

MANAGEMENT AND IMPACT MAIN ACHIEVEMENTS AND CHALLENGES



Main Achievements: Management

2

- Preparation of 3rd annual meeting / progress meetings
- Preparation and submission of 3rd Amendment (**Approved 26 Feb 2016**)
- Coordination of monthly Executive Management Committee teleconference updates
- Coordination and submission of deliverables and milestones for all partners
- Preparation of WP1 deliverables, milestones and reports
- General guidance and *ad-hoc* support to partners
- Fostering collaborations with NeurOmics and EUREnOmics- Joint meetings / tasks / committees and coordinator calls
- International initiatives – IRDiRC and Global Alliance
- Fostering collaborations with new collaborators / contributors



Achievements: Deliverables

3

- **All Deliverables due submitted between M25 and M36 (Total 33)**
 - WP1 (2) - D1.04, D1.11
 - WP2 (6) - D2.09, D2.10, D2.11, D2.12, D2.13, D2.14
 - WP3 (0) – No deliverables during period 3
 - WP4 (4) - D4.08, D4.09, D4.10, D4.11
 - WP5 (7) - D5.14, D5.15, D5.16, D5.17, D5.18, D5.19, D5.20
 - WP6 (5) - D6.10, D6.11, D6.12, D6.14, D6.15
 - WP7 (8) - D7.19, D7.20, D7.21, D7.22, D7.23, D7.24, D7.25, D7.27
 - WP8 (1) - D8.02

- **Ongoing - To be ready for M42 - 30th April 2016 (Total 3)**
 - D1.05 - Fourth progress and management report (FINOVATIS - M42)
 - D6.16 - Report on autonomy and solidarity (LUMC - M42)
 - D6.17 - Report on a framework for commercialisation and academic-industry interaction (LUMC - M42)



Achievements: Milestones

4

- **Milestones achieved between M25 and M36**
 - MS3 - Online registry for “undiagnosed” cases (ISS)
 - MS6 - Online searchable biobank catalogue launched (KI)
 - MS9 - Tools for molecular and clinical phenotype associations (MURDOCH)
 - MS10 - Guidelines for best practices in semantic enrichment and semantic data integration (LUMC)
 - MS20 - Training material development (PCB)
 - MS27 - Ethical platform for the platform endorsed (UU)

- **Delayed / Amended Milestones - None**

- **Ongoing - to be ready for M42**
 - MS31 - Midterm impact meeting WP7 (UNEW - M40) – Sustainability meeting report



Achievements: Amendment 3

5

- Submitted to the EC on the 27th October 2015
- Approved on the **26th February 2016**

- Additional Beneficiaries
 - BBMRI-ERIC (Partner 28)
 - UMCG (Partner 29)

- Partial transfer of rights and obligations
 - PCB (Partner 2) -> CRG (Partner 30)





Achievements: Reporting activities

6

RD-Connect annual reports are contractual and part of the deliverables list

- **Period 3 (1st Nov 2014 – 31st Oct 2015)**
 - **Third internal (M30) report**
 - Medium level reporting per WP and per institution (no payment information required)
 - **Third annual reporting (M36) submitted on 31st December 2015**
 - Reporting per task, coordination *via* WP leaders
 - Thorough assessment of outputs
 - Assessment of ALL deliverables and milestones
 - Triggers payment (Period 3 Form C)

- **Period 4 (1st Nov 2015 – 31st Oct 2016) – This period**
 - **Fourth internal (M42) - 31st April 2016**
 - **Fourth annual report (M48) - November 2016 (+60days)**



