



EXCELERATE Deliverable 10.3

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WP No.	10	
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1. Executive Summary

Genome assembly and genome annotation constitute areas where significant resources, both manpower and computational resources, are needed to come to the successful conclusion of a project. Experience, both organism-specific but also general knowledge of assembly and annotation, is necessary and this is often slowly built up over the years when working continuously on similar projects. Every project is unique, as all organisms are different and variety of different data types and tools exist. There are no golden standards, and the technology is rapidly advancing with new sequence technologies becoming available at a quick pace.

Experience in genome assembly and annotation exists in Europe but it varies between the ELIXIR Nodes, with some already having built up stable infrastructures and others just starting out. However, the need exists in all countries, not least because new sequencing technologies have made genome assembly and annotation more affordable and more feasible even over this past year. Consequently, one of the major goals of EXCELERATE task 10.3 is to build the capacity in genome assembly and annotation where it currently is insufficiently developed.

We report here on the first preparatory workshop for this task, which was held in Uppsala on 19–20 October 2015. Current genome annotation and assembly capacities at the eleven ELIXIR Nodes represented are summarised and a task roadmap is laid out.

2. Project objectives

With this deliverable, the project has reached or the deliverable has contributed to the following objectives:

No.	Objective	Yes	No
1	Implement a programme of organisational capacity building in newly formed ELIXIR Nodes, including sharing of best practice between partners in accessing EU Structural Funds (ESIF) for operating infrastructure.		x
2	Construct and coordinate ELIXIR-wide 'communities of practice' that support and develop the professionals who deliver advanced data and bioinformatics support and services in ELIXIR Nodes.	x	

3. Delivery and schedule

The delivery is delayed: Yes No

4. Adjustments made

The deliverable is slightly delayed because of extra time needed to collect information from participating nodes.

5. Background information

Background information on this WP as originally indicated in the description of action (DoA) is included here for reference.

Work package number	10	Start date or starting event:	month 1
Work package title	ELIXIR Node Capacity Building and Communities of Practice		
Lead	Jiří Vondrášek (CZ) and Bengt Persson (SE)		
Participant number and person months per participant			
1 – EMBL 6.00, 2 – UOXF 4.00, 5 – UTARTU 20.00, 7 – CNIO 1.00, 9 – CIPF 3.60, 13 – CSIC 2.00, 16 – FCG 2.00, 17 - INESC-ID 10.00, 20 – CSC 4.00, 21 – UiB 4.00, 23 – UiT 4.00, 26 – CNRS 5.00, 31 – LIU 24.00, 32 – UL 30.00, 34 – UOCHB 8.00, 35 – MU 26.60, 37 – VIB 10.00, 39 – BSRCAF 12.00, 40 – HUJ 8.00, 42 – FORTH 6.00			
This WP will address the issue of how to get people in Nodes coming together in capacity building, as detailed in the tasks below. There will be accompanying training needs in this capacity building and those training needs will be addressed in WP11. The training needs are in advanced training of the staff handling data and performing genome annotation and assembly. Other training needs for Use Cases will be in general addressed in WP11, but not specific to every Node. For Node capacity building, advanced training will be needed also in management and know-how on operating Nodes, performed in close collaboration with Task 10.1.			

A Community of practice is a group of people who share a craft or a profession, created to coordinate efforts to solve defined tasks and/or with the goal of gaining knowledge related to their field. ELIXIR is looking to establish such Communities of Practice of bioinformatics experts involved in advanced bioinformatics user support across the Nodes to effectively interact with bioinformatics infrastructure users at interfaces of different research fields. ELIXIR Communities of Practice would be the primary mechanism for ELIXIR to establish domain specific services, for example, forming a community of genome annotators across Nodes to meet the need from national researchers of ready access to genome annotation resources. Other examples could be to meet the needs of Rare Disease or Medical genomics research, agricultural or marine bioinformatics and chemical compounds for biology.

ELIXIR will start to build these Communities of Practice to enable coordination and knowledge exchange in selected areas in tasks 10.2 and 10.3. Task 10.2 is directed to create Good Practices in setting up data Nodes, of importance to create a sustainable and scalable data flow from laboratories to national Nodes and further to European or global databases. Task 10.3 is directed to coordinate and exchange expertise in the field of genome annotation and assembly and to create Good Practice in for this field. In the future, further communities of practice are envisioned, arising from needs identified by the Use Cases (WP6 to 9) and identified through community workshops and surveys (Task 10.4). The creation of a sustainable mechanism for establishment of communities of practice is also addressed in Task 10.4.

