



A European infrastructure for farmed animal genotype to phenotype research

Deliverable 7.1 Roadmap to set up an animal science community within ELIXIR

Grant agreement no°: 101094718

Due submission date

2023-December 31

Actual submission date

2024-01-02

Responsible author(s):

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Confidential: NO

This project has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement No 101094718. The content of this report reflects only the author's view. The European Commission is not responsible for any use that may be made of the information it

DOCUMENT CONTROL SHEET

Deliverable name	Roadmap to set up an animal science community within ELIXIR
Deliverable number	7.1
Partners providing input to this Deliverable	EMBL, INRAE, UEDIN, FBN
Draft final version circulated by lead party to: On date	21/12/2023
Approved by (on date)	
Work package no	7
Dissemination level	PU

REVISION HISTORY

Version number	Version date	Document name	Lead partner
Vs1	21/12/2023	20231221_EuroFAANG_Deliverable 7.1_Roadmap_to_set_up_an_animal_science_community_within_ELIXIR_V2	EMBL
Vs2	21/12/2023	20231221_EuroFAANG_Deliverable 7.1_Roadmap_to_set_up_an_animal_science_community_within_ELIXIR_V2	

Changes with respect to the DoA (Description of Action)

None

Dissemination and uptake

This is a public deliverable

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1. Executive Summary

Background	<p>In the ESFRI landscape of European Research Infrastructures for biology, ELIXIR plays a major role for data standardisation and data sharing which, makes it a major partner for the EuroFAANG Research Infrastructure (RI). Advanced collaboration with Elixir in the field of data standards and data management best practices is included in WP3. Here, the aim is to study the potential added value and the feasibility of establishing an Elixir Animal Agriculture Community to support animal science. This draws from the experience of the existing Plant Science Community, which has strong connections to the EFSRI EMPHASIS project. The governance of Elixir was contacted and agreed to discuss this objective with EuroFAANG RI representatives.</p>
Objectives	<p>To consult the scientific community and other infrastructures in the field of animal science to define the scope, the objectives, and the work program of the targeted 'Community' and to present it for approval by ELIXIR management.</p>
Methods	<p>The first major discussion with ELIXIR management was initiated at the ELIXIR All Hands meeting on 5th-8th June 2023 as part of an invited presentation of EuroFAANG RI at a symposium on "Biodiversity, Food Security and Pathogens" that is part of the developing ELIXIR 2024-2028 programme. Following these discussions a roadmap towards a future Animal Agriculture community was initiated. The first step to be taken was to apply for an ELIXIR focus group in order to mobilize the scientific community and animal phenotyping infrastructure projects. The focus group will list high priority topics, establish working groups and engage the Elixir nodes in order to set up the fully funded ELIXIR community.</p>
Results & implications	<p>The scope of the focus group was initially closely connected to the aim of EuroFAANG RI to enhance capacity for genotype to phenotype research in farm animals in Europe.</p> <p>At the same time, ELIXIR had identified a need for a group on food security as an identified priority area of the ELIXIR 2024-2028 programme. After some discussion between EuroFAANG RI and ELIXIR, there was an agreement to initially take a broader focus on large animal genomes instead of just food security, although this would remain a key area. The scope was then first enlarged to the community working on companion animals, because they share similar challenges as farm animals in terms of genome annotation, and also because this extension would also enlarge the basis of the scientific expertise.</p> <p>Crucially for the objectives of EuroFAANG RI, the scope was then further enlarged to incorporate the animal phenotyping community, which seemed needed because of the need to link in vitro phenotyping (EuroFAANG) to in vivo phenotyping infrastructures such as AQUAEXCEL and PIGWEB. WP6 of EuroFAANG RI will develop a network of projects working on in vivo and in</p>

	<p>vitro phenotyping and aims to link the two more efficiently for genotype to phenotype research.</p> <p>In addition, contacts have been made with representatives of the Plant Science Community to benefit from their experience establishing their ELIXIR community.</p> <p>Finally, the title of the focus group was chosen to be Domestic animal genomes and phenomes. Proposers and members are listed, including support from a number of Elixir nodes, which is a key factor for the ultimate success of moving from a focus group to a fully-fledged community.</p> <p>Next steps will be to present this focus group at the PAG meeting in January 2024, at regular meetings of ELIXIR national nodes of the EuroFAANG RI partners, at ELIXIR days, at the ELIXIR all hands, and at the EAAP conference in September 2024 in order to raise awareness in the scientific community. Discussions will continue with the Elixir hub management and with the heads of the nodes of representative countries to garner the necessary support and momentum to establish a full ELIXIR funded community.</p>
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2. Application for an ELIXIR focus group