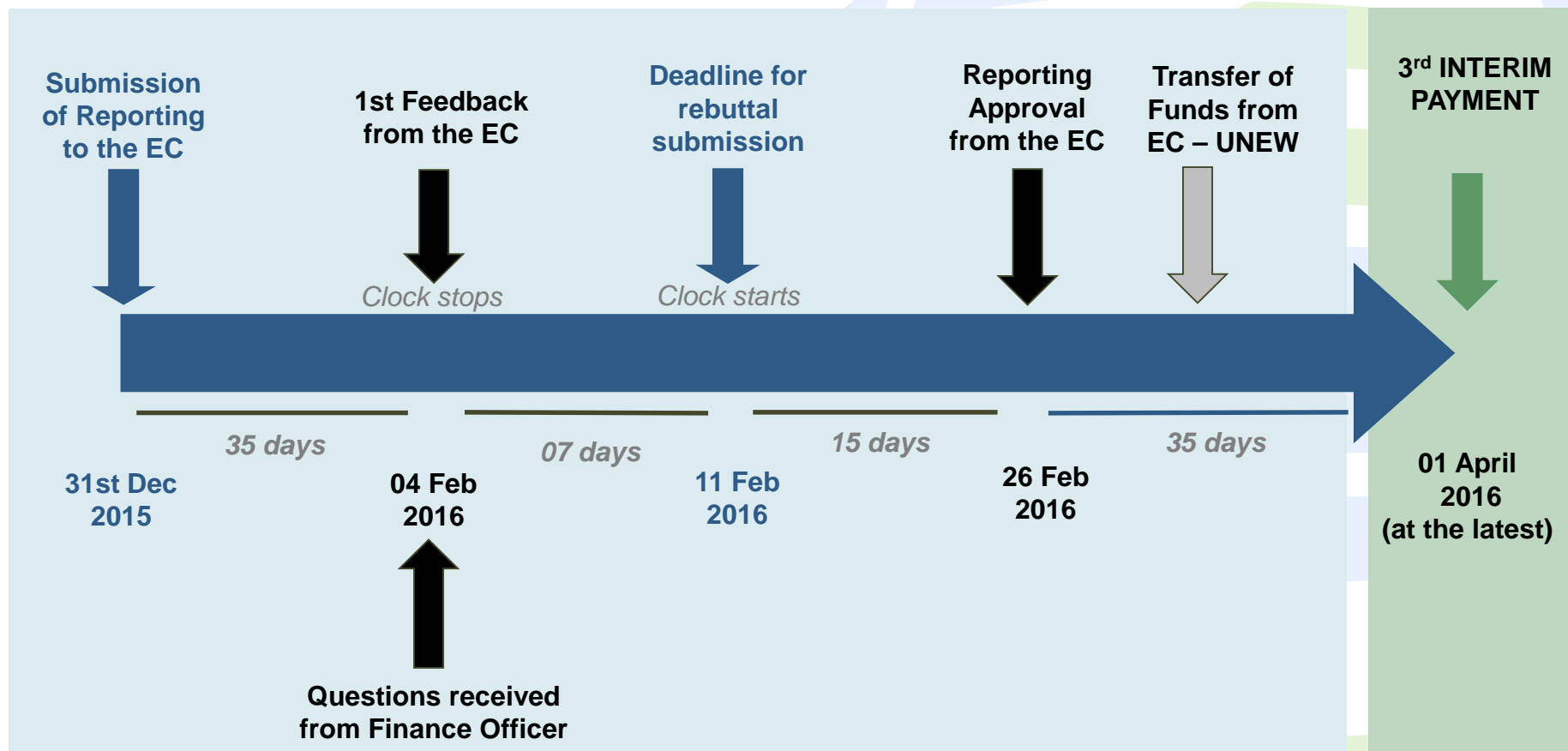
Challenges: Lessons learnt from 3rd PAR

7

- **Periodic Activity Report**
 - WP and task leaders responsible for completing the PAR- some concerns that **not all sections were updated fully**
 - The M30 interim reports helped somewhat with completing the annual reports and we will continue this practice in future reporting periods
- **Deliverables**
 - We encouraged partners to **submit M36 deliverables early** so as not to 'clash' with the annual reporting timelines – this worked in some cases, not in others. Will repeat.
- **Financial reporting / Form C**
 - We will **not** request interim financial reports for period 4
 - We will initiate the contractual financial reporting in period 4 as soon as possible (1st Nov 2016) so that **ALL** Forms C are signed electronically and submitted to the Coordinator before the Christmas holidays – this worked well this year and all Form C's were submitted on time
 - Direct interactions with partners financial departments helped the process
 - The coordination team will make contact with financial departments in October 2016 to inform them about reporting and deadlines to come



