcription profiling of human atrial and ventricular myocardium from pati
ventricular non-failing myocardium to identify the transcriptional basis for ul

Gender dependent differences in molecular electrophysiological targets in fa

NHLBI GO-ESP: Family Studies (Familial Atrial Fibrillation)

Using iPSC-derived neurons to uncover cellular phenotypes associated with f

Circulating microRNAs to predict neurological outcome after sudden cardiac

Valvular heart disease and atrial fibrillation regulate microRNA expression profiles in left and right atria differenti

LmnaN195K Mouse Model

Gene profiling of Hand2 target genes and transcriptional regulation of Hand2 expression in the postnatal myocardiu

Transcription profiling of rat heart transplants from Lewis to Lewis and Lewis to F344 strains with and without cold s
ischemia.

Gene expression analysis of cardiac left-ventricle tissue from hybrid mice harboring the Scn5a-1798insD/+ mutatio

Rac1-Induced Connective Tissue Growth Factor regulates Connexin 43 and N-Cadherin Expression in Atrial Fibrillati

Molecular Remodeling of Ion Channels in Human Atrial and Ventricular Myocytes Associated with Ischemic Cardiom

cardiac a



cardiac arrhythmia

Brugada syndrome
ventricular fibrillation
Familial short QT syndromw

atrial fibrillation

Catecholaminergic polymorphic ventricular

Familial long QT syndrome

Timothy syndrome
Jervell and Lange-Nielsen syndrome
Romano-Ward Syndrome

acquired long QT syndrome

sudden cardiac arrest

atrial flutter

cardiac atrium

ELIXIR 2014-15 Objectives

Build ELIXIR Community

- Pilot actions , workshops to drive Node-Node collaborations
- User communities / domain services

Visible and useful services

- Collate Node resources, tools and services – implement Node proposals
- Establish quality metrics, visualize through Tools&Service Registry

Sustainable Data management, archive and reuse

- Identify and Name “ELIXIR Core Resources”
- Data interoperability

What does this mean in practice?

- Establish ELIXIR Nodes:
 - Ensure services are visible through service registry. And the tools to support this
 - Put the legal agreements in place
 - Support national roadmap
- Run a series of *ELIXIR Pilot Actions*
 - Test assumptions from strategy through implementation...
 - Support the technical coordinator group – build on taskforces
 - Support the formation of “domain user communities”
- Establish *ELIXIR Core resources*
 - Data resources of exceptionally high value to user communities
 - Develop / coordinate data to support policy actions
 - Interoperability and good data management

Pilot actions

- 🔬 Pilot actions inform the development of ELIXIR Strategy and technical choices without committing the infrastructure to the long-term
- 🔬 Pilot actions will allow ELIXIR to test options for strategy implementations for subsequent upscaling and delivery
- 🔬 Pilot actions are a mechanism to run focused “sprints” to test / demonstrate services in collaboration with user communities and other research infrastructures
- 🔬 Pilot actions should foster collaborative working between the ELIXIR Nodes and allow for the development and exchange of best practice.



ELIXIR Pilots – technical challenges for biomedical research:



1. Cloud computing
“Embassy cloud”: Access reference data in a virtual environment – work as though you are at EMBL-EBI or SIB, Switzerland
2. Authentication & Authorisation
Improved methods and processes for access to clinical data
3. High-Performance Computing
“Lightpath”: Connections for on-demand reference data to remote HPC centres at EMBL-EBI and CSC Finland

How could ELIXIR build an agri – community?

A few ideas to start discussion:

- 🧬 Genome annotation working groups?
- 🧬 There is much work on multi omics / phenotype integration for disease biomarkers – pilot applicability of tools?
- 🧬 Sustainability planning for on-going projects?
- 🧬 Metagenomics pipelines, annotation and data sharing?
- 🧬 ...

