

Meeting report series

Report of the 2nd ISC Working Group on Biobanks teleconference

24 July 2014, 2-3pm

Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

Prof Hanns Lochmüller, Newcastle, UK (interim chair)
Dr Marina Mora, Milan, Italy
Dr Emmanuelle Rial-Sebbag, Toulouse, France
Dr Yaffa Rubinstein, Bethesda, USA
Dr Nikolajs Zeps, Crawley, Australia

Dr Lilian Lau, IRDiRC Scientific Secretariat
Dr Sophie Höhn, IRDiRC Scientific Secretariat

Apologies

Prof Mats Hansson, Uppsala, Sweden
Dr Veronika Karcagi, Budapest, Hungary
Dr Alastair Kent, London, UK
Prof Jan-Eric Litton, Stockholm, Sweden
Dr Susan Wallace, Leicester, UK

Agenda

- ▶ Report from the Executive and Scientific Committees
- ▶ Appraisal of road map contributions
- ▶ Election of chair
- ▶ AOB

REPORT

Report from the Executive and Scientific Committees

In the past year, following teleconference call and email exchanges, this working group (WG) fed back to the Interdisciplinary Scientific Committee (ISC). The information provided, along with feedbacks from other WGs, were reviewed, condensed and aligned, not only within the ISC but also in coordination with the Diagnostics SC (DSC) and the Therapies SC (TSC).

The EC was keen to have input through the WGs and the SCs in order to streamline their resources, to identify funding priorities for the next few years to work towards reaching the goals of IRDiRC, and to identify focus points of the WGs and of IRDiRC as a consortium. The first draft of the roadmap was presented to the Executive Committee (EC) during a meeting in Berlin in May 2014. It depicts both what the WGs do and the thought process, and lists what work should be done and funded. The funding mechanism remains to be defined by the funders; WGs currently do not have funding to do the works that they suggest.

Two priority ISC areas were picked up to be actioned upon as quickly as possible as they will enable and facilitate rare disease (RD) research in several areas. These actions may require new/adapted mechanisms for seed funding, and short proposals including budgets and timelines have been provided to the EC for decision:

- ▶ Data Standard Clearinghouse (Data Sharing and Bioinformatics WG): important that RD fields know what standards are out there, that they are collected and rated.
- ▶ Consent clauses (Ethics and Governance WG): good practice and best practice requirements in different countries currently not in conformity, making multi-national studies difficult, so consent clauses applied in IRDiRC works should be made available to everyone.

Members were advised to read the report of the EC's Berlin meeting in whole (available on the IRDiRC website at http://www.irdirc.org/?page_id=26). Members were also encouraged to read other reports, in particular those related to the ISC (available at http://www.irdirc.org/?page_id=25).

Appraisal of road map contributions

There isn't any immediate action/milestone defined yet for the Biobanks WG.

The proposed "work supporting policies" points received mixed feedback from the EC:

- ▶ Some of these work on quality in biobanking already taken on by larger consortia, which may not be RD-specific but nonetheless do similar work (e.g. BBMRI).
- ▶ Questioning how to further develop or improve cataloguing samples when there are few.

The proposed "funding policies" points:

- ▶ Questions were raised if there is good access to legacy and historical RD biomaterials, and how to rate them in terms of usefulness. Additional investments would be required if this is deemed worthwhile to look into.
- ▶ Incentives apply across several RD activities, making it vital to talk to other WGs to seek ongoing clinical trials and natural history data collection for participants that are well-phenotyped and characterized, therefore presenting ideal opportunity to collect new biomaterials that can feed into other studies apart from the primary study.

The EC sought specific examples and specific suggestions of work not already carried out by other consortia to avoid duplications. Standard mechanisms to fund research are in place but for some immediate actions, other mechanisms (e.g. seed funding) may be introduced.

Feedback from members of the WG

Any introduction and maintenance of quality standards for biobanking in RD need to be a sustainable effort. If funding is made available to improve the quality and homogenize the standards, a call could be put out for RD biobanks to apply. RD biobanks are often small and have additional/different problems compared to general or population biobanks. Some biobanking networks such as the Rare Diseases Human Biospecimens/Biorepositories (RD-HuB) and EuroBioBank have increased the visibility and identification of RD samples, but quality control and sustainability remain a concern.

There are general biobanks which unwittingly store rare disease samples and not further accessed by researchers. It is important to find a mean to identify these samples and made them available. Given a minimum dataset standard, the system should contain in-built RD-specific descriptions, by Orphanet or International Classification of Diseases (ICD), that can be mapped to samples initially not specified as rare (e.g. matching based on associated-disease names).

For uptake of works by the EC for further investment and investigation, WG members should try to illustrate the issues using case studies. A hypothetical example: a member searching for biomaterials in cancer biobanks but could not locate it due to misclassification and/or phenotyping in a different way. This would allow raising questions such as, if this is an issue of quality, how to overcome the problem, and how to measure its potential to reporting of outcome. A case study is useful for understanding the issue(s) involved and pointing to potential actionable solution(s).

One of the issues previously listed as issue to work on was that of pediatric research, which had not been integrated into the roadmap schematic nor currently highlighted as direct actionable item for Biobanks WG. However, the issue was highlighted in the full report of the ISC to the EC.

Should any member wish to raise specific case study for RD pediatrics that implicates biobanking, it would be not excluded from further discussion within this WG to determine if it would be an issue to be worked on, or an issue to be brought to attention of another WG. Similarly, other legal, ethical and societal issues related to RD may be cross-cutting and of interest for the WG on Ethics and Governance, but also need to be applied to specific RD biobanking questions which are in the remit of this WG.

Election of Chair

Dr Alastair Kent had stepped down as Chair of this WG. Members were asked to consider standing for Chair in the next teleconference, which will then be put to vote.

Additional query

Concern was raised regarding a plan to split the Registries and Natural History WG, another WG under the ISC. The concern was noted by the Chair but not further discussed as this particular teleconference was not the appropriate forum for it.

Main deliverables

- ▶ 2-3 most important issues related to biobanking and/or actionable points for further development of the roadmap by early September 2014
- ▶ Provide feedback from NIH meeting on rare disease biobanks and biospecimens
- ▶ Consideration to stand for the position of chair for this WG
- ▶ Plan the next teleconference to be held in September 2014