Objectives

WP10 is focused on strengthening the ELIXIR infrastructure by supporting coordination of Node activities and increasing the organisational capacities of ELIXIR Nodes. ELIXIR Nodes are at very different levels of maturity, ranging from national infrastructures that have existed for over a decade to newly formed consortia. Activities will focus on spreading the knowledge and bioinformatics best practice that exists within ELIXIR's larger and more established Nodes, with newer or smaller ELIXIR Nodes in less research-intensive areas of the EU. This will help to create a stairway to excellence for partners involved, and support the creation of a true European Research Area. One of the deliverables will be a set of "Good practices" for setting up and running an ELIXIR Node, which will be of substantial value for both current and future Nodes.

Its two Objectives are:

1. Implement a programme of organisational capacity building in newly formed ELIXIR Nodes, including sharing of best practice between partners in accessing EU Structural Funds (ESIF) for operating infrastructure.
2. Construct and coordinate ELIXIR-wide 'communities of practice' that support and develop the professionals who deliver advanced data and bioinformatics support and services in ELIXIR Nodes.

Description of work and role of partners

Task 10.1: ELIXIR Node Capacity Building (46PM)

This task will support the formation of an ELIXIR community. There are significant differences between existing ELIXIR Nodes in their capacity, level of expertise and maturity of services/tools/data. We will increase the joint competence and capacity for Nodes lacking a large national user community, large-scale projects and big data or having a limited record of offered tools and services. These Nodes will benefit from mutual collaboration and connection with well-established and more advanced Nodes they can utilize their know-how for a more rapid Node development. Altogether, this will help shape ELIXIR as an efficient pan-European infrastructure.

The major aim of this task is to provide management knowledge transfer among Nodes to create a set of well-balanced, well-functioning and compatible Nodes.

Support in coordinating national Nodes, including Skills and Knowledge exchange between ELIXIR Nodes. Nodes with different experiences will help to provide knowledge regarding good

practice in different situations and providing direct support to implementation of national infrastructures (e.g. by national / regional workshops with external experts, support to national community building efforts). The heterogeneity of Nodes established will help providing multiple effective ways for coordination and to get funds from national providers and their commitments to the infrastructure. Knowledge exchange will be catalysed by workshops, staff exchange programme and visits. This activity is based on the ELIXIR community practice experience but it is more general and should cover some features brought by larger staff community.

Identify and apply technical solutions at/between Nodes. The reason for particular technical solution must be explicitly formulated and the solution must be applicable on more than 2 Nodes. The capacity building deliverables would be primarily workshops based on Technical Services and/or Training WP deliverables.

Partners: CH, CZ, EE, NO, PT, SE, SI, UK, ES, EL, IL, EMBL-EBI

Task 10.2: Capacity Building in Data Nodes Network (34PM)

One of the aims of ELIXIR is to establish a network of data Nodes (Nodes with large data collections and databases with established way of data deposition and curation) to enable scalable data storage and their transferability by means of standardised formats. In this task, we will focus on establishing guidelines and good practices to facilitate efficient data collection into core data resources (cf. WP3), primarily focusing on data needed for selected Use Cases (WP6 to 9). This is tightly linked with IT solution by means of storage, dedicated networks and connections (cf. WP4). A distributed network following the same standards will also simplify international sharing of datasets for which this is ethically permitted.

This task both includes creation of routes for data publishing in a uniform manner across ELIXIR with data Nodes in each country and includes data repositories for replication of reference data allowing for fast access. The setting up of a data Nodes network has been identified by the technical experts within ELIXIR as a prioritised area.

Task 10.2 also includes development of Good Practices in setting up data Nodes enabling secure storage of sensitive data, such as sequence data related to patients. The task is interfacing with WP4 regarding technical developments on AAI and data transfer. Furthermore, there are connections with WP4 on data interoperability and the Use Case in WP9 on sensitive data.

Partners: SE, FI, CZ, EMBL-EBI, SI, PT, ES, EE. In due time, all ELIXIR Nodes are expected to have an ELIXIR data Node.

