



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 14th Consortium
Assembly Meeting**

March 19, 2019



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on March 19, 2019, via web/teleconference. It was attended by 33 participants representing 29 member organizations, the Scientific Committees (SC) and the Scientific Secretariat (Sci Sec).

1. Roadmap 2019

- Nine activities are planned in 2019: The Tool for Analysis of Funded Projects, Patient Participation in RD Research, The Galaxy Guide and Metrics on Goal 1 will be completed; Natural History Studies, Clinical Research Network, Indigenous Population, Clinical Data Sharing and Methodologies to Measure Goal 3 will be started.

2. Roadmap 2020

- The CA Chair announced a call for the identification of IRDiRC activities in 2020. The Sci Sec will collect the proposals and an agreement on the Roadmap 2020 will be found during the CA-SC meeting in Leiden, 22-24 of May 2019.
- The IRDiRC Roadmap 2020 should be drafted by early June 2019 since it is a deliverable of the EJP RD Annual Work Plan.

3. Leiden CA-SC meeting

- The general agenda of the meeting was provided to the CA and SC members.
- The Chairs and/or VC were invited to collect ideas from their Committee members for the development of a more detailed agenda. The Sci Sec will support this action and deliver the detailed agenda by May 3, 2019.

4. Upcoming meetings

- The second CA face-to-face meeting of the year is planned for October or November 2019. The CA Chair inquired if any member would like to host the IRDiRC fall meeting.
- The CA agreed to organise the next IRDiRC Congress jointly with RE(ACT) in March 2020.

5. Committee leadership and membership

- Durhane Wong-Rieger has been appointed Chair of the PACC
- The CA Chair highlighted the importance to identify candidates for the ISC Chair and FCC Vice-Chair positions.
- The CA appointed Thomas Morel as member of the TSC.

REPORT

1. Roadmap 2019

- Nine activities are ongoing and/or foreseen for the year 2019:
 - Activity A: The Funder Tool for Analysis of Funded Projects (led by the FCC)
 - Activity B: Patient Participation in RD Research (led by the PACC)
 - Activity C: The Galaxy Guide (led by the TSC)
 - Activity D: Natural History Studies (led by the ISC, joint activity between ISC, FCC & TSC)
 - Activity G: Clinical Research Network (led by the ISC, joint activity between ISC, FCC & TSC)
 - Activity H: Indigenous Population (led by the DSC)
 - Clinical Data Sharing (led by the DSC)
 - Metrics on Goal 1 (will be led by the DSC)
 - Methodologies to measure Goal 3 (will be led by an external group of experts)

- FCC activities
 - The FCC held a teleconference on March 12, 2019 to discuss ongoing matters, including the preliminary results of the survey developed in the context of the Activity A in collaboration with MyScienceWork (MSW) and targeting the identification of the most suitable processes to collect/extract the data on funded projects from FCC members. The completion of the questionnaire by all FCC members is expected before the CA-SC meeting in Leiden (22-24 May, 2019), and is the first step towards the creation of the IRDiRC database of funded projects.
 - MSW developed a beta version of the application which has already been tested on the existing databases combining ontologies for RD. It was able to detect if the project was focused on RD and classify the RD with an accuracy of 80%.
 - The FCC also discussed the first draft agenda for the Leiden meeting.

- CCC activities
 - The Penumbra Project/Krysalis activity: This activity is currently on hold. The CCC members are seeking to identify common goals and projects to advance the vision of IRDiRC. The Vice Chair of the CA and the Chair of the CCC will share a call to identify solutions.

- PACC activities
 - The Patient Participation in RD Research activity: The TF started in fall 2018 and two conference calls were organized on October and November 2018. The next phase of this activity will involve the distribution of a survey to external (non-IRDiRC) patient advocacy organizations in order to identify the barriers to patient participation in RD research.

- DSC activities
 - Indigenous Population activity: This TF has been approved in December 2018. The Sci Sec and the Chair of the DSC are currently working on the candidate profile. A call for nominations will then be launched.
 - Clinical Data Sharing activity: This TF will start in the last quarter of the year 2019.