RD-Connect Payment timeline - Period 3





Financial status

9

PM	ACTIVITIES	TOTAL	RTD	Management	OTHER
307	PERSONNEL COSTS	1,433,064.10 €	89%	5%	6%
	SUBCONTRACTING	7,557.62 €	62%	38%	0%
	OTHER DIRECT COSTS	199,857.24 €	88%	0%	12%
	INDIRECT COSTS	943,765.14 €			
	TOTAL COSTS	2,584,244.10 €			

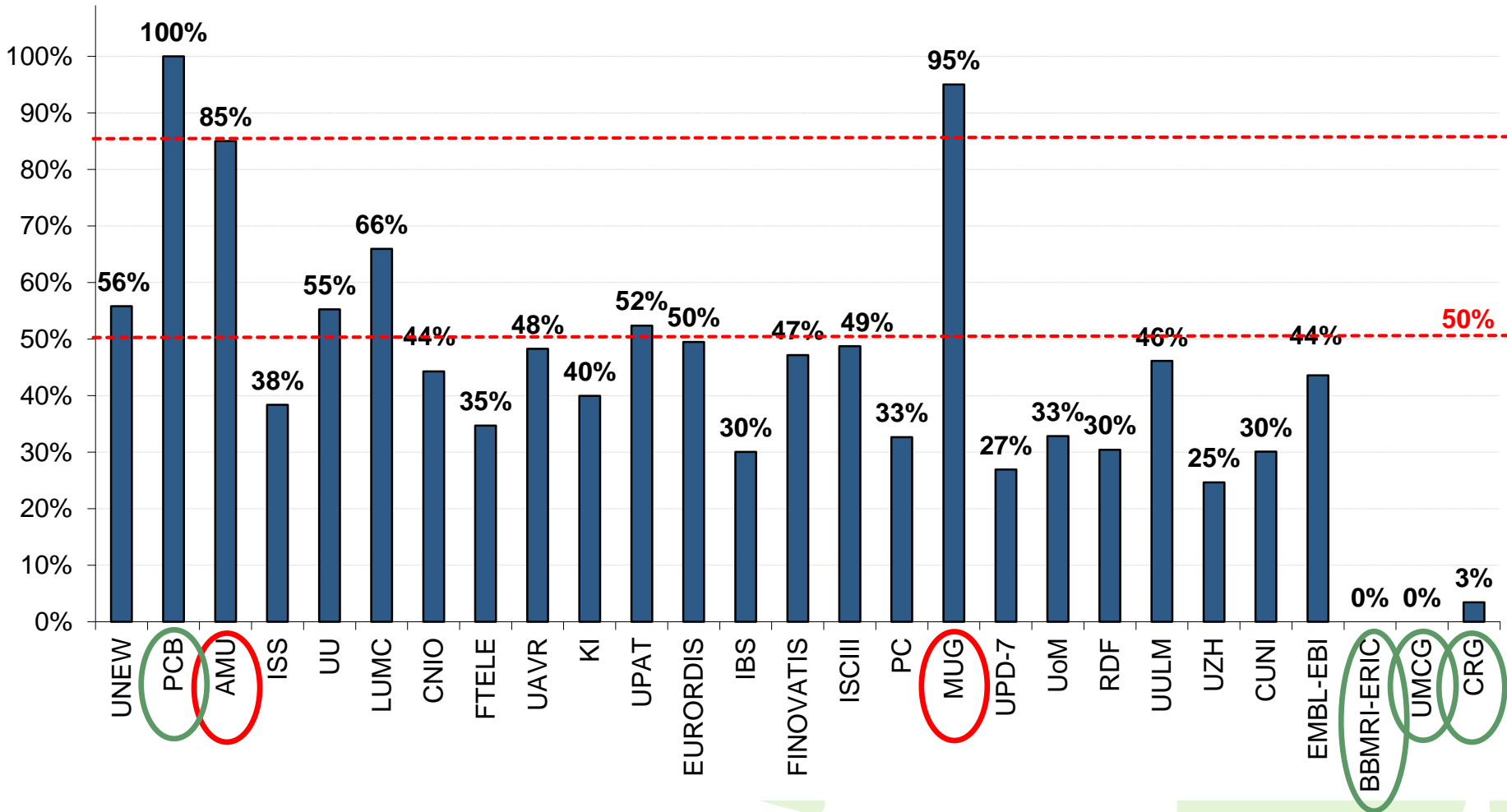
TOTAL REQUESTED EC CONTRIBUTION	2,008,613.83 €
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- Total requested EC contribution: **2,008,613.83 €** i.e. **17%** of total EC contribution for Period 3
- Corresponds to ~ 1/6 of Total EC Contribution for full duration of project, should a linear use of budget be considered over the course of the project.