2.1 Initial discussion at ELIXIR All Hands 2023

The first major discussion with ELIXIR management was initiated at the ELIXIR All Hands meeting on 5th-8th June 2023, in Dublin Ireland, as part of an invited presentation of EuroFAANG RI at a symposium on “Biodiversity, Food Security and Pathogens” that is part of the developing ELIXIR 2024-2028 programme. Here Peter Harrison presented the EuroFAANG RI and presented a proposal for how a collaboration with ELIXIR could be established (Figure 1). Following discussions with the senior management at this meeting a roadmap towards a future Animal Agriculture community was initiated. ELIXIR proposed that we should first establish a focus group to establish a strategy for engagement and gather the necessary support to transition to a full community. ELIXIR Focus Groups are agile structures that bring interested parties together around a particular topic. Focus Groups are open to any member of ELIXIR plus, where useful, external parties. They are established by a lightweight approval process, at the discretion of the ELIXIR Director. Focus Groups enable the tackling of a technical and scientific challenge that is not already accommodated within the ELIXIR structure to meet to explore possible approaches, in the early stages of development, before a specific strategy and community is developed. In contrast to Communities, Focus Groups won't be eligible for funding via a Community Implementation Study or a Community-led Implementation Study. They can however propose workshops for the annual ELIXIR all hands meeting, apply for travel grants and staff exchange programmes.

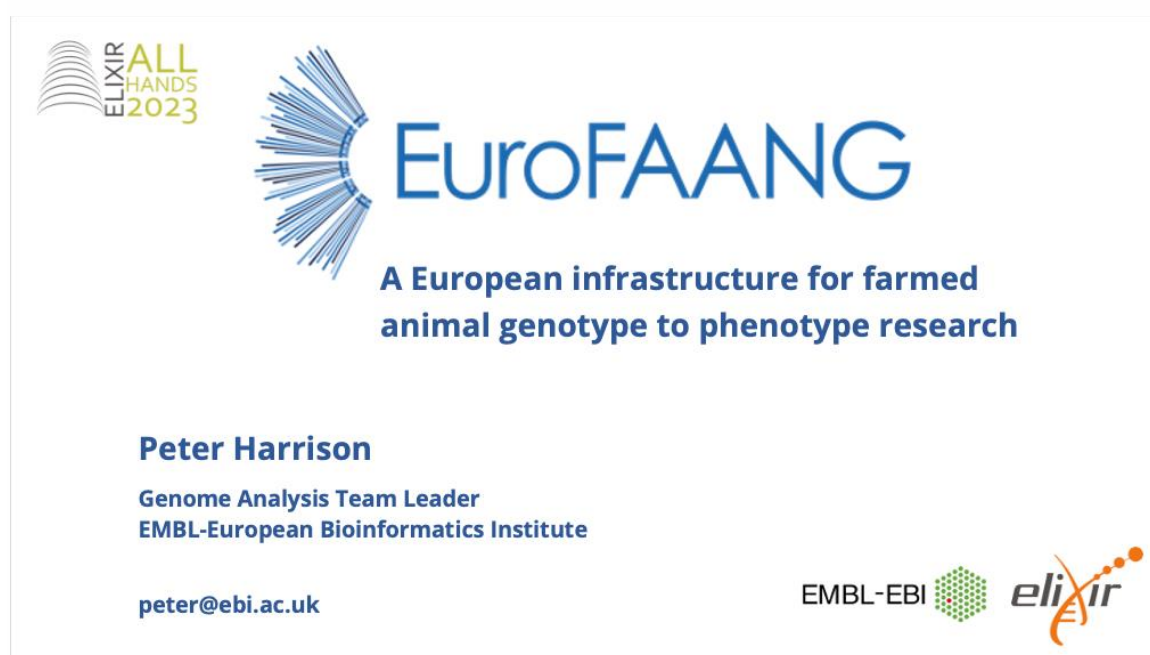


Figure 1. Opening slide of the EuroFAANG RI presentation at the Elixir All hands 2023.