Task 10.3 – Capacity Building in Genome Assembly and Annotation (44PM)

Specialised expert platforms for genome assembly and annotation are already available in several ELIXIR countries. They provide critical support to complex genome projects and deliver annotations that serve as the basis for scientific inquiry into the genomics of newly sequenced organisms. The specialised expertise at multiple ELIXIR Nodes would benefit from capacity building through competence-spreading advanced workshops and staff exchange.

The capacity-building efforts will benefit the Use Cases in WP6 on marine organisms and in WP8 on plant Use Cases. The genome annotation groups will contribute with domain-specific knowledge about different species, e.g. marine organisms (SE, NO), woody plants (PT) and crop plants (SI).

Furthermore, in order to facilitate access to genome annotation to the users, we propose a deployment of web services to enable genome projects in the scientific community to efficiently interact with the data. The development of such web services is intended together with the EnSEMBL team to create a pan-European collaboration on genomics resources to provide researchers with a unified analysis platform carried by multiple partners.

Partners: SE, NO, FR, PT, EBI, SI, BE, CZ, ES.

Task 10.4 – Sustainability of capacity building (30PM)

The main goal of Task 10.4 is periodical and long-term discovery of users with specific capacity needs at ELIXIR Nodes and/or research groups within Nodes. This knowledge of capacity needs/gaps will be gathered through surveys and face-to-face meetings. With capacity needs identified the Task 10.4 team will connect users with WP11 groups that have at their disposal training infrastructure, learning materials and knowledge needed to implement the capacity building. In order to ensure the sustainable flow of knowledge and stable capacity maintenance we need to provide long-term networking of capacity seekers and providers. They will be focused to the great extent to the Good Practices from Task 10.2 and 10.3 (and WP6 to 9). With well-formed ELIXIR Communities of practice, the Task 6.4 will be able to lead the reuse or even suggest the adaptation of WP11 courses and training materials for specific capacity building needs.

It is of great importance that capacity needs will be periodically (but in long-term perspective) tested through surveys, which will also contribute to the sustainability of training infrastructure and learning materials provided by WP11. Task 10.4 will monitor the implementation of capacity building in Tasks 10.1, 10.2 and 10.3 in order to extract good practices and compile good practice

recommendations and guidelines which can be used in other capacity building contexts.

Partners: SE, SI, CZ, BE, EE, EL, IL, EMBL-EBI

Task 10.5: Supporting ELIXIR Nodes in understanding Smart Specialisation Strategies and accessing EU Structural and Investment Funds (ESIF) (36.2PM)

The potential for exploiting funding synergies between EU Research programmes and ESIF are well known⁶². Those ELIXIR Nodes eligible for ESIF are therefore presented with a real opportunity for local funding of their Node, particularly in light of the proposed focus on ESIF and ESFRI that many Member States are making within their national plans to the Juncker Investment Plan. However, understanding the local priorities for funding, rules, and application procedures presents is complex and time consuming and securing ESIF for operational costs of life science infrastructures is a real challenge. For ELIXIR Nodes to access ESIF in any meaningful way, support needs to be targeted at the local level, allowing scientists to build up an understanding of their local Smart Specialisation Strategy, which dictates the funding opportunities for that region, and then develop a strong business case that can be used for subsequent funding applications.

Partners: CZ, SI, EE, EL

ELIXIR ESIF Task Force (Months 1-12)

ELIXIR Structural Funds Task Force grouping funding specialists across ELIXIR Nodes will be established to share best practice in ESIF use for research infrastructures. The Task Force would also engage external experts such as ones from national managing authorities for ESIF, DG REGIO, DG EMPLOY, DG Enterprise and Industry and Jaspers and would make use of existing reports such as the ESPON KIT report (www.espon.eu).

An ELIXIR-wide Workshop early at start of the project to pool good practice on using Structural Funds to support research infrastructures and facilitate personal interactions. Meeting will be hosted and organised by CEITEC, who leads this task.

This would include talks from ELIXIR Nodes with experience of accessing Structural Funds (Estonia, Czech Rep, Slovenia), as well as other ESFRIs such as ELI that have done this successfully in other disciplines

Local priorities and their overlaps identification towards Business Case (Months 6-24)

As all regional priorities are different, and as the application process for funding is done in the

local language and following local rules, target Nodes will work with their regional partners to understand the priorities. This task will support Nodes in understanding their local Smart Specialisation Strategy and the regional priorities relating to research and life sciences. Access support from Jaspers following the connections built up within Months 1-12.