% of EC accepted costs (M1-M36)

10



Y1 + Y2 + Y3 = 47% total EC contribution spent



Challenges: 85% project budget

11

- Partners under 85% spend should expect to receive funds up to the amount approved in Period 3 Form C's
- Partners 'spending' 85% or over will receive a reduced payment in order to respect the limit
- New partners propose to distribute 63% to BBMRI-ERIC and UMCG as pre-financing
- CRG received budget directly from PCB as pre-financing



RD-Connect Mid-Term Review

12

- **Project officer Dr. Iiro Eerola confirmed that the Mid-Term review would be carried out by an external reviewer (Prof. Erwin Böttinger CEO of the Berlin Institute of Health) and would be based on:**
 - **All submitted deliverables (up to M36)**
 - **Period 3 Activity Report**

- **No face to face meeting in Brussels**





Next joint project meeting - 2017

13

- Meeting to be held jointly with NeurOmics and EUREnOmics (as their final project meeting)
- Proposed location: Berlin, Germany
- Suggested dates:
 - 24-28 April 2017
 - 2-5 May 2017
 - 8-12 May 2017





Main Achievements: Impact

14

- Further developed strong communication channels (Website, Newsletter and Twitter) for disseminating the outputs from RD-Connect.
- Over 70 peer-reviewed publications acknowledging or referencing RD-Connect have been published including a review on RD-Connect (44 in Period 3) .
- Additional stakeholders beyond NeurOmics and EUREnOmics, including industry, new data suppliers and other projects have expressed interest in contributing data to the RD-Connect platform.
- The *international charter of principles for sharing bio-specimens and data* which provides guidance for effective legally and ethically grounded sharing has been awarded the IRDiRC recommended label in August 2015.
- Establishment of the Unique identifier Task Force (RD-C, GA4GH and IRDiRC)
- The joint communication group between NeurOmics, EUREnOmics and RD-Connect has met over the past 12-months.
- Interaction with EU priority areas remains strong.
- Facilitate training events for partners and collaborators.



Website updates

15

- Website was updated with content from M24 / M30 deliverables.
- WP leaders / partners involved in the updating as appropriate.
- Most updates coincided with monthly newsletters which focused on specific topics or themes.
- Biobank and Registry sections of the site have been updated to include information on and links to resources for the RD-Connect online catalogue / ID-Cards.
- Interface to the central RD-Connect platform has been integrated with the website to create a comprehensive and user-friendly online portal that combines the data and tools interface with an extensive online communication and information site.
- Portal provides access to the Genomics Platform itself as well as various associated tools and resources such as Phenotips, the ID-Card system and links to the Biosample Catalogue.
- New section added with information about the collaborating and contributing projects to RD-Connect.
- Publication and Outreach section updated.



Dissemination activities

16

	Year 1 (Jan 2013 – Dec 2013)	Year 2 (Jan 2014 – 31 Oct 2014)	Year 3 (1 Nov 2014 – 31 Oct 2015)
Sessions	10,832	13,303	19,462
Unique users	7,294	9,746	14,662
Page views	27,807	29,693	40,787

- Website** users increased year on year - 14,662 users
- Twitter (@ConnectRD)** has a total of 809 followers and has made a total of 771 tweets
- Newsletter** - 18 issues have been published. At M36 we have 605 subscribers
- 44 **peer reviewed publications** referencing or produced as a direct result of RD-Connect in period 3.