A key part of this process was to establish an appropriate scope of the group. This went through a number of iterations as the group broadened and strengthened its support across plant and phenotyping communities and at the ELIXIR country nodes. The outcomes of these discussions account for the bulk of this deliverable.

The other key discussions that occurred in order to shape in detail the scope of the focus group included:

- 28th July 2023: A virtual meeting with Katherina Heil (ELIXIR Programme Manager for Communities and Training, ELIXIR Hub, EMBL) to discuss the focus group application process and scope definitions. Following this meeting, the initial scope of the focus group was written by Peter Harrison and Michèle Boichard, with further input by email from the EuroFAANG executive and WP leads.
- 22nd September: virtual meeting of the EuroFAANG executive committee to discuss and review the scope from the perspective of EuroFAANG (see 2.3). In this meeting the expansion to include companion animals was agreed and an initial proposal for the chairs of the focus group was established.
- 29th September: A meeting was held between Peter Harrison (EuroFAANG) and Niklas Blomberg at EMBL Heidelberg to run through the high-level scope and ensure support from Elixir Hub for the direction taken.
- Two meetings were held with Cyril Pommier (INRAE) - who chairs the ELIXIR plant animal sciences community and played a key role in establishing the EMPHASIS EFSRI - as an in person meeting at the ELIXIR All hands and a virtual meeting in November. Cyril has agreed to join and support our focus group, and to explore the shared food security focus of the two groups. His expertise in the formation of the plant community and in the interaction between ELIXIR and the EMPHASIS EFSRI will be highly valuable to our efforts. The future success of the community and the future RI will rely on the interaction with the plant agriculture genomics and phenotyping communities.
- 13th November: A virtual meeting between EuroFAANG RI WP leads and René Baumont (INRAE; ELIXIR France), Catherine Larzul (INRAE; ELIXIR France) and Cyril Pommier (INRAE; ELIXIR France) to discuss the role of the emerging phenotyping research infrastructure in the ELIXIR focus group. This resulted in the final updated version of the scope document and the title change to include phenomes (see 2.4). This is a key

development for the future success of both the focus group and EuroFAANG RI.

2.2 First version of the scope

The first version of the focus group scope was tightly linked to the objectives of EuroFAANG RI, whilst factoring in the desire of Elixir to form a group that would tackle key food security issues for Europe.

Preferred title: **'Farmed animal genomes'**

ELIXIR Original Proposed Title: Large Animal Genomes

Other alternative title: 'Agriculture and aquaculture genomes'

One liner: **A scientific community to support genotype to phenotype analysis for agriculture and aquaculture species.**

Vision statement:

Large animal species, both terrestrial and aquatic, have been bred for centuries by human societies. Today, the farming of such species provides an important source of nutrition to humans and plays a critical role in feeding a growing global population. Farmed animal genomes also have a role to play in the One Health challenge, because of the risk of zoonotic diseases. Continuing human population growth and a rapidly destabilising climate poses significant insecurity to our global food system. Agriculture and aquaculture need to adapt to these challenges in order to maintain the benefits they provide to human societies, whilst reducing their environmental impact, conserving biodiversity, and flexibly adjusting to changing societal expectations. Adoption of genomics enabled breeding and management and an improved ability to predict an animal's phenotype using genotype information are key adaptations that are required to meet these challenges. Connecting researchers and data scientists across species and countries is needed to accelerate genotype to phenotype research in agriculture and aquaculture and develop a coordinated open and standardised data life cycle. This ELIXIR community aims at promoting tools, databases, standards, and best practices for farmed animal genomics genotype to phenotype research.

More specifically this ELIXIR community will:

- coordinate discussions and explore potential for data/technology solutions for addressing key issues in welfare, behaviour, health, infectious diseases, and preservation of genetic diversity
- develop standards, coordination, workflows, and visualisation for key developing areas of pangenomics, functional genomics, genome editing, phenotyping and biorepositories, which are needed to enhance genotype to phenotype data analyses
- develop FAIR data management standards and ontologies and promote best practices in data coordination and archiving
- collaborate with key EU research infrastructures in animal science, including EuroFAANG RI, INFRAFRONTIER, PIGWEB and AquaExcel, and key global initiatives such as FAANG and AG2PI
- organise tools and data training and knowledge transfer events

- enhance data reuse and the compatibility of combining data for large scale analyses
- enhance usability and interoperability of the EU data infrastructure for animal genomics
- exchange with other ELIXIR communities, such as the plant sciences and biodiversity communities, on overlapping challenges and potential data/technology solutions.