Supporting Nodes in actually developing the Business Cases and applications for Structural funds to support the construction and/or operation of the Node. The timing of this work will depend on when the calls will be opened for each region.

Partners: CZ, SI, EE, EL

Appendix 1: REPORT Preparatory workshop for ELIXIR-EXCELERATE Task 10.3

REPORT

Preparatory workshop for ELIXIR- EXCELERATE Task 10.3 Capacity Building in Genome Assembly and Annotation, including early phase activities

Summary

This preparatory workshop for EXCELERATE Task 10.3, at which eleven ELIXIR Nodes were represented, was held in Uppsala on Oct 19–20 2015. The level of experience in assembly and annotation varies between the Nodes, with some already having built up stable infrastructures and others just starting out. The workshop was divided into two parts, with the first being dedicated to presentations by the participating Nodes about their interests, competences and needs in genome assembly and genome annotation. This was followed by a discussion around a number of defined topics to further outline and detail the project work in task 10.3, and a closing meeting where tasks to be performed were defined.

Topics discussed included:

- What areas should we focus on in the task 10.3?
- Is there any interest in a helpdesk-activity where for example Nodes with organism-specific experiences help other Nodes with similar projects?
- How should we communicate within the task?
- What can the different Nodes contribute with?
- Do we see a need for any specific web-services?
- How do we best help inexperienced Nodes set up assembly- or annotation-structures of their own?

Some areas were chosen as prioritized and the following tasks were defined:

- Create a wiki page of expertise present in the different Nodes.
- Write best practices documents with minimum requirements of what an assembly- or annotation project should include.
- Enable Nodes to develop expertise in annotation and assembly by immersive training.
- Create definitions of terms, possibly as a wiki
- Investigate ways of communicating within the task

Meetings planned for 2016

For continuation of the task 10.3, two meetings in 2016 are scheduled. These meetings, which will be held in connection with major community events, the progress of each task will be reported and further tasks will be added.

1. ISCB meeting in Barcelona Apr 4–6 2016
2. ECCB meeting in The Hague, Sep 3–7 2016

Summaries of presentations

Belgium: The Node is based in Gent and focuses on annotation of plants, but also works with marine algae, fungi and animals. Has developed an annotation pipeline and a tool for community annotation - OrcAE. The Node sees a need for functional annotation tools and better tools for assembly of PacBio and Nanopore data.

Czech republic: The Czech consortium has 14 members. Has expertise in annotation of repeated elements at the LAMC center. The Node is interested in setting up annotation services.

EMBL-EBI: Wants to enable genomic science. Focuses on annotation of vertebrates, but also annotates other taxonomic groups in Ensembl Genomes.

France: The Node is distributed over 6 regional centers. There are in France several separate annotation services such as Microscope, GNPannot, and the URGI platform. Focus is on plants, bacteria, and viruses, and also metagenomic pipelines.

Italy: Currently an observer Node, with several universities involved. The Milano group focuses on genome assembly with interest in library QC, preprocessing of data, coverage biases, effects of amplification, estimation of repeat numbers and scaffolding.

Norway: The Node is distributed over several cities, with main Node in Bergen. Focus is on marine metagenomics; a pipeline for metagenomic annotation called Meta-pipe has been developed. Can contribute with pipelines for analysis of metagenomic data, marine specific data resources, expertise in metagenomic analysis and annotation standards, and courses.

Portugal: Several different institutions constitute the Portugal Node. There are several plant-based projects in the country, including assembly and annotation of the cork oak genome, but also projects on pine and eucalyptus. The Node is interested in collaborating with the work packages in marine organisms and plants (WP 6 and 7, resp.).

Slovenia: The Node has 15 partners in 11 legal entities. They are interested in capacity building and training in genome-assembly and -annotation and can provide use cases for plants, rare diseases and marine organisms. They also have expertise in health-oriented ontologies.

Sweden: The Node has 45 experts distributed over 6 cities including a team of 5 bioinformaticians supporting Swedish research projects with assembly and annotation expertise. The team works with all types of organisms and all types of data, can contribute with technical expertise and teaching.