Dissemination activities

17

- 200 individual **events / activities** which have involved RD-Connect over Period 3 (31 Oct 2014 – 1 Nov 2015):
 - Presentations to a scientific event: 99
 - Posters: 43
 - Workshops: 22
 - Films, interviews, TV-clips and videos: 9
 - Presentation to a wider public: 6
 - Conference: 5
 - Exhibition: 4
 - Websites/applications: 4
 - Press release: 4
 - Articles published in the popular press: 3
 - Flyers: 1



IRDiRC and Global Alliance

18

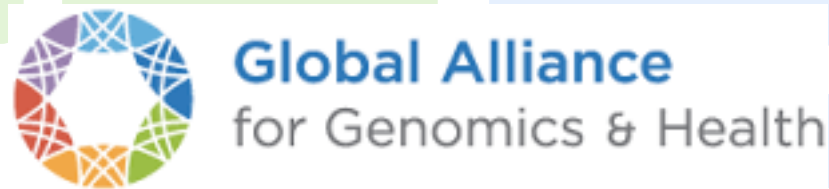
□ IRDiRC

- Committees and Exec
- Task Forces
- IRDiRC Recommended
 - *The international charter of principles for sharing bio-specimens and data which provides guidance for effective legally and ethically grounded sharing – August 2015.*
 - *Potential future applications include ID-Cards and the RD-Connect platform*



□ Global Alliance

- Matchmaker Exchange
- GA4GH Beacon
- Joint session at the 13th Mutation Detection meeting in Leiden
- Unique Identifier Task Force (RD-Connect – IRDiRC – GA4GH)





ESFRI Infrastructures

ESFRI

19

- Refocused interactions with ESFRI infrastructures (BBMRI-ERIC, ECRIN, EATRIS and ELIXIR, EU-OPENSOURCE and INFRAFRONTIER) to
 - Concentrate work around funded projects such as **EXCELERATE (ELIXIR)**, **ADOPT-BBMRI-ERIC (BBMRI)** and **CORBEL (multiple RIs)** (EU Horizon 2020). Participation of RD-Connect partners, dedicated RD work streams led by RD-Connect partners = extensive interaction in areas relating to data interoperability and identifiers. .
 - Identifying opportunities for funding for projects not currently funded.
- Collaborations already in place
 - BBMRI-ERIC - full partner / sustainability / BBMRI-UK leadership
 - ELIXIR - BYOD workshops / Linked data approaches
- Additional Use cases – presented at ESFRI infrastructure meeting in Lux 18th Sept 2015, Hinxton, Elixir interoperability meetings (Amsterdam and Barcelona)

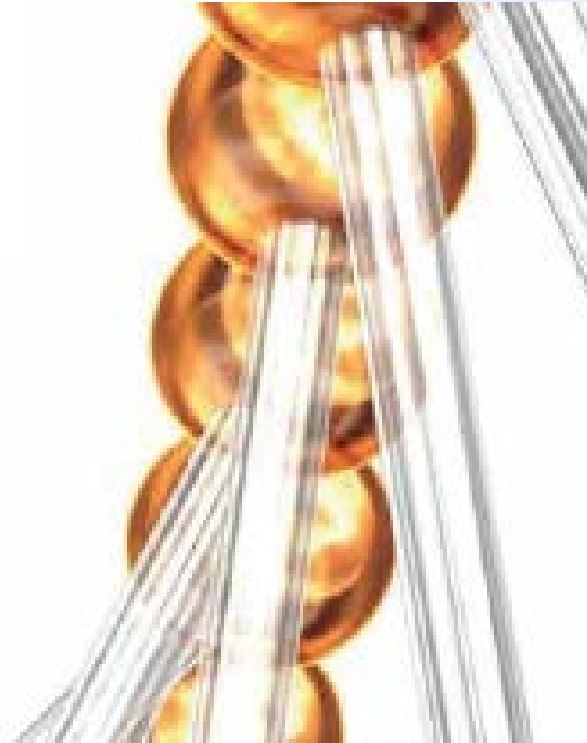


Mediterranean Collaborations

20



Genomics of
Neurodegenerative
Disorders



14-16
J a n
2016

University of Mansoura,
Mansoura, EGYPT



Challenges: Impact

21

- Staffing –Dorota Badowska (UNEW) and Virginie Bros-Facer (EURORDIS)
- Engagement of ALL work packages
- Impact working group (UNEW, RDF, AMU and CNAG...others?)
- Engagement with EUREnOmicS
- Sustainability





EURen  Omics

Neur  Omics

Thank you!