2.3 Second version of the scope

The expanded second version of the scope included the addition of Companion animals that would broaden and strengthen the effectiveness of the group by drawing on greater expertise. This version also included a first list of the focus group proposers from which the 3 focus group chairs would be elected and an initial list of experts that would form the initial group membership, crucially including a diverse set of Elixir node members.

Title : Domestic animal genomes

One liner : **A scientific community to support genotype to phenotype analysis for farmed and companion animal species.**

Proposers

Peter Harrison (EMBL-EBI; ELIXIR Hub)

Martien Groenen (Wageningen University; ELIXIR Netherlands)

Sigbjørn Lien (NMBU, ELIXIR Norway)

Initial Members to add once launched:

Alexey Sokolov (EMBL-EBI; ELIXIR Hub)

Richard Crooijmans (Wageningen ; ELIXIR Netherlands)

Martijn Derks (Wageningen ; ELIXIR Netherlands)

Marta Godia (Wageningen ; ELIXIR Netherlands)

Thomas Derrien (INRAE; ELIXIR France; TBC)

Scope:

A scientific community to support genotype to phenotype analysis for farmed and companion animal species. Connecting researchers and data scientists across species and countries is needed to accelerate genotype to phenotype research in domestic animals and develop a coordinated open and standardised data life cycle. This ELIXIR community aims at promoting tools, databases, standards, and best practices for domestic animal genomics genotype to phenotype research. More specifically this ELIXIR community will:

- coordinate discussions and explore potential for data/technology solutions for addressing key issues in welfare, behaviour, health, infectious diseases, and preservation of genetic diversity
- develop standards, coordination, workflows, and visualisation for key developing areas of pangenomics, functional genomics, genome editing, phenotyping and biorepositories, which are needed to enhance genotype to phenotype data analyses
- develop FAIR data management standards and ontologies and promote best practices in data coordination and archiving
- collaborate with key EU research infrastructures in animal science, including EuroFAANG RI, INFRAFRONTIER, PIGWEB and AquaExcel, and key global initiatives such as FAANG and AG2PI
- organise tools and data training and knowledge transfer events
- enhance data reuse and the compatibility of combining data for large scale analyses
- enhance usability and interoperability of the EU data infrastructure for animal genomics
- exchange with other ELIXIR communities, such as the plant sciences and biodiversity communities, on overlapping challenges and potential data/technology solutions

Justification:

Large animal species, both terrestrial and aquatic, have been bred for centuries by human societies. Today, the farming of such species provides an important source of nutrition to humans and plays a critical role in feeding a growing global population. Farmed animal genomes also have a role to play in the One Health challenge, because of the risk of zoonotic diseases. Continuing human population growth and a rapidly destabilising climate poses significant insecurity to our global food system. Agriculture and aquaculture need to adapt to these challenges in order to maintain the benefits they provide to human societies, whilst reducing their environmental impact, conserving biodiversity, and flexibly adjusting to changing societal expectations. Adoption of genomics enabled breeding and management and an improved ability to predict an animal's phenotype using genotype information are key adaptations that are required to meet these challenges. Companion animals also have a major role to play in human society, and significant research is ongoing to improve animal health. This focus group will explore data challenges across a range of phylogenetic taxa covering companion animals, agriculture, aquaculture, and additionally other related taxa such as the role of insects as farmed animal pests.

As highlighted at the ELIXIR all hands 2023, this proposal sets out to meet the desire from ELIXIR and the EU research and industry researchers for a large animal genomics and food security focussed ELIXIR Community, filling a long-standing gap in the data coordination

landscape. This focus group has the backing of emerging Research Infrastructures in farmed animal genomics (EuroFAANG) and phenotyping, as well as a large number of researchers and established community from funded Horizon research consortia (AQUA-FAANG, BovReg, HoloRuminant, RUMIGEN, GENE-SWitCH, GeroNIMO), and the wider global FAANG initiative.