Switzerland: SIB, the Swiss Institute of Bioinformatics constitute the Swiss Node. Their mission is to provide world-class bioinformatics to the life science community.

They have extensive experience in proteome annotation, for example using the HAMAP tool, and see a need for more consistent annotation.

United Kingdom: The Node focuses on training. Has experience with vertebrates and plants and can contribute with technical expertise, tools development, training, and best practices. The UK community wants to have access to more compute power and more storage, wants help to adopt new technologies, and is interested in developing standards to exchange and integrate information.

Roadmap

Genome assembly and genome annotation constitute areas where significant resources, both manpower and computational resources, are needed to come to the successful conclusion of a project. Experience, both organism-specific but also general knowledge of assembly and annotation, is necessary and this is often slowly built up over the years when working continuously on similar projects. Every project is unique, as all organisms are different and variety of different data types and tools exist. There are no golden standards, and the technology is rapidly advancing with new sequence technologies becoming available at a quick pace.

This hard-bought expertise exists in Europe, but it is unevenly distributed with some countries having built up considerable expertise in the area, while others have not started. The need exists in all countries though, not the least because new sequence technologies have made assembly and annotation more affordable and more feasible only in the last year. This need was correctly identified in the ELIXIR-EXCELERATE proposal and one of the major goals of task 10.3 is to build the capacity in genome assembly and annotation where it currently is insufficiently developed.

Current activities

We are currently in a build-up phase where structures need to put into place to ensure easy communication and information needs to be collected to build later initiatives on. The kick-off meeting in Uppsala in October identified some key areas to start focusing on.

Existing expertise

A list has been compiled of existing genome assembly and genome annotation expertise for the Nodes working in the task. Although already informative, some details are missing and the list will be continuously updated as more Nodes are joining. A condensed list is presented here, representing the status of January 2016.

Belgium - Experts on annotation of plants, but also annotation of algae, fungi, and animals. Own annotation pipeline developed as well as online tool for cooperative curation of annotated genomes.

Czech Republic - Extensive work on annotation of repetitive elements. Also phenogenomics, i.e., the use of mouse and rat models to infer functional annotation of orthologous human genes.

EMBL-EBI - Hosts Ensembl, the largest annotation resource in Europe. Focus on the annotation of mammals, but also extensive annotation of other organisms in Ensembl genomes and Ensembl bacteria. Supplies powerful online interfaces for data mining and functional genomics of hosted organisms.

France - Long experience of diverse assembly- and annotation-projects, including plants, fungi, bacteria, viruses, and marine algae, as well as environmental samples. Has several online services available in particular for annotation.

Norway - A focus on marine metagenomics. Maintains meta-pipe, an online tool for the annotation and analysis of metagenomic data.

Portugal - Genomics marine organisms and forest trees, of which the cork oak genome is a flagship project.

Slovenia - Focus on annotation of genetic diseases.

Sweden - Has a dedicated support team for assembly and annotation of any organism. Extensive experience of annotation of eukaryotes, in particular birds, butterflies, and fungi

List of terms

To enable communication, a list of definitions of common terms used in assembly and annotation is being compiled. The first version will be ready by April 2016.

Communication

Ways of efficient communication within the task such as a discussion forum and chat services is being investigated and will be reported on at the WP10.3 April 2016 meeting.

