



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 16th Consortium
Assembly Meeting**

September 12, 2019



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on September 12, 2019, via web/teleconference. It was attended by 27 participants representing member organizations, the Scientific Committees (SC) and the Scientific Secretariat (Sci Sec).

1. New Consortium Assembly members

- The CA Chair announced the nomination of 3 new members in the constituency
 - INSERM (FCC)
 - Yposkesi (CCC)
 - Eloxx Pharmaceuticals (CCC)

2. Review of the Roadmap 2020

- The CA Chair provided a summary of the main comments provided by the Scientific Secretariat and the Op Comm to the 10 Task Force/Working Group proposals received and then described the Gantt chart of activities.
- CA members were invited to send their comments to the SciSec by September 20, 2019.
- These comments will be communicated to the applicants on September 27, 2019.
- The revised version of the proposals will be requested by October 18, 2019.
- The final roadmap will be validated during the CA meeting in Paris, November 21-22.
 - Proposals ranked with high priority in Leiden (CA meeting, May 24, 2019)
 - Chrysalis Project
 - New Technologies and Integrative OMICS
 - Rare Diseases Treatment Access
 - Shared Molecular Etiology Underlying Multiple Rare Diseases
 - Galaxy Guide Phase 2
 - Big Bang Project – Drug Repurposing Guidebook
 - Proposals ranked with medium priority in Leiden (CA meeting, May 24, 2019)
 - Primary Care
 - Machine Readable and Computable Consent
 - Pluto Project on Disregarded Rare Diseases
 - Alternative Business Models for Rare Disease Drug Development

3. Upcoming meetings

- CA face-to-face meeting, November 21-22, Paris: The agenda of the meeting was provided.
- RE(ACT)-IRDiRC Congress and IRDiRC CA-SC meeting, March 11-15, Berlin: The general structure of the event was announced.

REPORT

1. New CA members

- INSERM (French Institute of Health and Medical Research):
 - Member of the FCC
 - Represented by Dr. Catherine Nguyen

- Yposkesi
 - Yposkesi is a pharmaceutical company dedicated to the development and GMP manufacturing of gene and cell therapy products
 - Member of the CCC
 - Represented by Dr. Jean-Pierre Gaspard

- Eloxx Pharmaceuticals
 - Eloxx Pharmaceuticals is a clinical stage pharma company developing novel RNA modulating drug candidates across several RD therapeutic areas
 - Member of the CCC
 - Represented by Dr. Kristie Kapinas

- New members will be invited to present their Organizations/Companies at the next Consortium Assembly meeting in Paris on November 21-22, 2019.

2. Review of the Roadmap 2020

The CA Chair provided a summary of the main comments delivered by the Scientific Secretariat and the OpComm to the 10 Task Force/Working Group proposals received and then described the Gantt chart of activities

- (please **see Annex 1**)

- The main comments formulated during the meeting addressed the following points:
 - Complementarity of the projects: Some projects share complementary goals and may address redundant questions. This matter can be overcome by inviting CA and SC members to participate in activities proposed by other Committees (stimulate interaction and collaboration within IRDiRC).
 - Timeline prioritization of the projects should be developed based on the ongoing activities and the complementarity of the projects (e.g Chrysalis and Pluto projects)
 - Terminology/wording: Regarding “The Pluto project of Disregarded Rare Diseases”, it was questioned why the word “disregarded” was used and not “neglected” as it was described during the CA-SC meeting in Leiden. CA members are invited to provide another terminology. Orphanet may also be contacted to identify the correct wording.

- CA members were invited to further comment the proposals (by email to the Scientific Secretariat) by September 20, 2019.
- Feedback from CA members will be communicated to the applicants on September 20, 2019, and the revised proposals will be requested by October 18, 2019. The final Roadmap 2020 will be validated during the CA meeting in Paris, November 21-22.

3. Upcoming meetings

- The second CA face-to-face meeting of the year will take place in Paris on November 21-22, 2019. The provisional agenda of the meeting was presented (please see **Annex 2**).
- The next IRDiRC Conference will be organized jointly with the RE(ACT) Congress in Berlin on March 11-14, 2020. This event will be immediately followed by the IRDiRC CA-SC Conference on March 14-15 (same venue).
- The general structure of the events is described below:

	Wed 11 March	Thu 12 March	Fri 13 March	Sat 14 March	Sun 15 March
Morning		Session B Diagnostic, WGS, artificial intelligence, new technologies	Session D Molecular etiology of RD, innovative clinical trials, precision medicine	Session F Patient engagement in drug development and clinical trials	IRDiRC committee meeting
Afternoon	Session A (workshop) (1) Methodologies to assess the effect of diagnoses and therapies on RD patients (2) Presentation of the Galaxy Guide	Session C Advanced therapies: gene editing, cell therapy	Session E Access to diagnosis and drugs for all	IRDiRC committee meeting	IRDiRC committee meeting
Evening	Opening Ceremony	Speakers dinner	Social event (if sponsored)		

- Speakers have been identified and invited for each session by the joint Conference/Congress committee; several of them have already confirmed their participation.

Actions and deliverables

- Review of the Roadmap 2020
 - CA members were invited to send their comments to the SciSec by September 20, 2019.
 - Feedback from CA members will be communicated to the applicants on September 27, 2019.
 - The revised version of the proposals will be submitted to the SciSec on October 18, 2019.
 - final Roadmap 2020 will be validated during the CA meeting in Paris, November 21-22.
- Agenda and logistics of the CA meeting in Paris will be circulated in due time.