- ISC activities
 - There is currently a transition period in the ISC since the former Chair was not yet replaced and new members must be identified to join the Committee. The Vice Chair of the ISC is assisted by S. Groft and the Sci Sec to work on these issues, and a timeline of the actions will be proposed during the CA-SC meeting in Leiden, 22-24 May 2019.
 - In the absence of the VC of the committee, it has been agreed that the ISC will be chaired by S. Groft during the CA-SC meeting in Leiden.
 - Clinical Research Network activity: The proposal of this activity was approved in July 2018 and consequently, the start of this activity will be prioritized. The Sci Sec is assisting the VC in the identification of the TF experts. In addition, a call for nominations will be addressed to the CA and published on the IRDiRC website in order to allow internal and external experts to be nominated.
 - Natural History Studies activity: The revised proposal was accepted in November 2018. The Sci Sec will assist the VC in the identification of the TF experts. In addition, a call for nominations will be addressed to the CA and published on the IRDiRC website in order to allow internal and external experts to be nominated.

- TSC activities
 - Galaxy Guide activity: A workshop was organized in December 2018 in Dublin, and an internal report was communicated to the TSC. The group is currently reviewing the building blocks and developing a graphical representation of them. The first version of the guidebook will be ready by April 2019, and the dissemination of the results will start in July 2019.
 - Measure of drug count: This work was supported by the former Sci Sec and therefore, the new Sci Sec and the TSC have to understand how the information for measuring drug count was shaped, and how the metrics were assessed. A dedicated conference call will take place to discuss the issue with TSC chair & VC, Orphanet representatives and the SciSec. This work is currently in progress.
 - The drug counter manuscript is in preparation. It describes the data provided by the former Sci Sec.
 - Data mining and repurposing Task Force: A manuscript is in preparation.
 - Prioritization of the research agenda for RD: This activity is currently on hold since the Committee wishes to prioritize activities emerging from the Galaxy Guide TF.
 - TSC membership: Four new positions will be opened.

- Other activities

- Metrics on Goal 1: This activity was under the responsibility of former IRDiRC members. It will now be led by the DSC.
- Methodologies to measure Goal 3: The Working Group on Goal 3 will develop recommendations on methodologies to assess the impact of diagnoses and therapies in RD patients. A list of experts was identified and invitations were sent. The activities will start as soon as the group is formed. The CA VC offered to chair this Working Group.
- Access to approved therapies: IRDiRC was approached to address the issue of RD drugs availability in countries that cannot afford to pay for them. A dedicated Working Group might be included in IRDiRC activities and therefore, a template for activity proposal will be forwarded to him, as well as to the DSC Chair who will be invited to present the project during the CA-SC meeting in Leiden.

2. Roadmap 2020

- A call for the identification of IRDiRC activities in 2020 was announced during the CA conference call on March 19, 2019. The Sci Sec will collect the proposals from each Committee and an agreement on the activities foreseen in 2020 will be found during the CA-SC meeting in Leiden (May 23, 2019).
- The IRDiRC Roadmap 2020 must be drafted by the beginning of June 2019 since it is a deliverable of the EJP RD annual work plan. The delivery of the Roadmap in due time is very important since EJP RD relies on the scientific recommendations of IRDiRC and the TF outcomes to develop its strategic plan.

3. Leiden CA-SC meeting

- The general agenda was previously communicated to the CA. The meeting will be structured as follows:
 - Day 1 (May 22, 2019): Scientific Committees in parallel and Joint Scientific Committees
 - Day 2 (May 23, 2019): Joint SC and CA, and CA in parallel
 - Day 3 (May 24, 2019): CA alone
- A more detailed agenda needs to be drafted for the meeting. Therefore, the Chairs and/or VC are invited to collect ideas from their Committee members e.g. the confirmation or update of activities on hold, the development of new activities, or the revision or update of recommendations/tools from previous TF. The Sci Sec will provide its support and, if requested, organize conference calls with each Committee.
- The propositions for the Roadmap 2020 will start to be discussed during the next Operating Committee (April 3, 2019), and communicated to the Sci Sec who will deliver the detailed agenda by May 7, 2019. Joint slots will be included in the agenda to ensure a good interconnection between the Committees.