Timeline

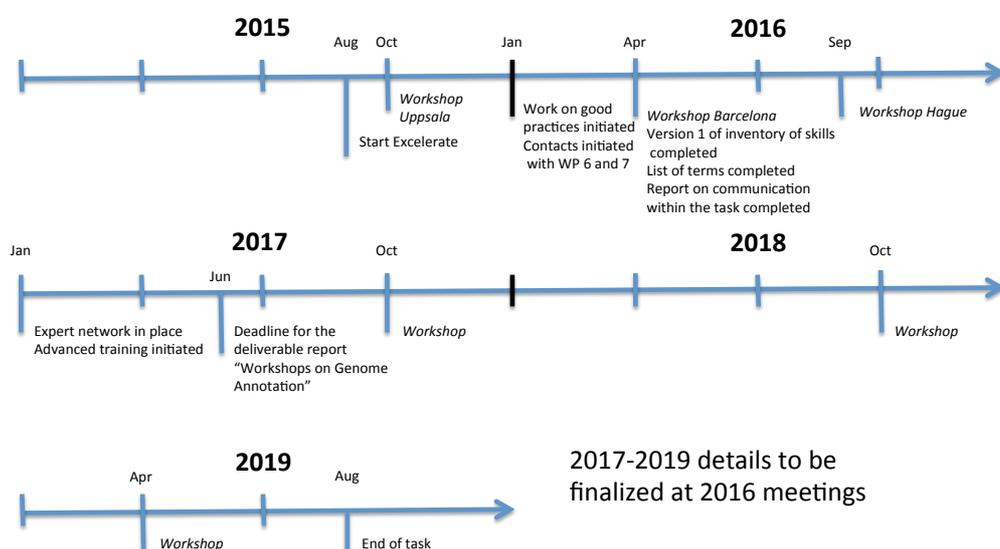


Figure 1: Timeline for the ELIXIR-EXCELERATE Genome Annotation Capacity Building Programme

Collaborations with other work packages

This task clearly shares areas of interest with other work packages, and this was also identified already in the task description. Work packages 6 (Use case A: Marine metagenomics) and 7 (Use case B: Crop and forest plants) will benefit from the assembly and annotation expertise present in this task. Annotation of genes is a key component of metagenomics analyses and the genotypic component of the plant use case is also heavily dependent on annotation. Both of these use cases also deal with standardization of terms, including gene names, which is one of the major results of the gene annotation work focused on in this task. There is also currently a discussion on comparability of annotations, which would benefit the standardization issues encountered in both use cases. During 2016Q1, we will investigate the needs of the WP6 and WP7 regarding annotation issues.

Advanced training

The need for assembly and annotation expertise in Europe at the moment greatly exceeds the available expertise. A key component of this task is therefore to spread the knowledge from the Nodes where developed assembly and annotation infrastructures exist, to Nodes where this knowledge is still lacking. We want to encourage and enable the growth of these services by high quality training activities, and two activities have been chosen as most important. We plan to have the first activities in 2017 and then run these activities regularly.

Courses. Well-developed courses in genome assembly and annotation already exist in several Nodes. These can be adapted and used in the developing Node's countries or at the facilities of the expert Nodes. There is also the possibility to make organism- or more subject-specific courses and run these in collaboration with the use cases A, B, and C (marine metagenomics, forest and crop plants, and rare diseases). Efforts will be coordinated with the training work package (WP 11).

Immersive training. It was decided at the Uppsala kick-off that the developed Nodes would accept bioinformaticians from other Nodes for immersive training. This would take the shape of shorter internships, perhaps one week at the time, where the training bioinformatician would get a possibility to join in running assembly- or annotation-projects to learn the necessary tools and workflows. There will also be a possibility to bring own data to these occasions to get access to expert advice while working on the data.

Network of experts

With currently investigated communication tools in place, efficient exchange of technical expertise will be easy. Together with the list of existing expertise, this will enable experts to get in touch with each other to help and to increase the overall expertise of genome assembly and annotation in Europe. This expert network is planned to be in place early 2017.

Good practices. A "best practices" document was much requested at the Uppsala workshop. This will be used as a guide by Nodes interested in setting up assembly or annotation services of their own. A "good practices" document is also mentioned in the ELIXIR-EXCELERATE proposal as an important deliverable from this task.

The good practices document will include, but not necessarily be limited to:

- A definition of minimum requirements for assembly and annotation projects

- An outline of necessary computational resources
- Important biological and technical considerations to remember when going into an assembly or annotation project
- Minimum human resources necessary to run a successful assembly and/or annotation service

Future possibilities

Project planning. Assembly and annotation projects are long and time and consuming with multiple steps involved and it is of great importance that the projects are planned correctly already from the start before ordering data. To offer the possibility of expert advice in the planning stage to starting projects would cost little resources but be of great benefit for the new projects. A cross-Europe assistance between ELIXIR Nodes would help in this capacity building. This will be further investigated at upcoming meetings in April and September.

Web services. Co-ordinated web services in this area would be beneficial for the assembly and annotation platforms across the ELIXIR Nodes. We will outline how to best coordinate these at the upcoming meeting in April 2016.

List of participants

Belgium

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