2.4 Final version of the scope

This final version included the addition of scope and objectives on phenomes, a broader leadership of proposers and more experts for greater support across the ELIXIR nodes. This was the version submitted to the ELIXIR director for review.

Title: Domestic animal genomes and phenomes

One liner: A scientific community to support genotype and phenotype analysis for farmed and companion animal species.

Proposers:

Peter Harrison (EMBL-EBI; ELIXIR Hub)

Emily Clark (University of Edinburgh; ELIXIR UK)

Martien Groenen (Wageningen University; ELIXIR Netherlands)

Sigbjørn Lien (NMBU; ELIXIR Norway)

Michèle Boichard (INRAE; ELIXIR France)

René Baumont (INRAE; ELIXIR France)

Initial Members to add once launched:

Alexey Sokolov (EMBL-EBI; ELIXIR Hub)

Yogmatee Roochun (EMBL-EBI; ELIXIR Hub)

Richard Crooijmans (Wageningen; ELIXIR Netherlands)

Martijn Derks (Wageningen; ELIXIR Netherlands)

Marta Godia (Wageningen; ELIXIR Netherlands)

Dan Macqueen (University of Edinburgh; ELIXIR UK)

Javier Santoyo-Lopez (University of Edinburgh; ELIXIR UK)

Elisabetta Giuffra (INRAE; ELIXIR France)

Guillaume Devailly (GenPhySE; ELIXIR France)

Thomas Derrien (CNRS; ELIXIR France)

Christophe Hitte (CNRS; ELIXIR France)

Catherine Larzul (INRAE; ELIXIR France)

Cyril Pommier (INRAE; ELIXIR France)

NB. EFFAB and FBN can only be invited once the focus group has launched as they are not ELIXIR member institutes

Scope:

A scientific community to support genotype to phenotype analysis for farmed and companion animal species. Connecting researchers and data scientists across species and countries is needed to accelerate genotype to phenotype research in domestic animals and develop a coordinated open and standardised data life cycle. This ELIXIR community aims at promoting and align tools, databases, standards, and best practices for domestic animal genomics and phenomics research. More specifically this ELIXIR community will:

- coordinate discussions and explore potential for data/technology solutions for addressing key issues in welfare, behaviour, health, infectious diseases, metabolism and nutritional efficiency and preservation of genetic diversity and environment
- develop data standards, coordination, workflows, and visualisation for key developing areas of pangenomics, functional genomics, genome editing, phenotyping and biorepositories, which are needed to enhance data analyses
- develop FAIR data management guidelines, standards and ontologies and promote best practices in data coordination and archiving
- collaborate with key EU research infrastructures in animal science, including EuroFAANG RI, INFRAFRONTIER, PIGWEB, SmartCow (that is now in the AgroServ project) and AquaExcel, and key global initiatives such as FAANG and AG2PI
- organise tools and data training and knowledge transfer events
- enhance data reuse and the compatibility of combining data for large scale analyses
- enhance usability and interoperability of the EU data infrastructures for animal sciences
- exchange with other ELIXIR communities, such as the plant sciences and biodiversity communities, on overlapping challenges and potential data/technology solutions

Justification:

Domestic animal species, both terrestrial and aquatic, have been bred for centuries by human societies for a range of uses, such as workforce, leisure, companionship, and food production. Understanding the pathway from genotype to phenotype is a shared issue across domestic animals, with important requirements for data management and data analysis. For this reason we propose a focus group for Domestic Animal species. Domestic animal genomes can inform biomedical and veterinary models for disease and are important in the One Health challenge, because of the risk of zoonotic diseases. Regarding the current objective of food production, the farming of such species plays a critical role in feeding a growing human population. Farmed animal genomes also have a role to play in the One Health challenge, because of the risk of zoonotic diseases. Farm animals also contribute to closed nutrient cycles. Continuing human population growth and a rapidly destabilising climate poses significant insecurity to our global food system. Agriculture