RD  Connect

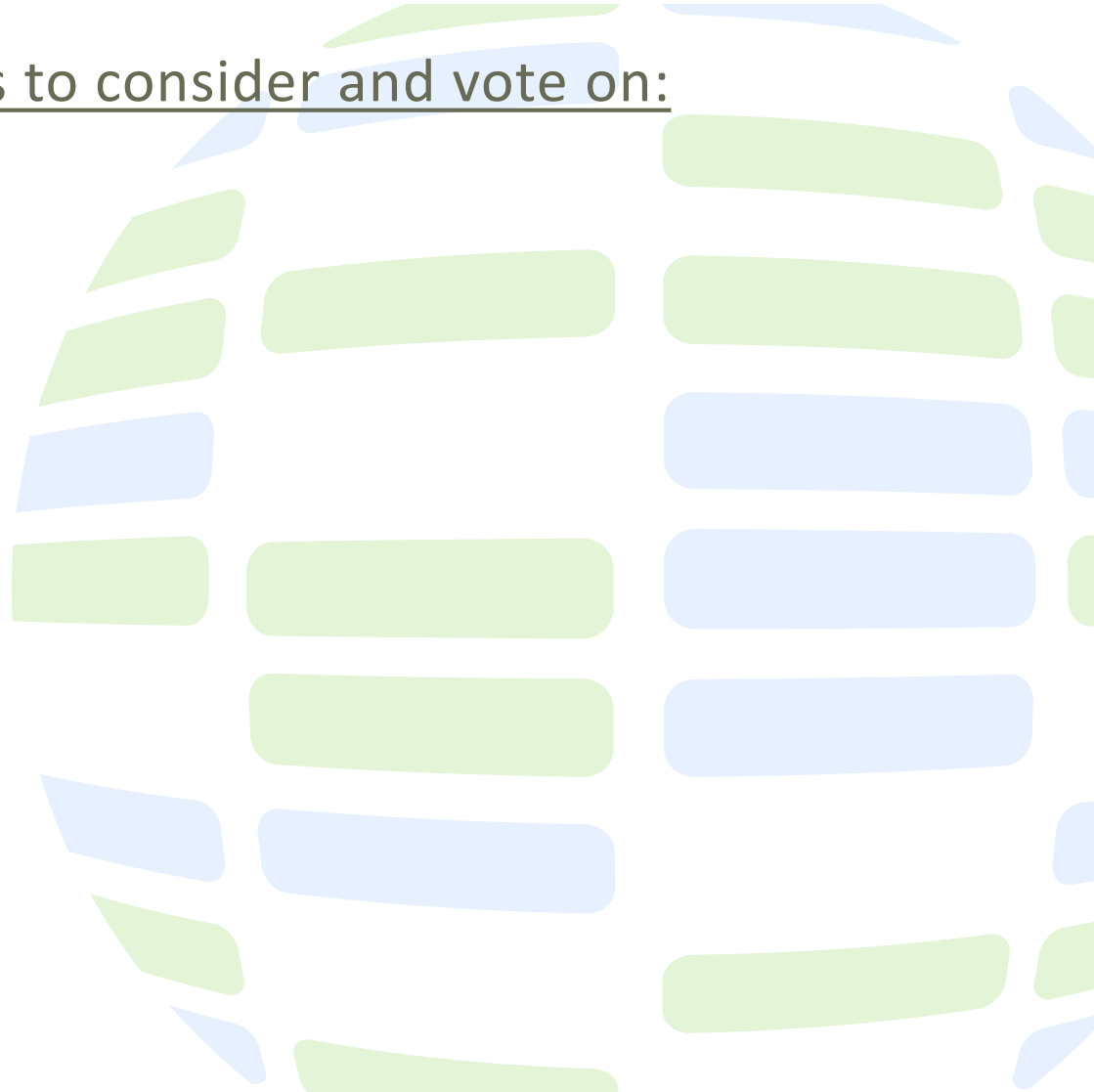


Governing Board

23

Governing board needs to consider and vote on:

- XX



Governing Board Members

Institution	GB member	Representative or Default
University of Newcastle upon Tyne (UNEW)	Prof Hanns Lochmüller	
Centro Nacional de Analisis Genómico (CNAG)	Dr Ivo Gut	
Aix-Marseille University Medical School (AMU)	Prof Christophe Béroud	
Istituto Superiore di Sanità (ISS)	Dr Domenica Taruscio	
Uppsala University (UU)	Dr Mats G. Hansson	
Leiden University Medical Center (LUMC)	Dr Peter-Bram 't Hoen	
Centro Nacional de Investigaciones Oncológicas (CNIO)	Dr Alfonso Valencia	
Fondazione Telethon (FTELE)	Dr Lucia Monaco	
University of Aveiro (UAVR)	Prof Jose Luis Oliveira	
Karolinska Institutet (KI)	Prof Jan-Eric Litton	
University of Patras (UPAT)	Prof George P. Patrinos	
European Organisation for Rare Diseases (EURORDIS)	Dr Yann Le Cam	
Interactive Biosoftware (IBS)	Dr André Blavier	
Finovatis (Finovatis)	Dr David Koubi	
Instituto de Salud Carlos III (ISCIII)	Dr Manuel Posada	
Innolyst, Inc., Patient Crossroads (PC)	Dr Kyle Brown	
Medical University of Graz (MUG)	Prof Kurt Zatloukal	
University Paris Diderot – Paris 7 (UPD-7)	Prof Odile Boespflug-Tanguy	
University of Malta (UoM)	Prof Alex Felice	
Fondation maladies rares (Fondation MR)	Prof Nicolas Lévy	
University Hospital of Ulm (UULM)	Dr Michael Orth	
University of Zürich (UZH)	Prof Nenad Blau	
Charles University (CUNI)	Prof Milan Macek	
National Institutes of Health – Office of Rare Diseases Research (NIH-ORDR)	Dr Yaffa Rubinstein	
Murdoch University (MURDOCH)	Prof Matthew Bellgard	
Office of Population Health Genomics (OPHG)	Dr Hugh Dawkins	
European Bioinformatics Institute (EMBL-EBI)	Dr Paul Flicek	



Voting by Governing Board - rules

25

- Each Governing Board shall not deliberate and decide validly unless two-thirds ($2/3 = 18$) of its Members are present or represented (quorum)
- Each Member of a Governing Board present or represented in the meeting shall have one vote
- Defaulting Parties may not vote
- Decisions shall be taken by a majority of two-thirds ($2/3$) of the votes = at least 12 votes in favour



New partner- 28

Jan-Eric Litton (BBMRI-ERIC)



26

- **An existing partner in RD-Connect (Karolinska Institute, KI)**
- **Appointed BBMRI-ERIC's first Director General on 22 January 2014 in Graz, Austria**
- **80/20 split in time between BBMRI-ERIC and KI therefore in order to continue work in RD-Connect BBMRI-ERIC wishes to become a full partner**
- **Additional considerations:**
 - Sustainability - RD-Connect may move to a Common Services for RD in BBMRI-ERIC.
 - Formalising the relationship between BBMRI-ERIC and RD-Connect may secure achievements through a stable infrastructure
 - BBMRI-ERIC has adopted BRIF (Bioresource Impact Factor), use in catalogue
 - BBMRI-ERIC sets up a Common Services for IT, Molgenis will be part of that service
 - BBMRI-ERIC launched a Common Services for ELSI
- **Budget to be transferred from KI to BBMRI-ERIC (€100,000 EC contribution)**



New partner - 29

Morris Swertz (Groningen)



27

- **University Medical Center Groningen (UMCG) currently associated partner**
- **Background**
 - Head of the Genomics Coordination Center (GCC) University of Groningen.
 - Developed MOLGENIS, a bioinformatics toolbox to rapidly generate suitable software infrastructures for life scientists to manage, QC, analyze and integrate complex phenotype and genotype data sets (Swertz, 2010)
 - Dutch ELIXIR hub / BBMRI-NL / BBMRI-EU
 - Involved in several EU projects including EU-GEN2PHEN, EU-BioMedBridges, EU-PANACEA and EU-SYSGENET
- **Partnership contributions:**
 - Plays a key role in WP3, enabling development of the sample-level database for biosamples
 - Other work related to genomics platform
- **Additional opportunities:**
 - Participation in the 'pipeline' benchmark and contribution in processing area
 - Contribute NGS data sharing/interpretation toolbox for integration in platform
 - Easing the generation of linked data with LUMC (Peter-Bram/ Marco) and UAVR (Pedro)
- **Budget to be transferred from KI to UMCG (€100,000 EC contribution)**