Thank you



Belgium



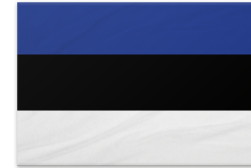
Czech Republic



Denmark



EMBL



Estonia



Finland



France



Greece



Israel



Italy



Netherlands



Norway



Portugal



Slovenia



Spain



Sweden



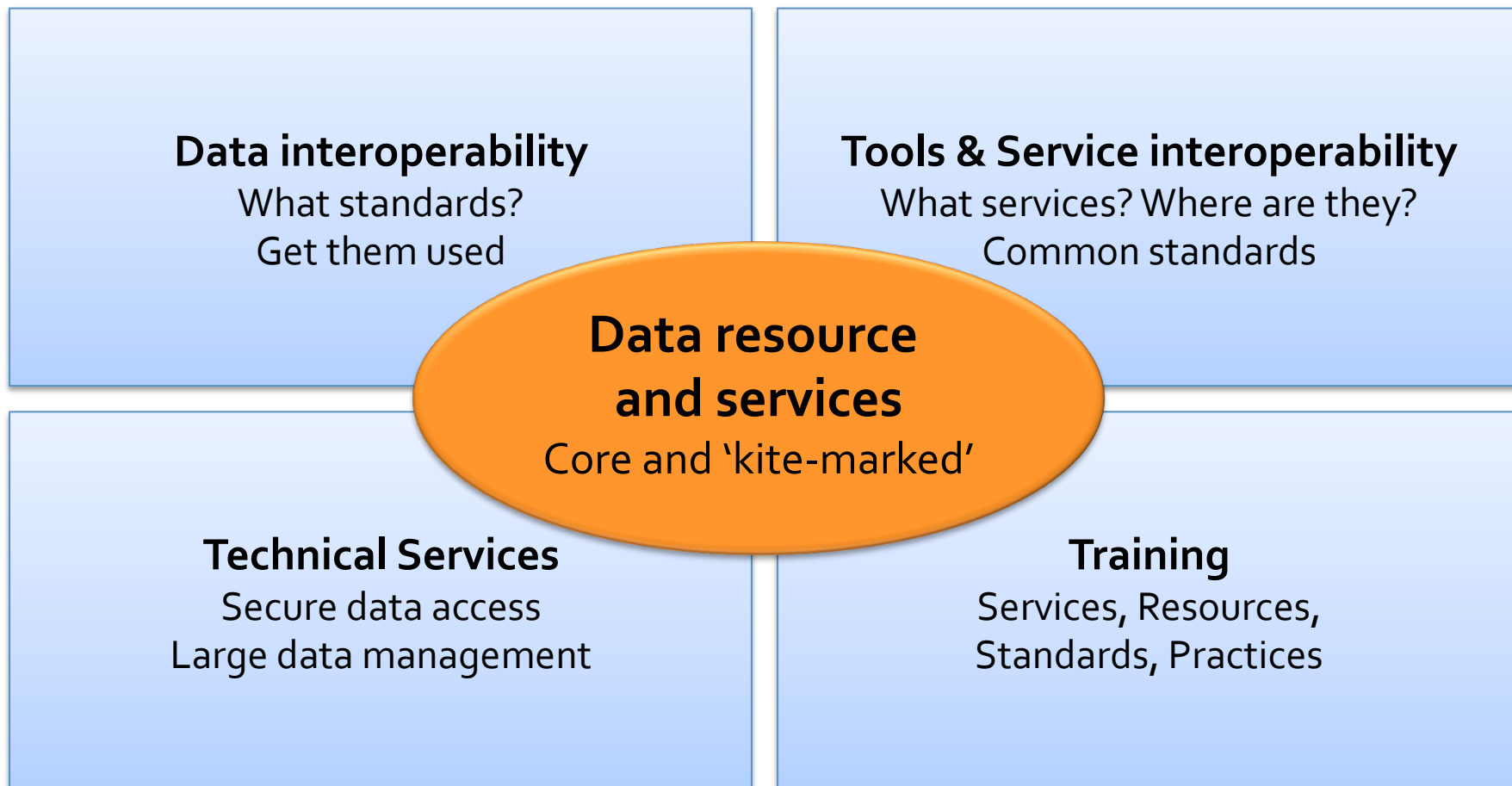
Switzerland



United Kingdom

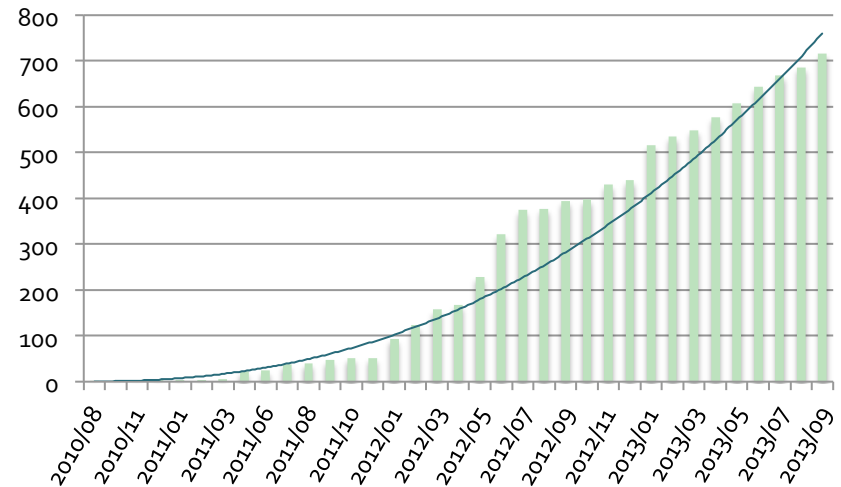


ELIXIR Programme



Sustaining core data resources

- Primary archive for any data consented for sharing in the context of research but not for fully public distribution
 - Secure storage, management and dissemination of data – raw or processed - from biomedical research projects.
 - Phenotypic data collected from the subjects.
 - Submissions must be de-identified and in accordance with the informed consent.
 - Data are packed into datasets that are governed by a Data Access Committee (DAC).
 - Authentication - each DAC approved individual will have a personal EGA account.
 - Authorization – DACs attach access permission(s) to the EGA account(s).
- EGA hosts more than 450 studies and discoverability to the 732 that are in both EGA and dbGaP
- EGA supports more than 400 user requests per month



Under ELIXIR the CRG and the EBI have agreed to “Explore ways in which the CRG’s emerging Node could share responsibility for production of the EGA in future” ...

... which translates into managing peer database representations of the EGA Project hosted jointly by the Hub and the Spanish node of ELIXIR



Challenges for life-science data services

Scale and
Sustain
funding



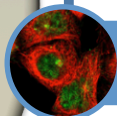
Distributed infrastructure with >1M users



Managing and interoperate big and heterogeneous data



Capacity Compute. Capability Storage



Integrating clinical and translational data



Privacy and ethical concerns



Algorithms to data – clouds, research environments...

Programme of work