and aquaculture need to adapt to these challenges in order to maintain the benefits they provide to human societies, whilst reducing their environmental impact, conserving biodiversity, and flexibly adjusting to changing societal expectations. The adaptation of the productive, reproductive and non-productive functions of animals and populations to multiple environmental perturbations (genetic and epigenetic mechanisms, phenotypic plasticity, interactions between functions, etc.) require increased animal phenotyping capacities combining both vertical phenotyping (functional exploration) and horizontal phenotyping of large populations using the most advanced data acquisition technologies (high- throughput automatic devices, precision breeding, biomarkers, proxies, etc.). Adoption of genomics enabled breeding and management and an improved ability to predict an animal's phenotype using genotype information are key adaptations that are required to meet these challenges. Companion animals also have a major role to play in human society, and significant research is ongoing to improve animal health. This focus group will explore data challenges across a range of phylogenetic taxa covering companion animals, agriculture, aquaculture, and additionally other related taxa such as the role of insects as farmed animal pests and feed.

As highlighted at the ELIXIR all hands 2023, this proposal sets out meet the desire from ELIXIR and the EU research and industry researchers for a large animal science and food security focussed ELIXIR Community, filling a long-standing gap in the data coordination landscape. This focus group has the backing of emerging Research Infrastructures in farmed animal genomics (EuroFAANG) and phenotyping (AquaExcel, SmartCow and PIGWEB communities are building a new INFRA-DEV project), as well as a large number of researchers and established community from funded Horizon research consortia (AQUA-FAANG, BovReg, HoloRuminant, RUMIGEN, GENE-SWitCH, GEroNIMO), and the wider global FAANG initiative.

2.5 Submission of the application

The above scope and justification was submitted to the ELIXIR hub senior management and director on 17th November 2023. The ELIXIR Directors decision on the focus group is based upon relevance to ELIXIR in the wider context, having broad scope, uniqueness compared to existing communities and inclusiveness. We are still awaiting a decision from ELIXIR, this process is being delayed as ELIXIR is currently transitioning to a new director, Tim Hubbard. With the extensive pre-discussion with the ELIXIR senior management and the outgoing director Niklas Blomberg, the alignment to the 2024-2028 ELIXIR programme and the extensive positive support we have received from a number of ELIXIR nodes, we are expecting a positive outcome.

3. Next steps

Once the focus group has been approved, we will collectively adjust and expand our final scope and justification to make a landing page on the ELIXIR website (<https://elixir-europe.org/focus-groups>) and an announcement in the ELIXIR weekly brief. A kickoff meeting will be organised during Q1 2024 by the appointed chairs.

We will then focus on expanding the membership to gather all the required expertise to tackle our focus objectives, and establish leaders for work packages to tackle key topics. This will include targeting FAANG, domestic animal genotyping and phenotyping, and plant science communities. We will for example foster connections to key partners such as the nf-core community (<https://nf-co.re/>) that already have strong connections to EuroFAANG and connect to the community workflow development of ELIXIR, that would form a working group within the future community.

The new focus group will be presented as part of the 10 years celebration of FAANG workshop at the Plant and Animal Genome Conference in San Diego in January 2024. The morning of this workshop will host the *Global EuroFAANG animal agriculture G2P networking event* that forms deliverable 8.1, so this is the ideal time and context to ensure broad scientific support from European and global institutes. The group will also be presented at ELIXIR days, National ELIXIR nodes meetings, as part of our discussions with other RIs and at the 2024 EAAP meeting.

The focus group will organise the first EuroFAANG RI and Elixir joint virtual workshop on data standards and information systems, that forms milestone 7. This would focus on the opening data issues for animal genomes and phenomes and the first priorities for the workplan and identifying working groups. The existing blueprint of the EMPHASIS ELIXIR collaboration will be used as a foundation. It is likely that a great effort will be needed for standards on phenotyping and on cellular models, where harmonization and standardization are more recent and challenging than for genomics. The ELIXIR community will align and contribute to the key deliverables and timelines of the EuroFAANG RI project deliverables in WP3, 4, 5 and 6. For example, it will provide a broader view on deliverable 3.4 that will assess data infrastructure gaps and make proposals for required data infrastructure development.

The focus group will aim to build a solid foundation through 2024, developing a regular meeting schedule and strong presence at ELIXIR and animal science events. Clear targets will be established to track progress on the scoped objectives. In parallel to this we will continue the discussions with the Elixir hub management and with the heads of the nodes of representative countries to garner the necessary support and momentum to establish a full ELIXIR funded community.