

Meeting report series

Report of the 2nd Diagnostics Scientific Committee Meeting

Dublin, Mespil hotel
15th April 2013

Participants

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Apologies

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Agenda

1. Role of Support IRDiRC
2. IRDiRC policy and guidelines document: update
3. IRDiRC Roadmap: report from the Executive Committee meeting
4. Roadmap for Diagnostics SC
5. Finalization of WG membership
6. Definition of WG mandate and tasks
7. Reported needs and clarification
8. Next meeting- data and location

EXECUTIVE SUMMARY

The second meeting of the IRDiRC Diagnostics Scientific Committee (DSC) took place in Dublin on 15 April 2013. This meeting was organized at Mespil Hotel, Dublin.

This meeting took place right after the Executive Committee Meeting that the chairs of the three Scientific Committees attended. It was reported that the Executive Committee reviewed and adopted the Policies and Guidelines document earlier this afternoon.

The organization, role and budget of the Scientific Secretariat were presented.

The composition, mandate and major tasks of the Working groups were finalized so that invitations could be sent. It was suggested to invite representatives from BGI and HVP to further complete the Working Groups.

REPORT

1. Role of SUPPORT IRDiRC

The role of Support-IRDiRC was jointly presented to the DSC and the Therapeutics SC (TSC) by the Scientific Secretariat. The three Work Packages of the project Support-IRDiRC (FP7-funded project) were presented (Implementation, Cooperation and Dissemination), along with the budget, the role of the IRDiRC Committees and the IRDiRC Scientific Secretariat.

Two main points were discussed:

- ▶ Presentation and definition of each SUPPORT IRDiRC Work Package (WP) and Communication plan. Clarification was requested regarding the role of the Scientific Committees in reference to the presentation note: “IRDiRC Scientific Committees will act as reviewers of the Support-IRDiRC activity reports”.
- ▶ Presentation of the SUPPORT-IRDiRC budget. SC members requested a more detailed breakdown of the budget.

2. IRDiRC Policies and guidelines document: update

The DSC was joined by the TSC for this update.

- ▶ Governance structure of IRDiRC.
 - Comment was raised about the governance diagram on the flyer to be disseminated during the Dublin Conference (16-17 April), which might better show the interaction between the different groups.
 - Regarding the structure of each funding organization, international coordination is possible but there will be no common pot for funding projects.
- ▶ The policies and guidelines document has been discussed and modified by the Executive Committee during a meeting earlier in the day. The modified document will be sent around to all SCs when it has received final approval from the funding organizations.

3. IRDiRC Roadmap: report from the Executive Committee meeting

The term “roadmap” has been discussed and found not appropriate and so it has been renamed as an “Action plan”. It has been emphasized that the action plan should indicate achievable activities as well as milestones.

4. Roadmap for Diagnostics SC

Regarding the document disseminated to all SCs, the DSC has considered the following ‘actions’ as being under the objectives of the DSC and its WGs:

- 1-Contribute to a well-curated list of all rare disease -> Ontologies WG
- 2-Promote the discovery of all the new genes that underlie RD -> Sequencing WG

5-Promote coordination between RD and model organism research -> Models WG

8-Contribute and promote a set of standards for RD diagnostic testing and reporting -> Sequencing WG

13-Promote the harmonization and interoperability of ontologies to be applied to databases, registries, and biobanks -> Ontologies WG

14-Promote the interoperability between genotype-phenotype databases -> Genome-Phenome WG

15-Guidelines for the clinical reporting of genomic data sequencing -> Sequencing WG

5. Definition of WG mandate and tasks

Major tasks

- ▶ Promote cooperation
- ▶ Identify opportunities and gaps
- ▶ Suggest areas for rare diseases research to the SCs

Mandate

Each SC has 4 WGs. For the DSC: Ontologies, Sequencing, Models, Genome/Phenome. At least 1 SC member is expected to attend each WG. It is stressed that chronograms and milestones should be defined for each WG to communicate to SC who will present to the Executive Committee.

6. Finalization of WG membership

It was recognized that BGI and HVP may suggest additional key members for these WGs in the near future.

Invitations will be sent to each proposed WG member as soon as possible with the plan that they meet by teleconference in June or July of 2013 – in time to report to the DSC prior to their next meeting.

7. Reported needs and clarification.

Reported needs include the following:

- ▶ How do we calculate the number of new genes discovered?
 - Discussion centred around using OMIM or Orphanet to track new genes discovered as both capture this data. Clarification with Orphanet as to how they capture these data will be sought and if we could use this as the counting mechanism for IRDiRC.
- ▶ Is there a catalog of projects and funding agencies?
 - To be collected by SUPPORT IRDiRC and available on the website.
- ▶ Will an IRDiRC label be defined for such projects?

8. Next meeting – date and location

The next face-to-face meeting of the DSC will be held prior to the next Executive Committee that will take place September 23-24 2013, in Miami.

Dinner: Glaze Bistro of the Mespil Hotel