4. Upcoming meetings

- The second CA face-to-face meeting of the year is planned for October or November 2019. Since many members need to have the confirmation of the meeting at least 6 months in advance, the CA Chair inquired if any member would like to host the IRDiRC fall meeting. This event is dedicated to the face-to-face meeting of the CA representing all Constituent Committees and the Chairs & VC of the three Scientific Committees. The host is expected to find a venue and provide a logistical help to the Sci Sec for the organisation of the meeting.
- The last IRDiRC Congress took place in February 2017 and it is expected that the next one should be organised soon. Since other relevant events will be organized within first semester of 2020 (RE(ACT), ECRD), it has been suggested to organize a joint congress between IRDiRC and RE(ACT) in order to increase the impact and avoid dilution of participants. In addition, RE(ACT) 2020 will be organised within the EJP RD, which would facilitate the co-organisation from the administrative point of view. It was proposed that joint scientific sessions (to be developed by a joint scientific committee) could represent the core of the meeting. In addition, to underline the specificity of each organisation (IRDiRC and RE(ACT)), some specific sessions could be proposed, e.g. dedicated workshops (typically organised by RE(ACT) and considered as beneficiary for the research community) would be proposed before the official opening of the congress; a specific policy session (typically organised by IRDiRC) would close the meeting. The event is planned for early in March 2020 in Berlin, Germany. The CA approved the co-organisation of joint IRDiRC-RE(ACT) congress.
- The participation of IRDiRC members in other relevant meetings has been addressed. Several IRDiRC members including the CA VC and the DSC Chair will be present in India for the Conference of the International Undiagnosed Diseases Network. In addition, the CA Chair invited IRDiRC members to highlight their contribution to the consortium when they are invited as speakers.

5. Committee leadership and membership

- PACC Chair election
 - Durhane Wong-Rieger, President & CEO of the Canadian Organization for Rare Disorders (CORD), has been elected Chair of the PACC for a 3-year term, beginning on March 20, 2019 and extending through March 19, 2022.
- ISC Chair election
 - The VC of the ISC and the Sci Sec are currently looking for nominations and/or self-nominations. The election process will be resumed as soon as a candidate is identified.
- FCC Vice-Chair election
 - Since no candidates were nominated or self-nominated in February 2019, a new call for nominations was opened on March 19, 2019. The goal of the Committee is to have a candidate appointed before the CA-SC meeting in Leiden.
- New members: Membership validation
 - Thomas Morel was appointed member of the TSC by the CA.

6. AOB

- Endorsement of the proposal for a moratorium on heritable genome editing: the CA Chair contacted the IRDiRC CA members on 15-03-2019 to ask for their consensus for issuing an endorsement statement on the IRDiRC website. In the absence of objection from the people involved in IRDiRC, the endorsement statement was published on the website on 18-03-2019. During the CA conference call on 19-03-2019, some members pointed out that the endorsement cannot represent the official position of specific IRDiRC organizations. Therefore, a disclaimer was added on the website in order to express this point. It was agreed to further address this issue in the Leiden meeting.

Actions and deliverables

- Roadmap 2020
 - The Sci Sec will collect proposals and the CA and SC will agree on the Roadmap content in Leiden
 - Roadmap 2020 is a deliverable of the EJP RD annual work plan, June 2019
- Leiden CA-SC meeting
 - The Sci Sec will collect ideas from Committees and develop a detailed agenda by May 7, 2019
- CA fall meeting
 - CA members are invited to propose to host the meeting
- Committee leadership
 - CA and SC members will provide their help for the identification of candidates for the ISC Chair and FCC Vice-Chair positions