



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 1st Consortium
Assembly Meeting**

August 2, 2016



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on 2 August 2016, via web/teleconference. It was attended by 24 participants representing 22 member organizations and the Scientific Secretariat (Sci Sec).

1. Updates by the Chair of CA
 - Roche has joined IRDiRC and is represented by Dr Sangeeta Jethwa
 - AMED is now represented by its president, Dr Makoto Suematsu
 - The mechanism to manage the voluntary membership fees account is still a work in progress
2. IRDiRC governance and constituent committees
 - Conference calls of the constituent committees will take place at the end of August
 - CA members not already contacted to participate in a constituent committee should inform the Scientific Secretariat (Sci Sec)
 - Call purposes: identify priorities and goals, elect Chair and point of contact for each group
3. IRDiRC CA face-to-face meeting
 - The meeting will take place at the Grand Hotel Baia Verde in Catania on 23 September 2016
 - Joint meeting, as well as lunch and dinner, with E-Rare will take place on 22 September 2016
 - Members intending to attend should book accommodation directly with the hotel
4. The 3rd IRDiRC conference
 - IRDiRC meetings (CA, constituents, scientific committees) will take place on 6-7 February 2017
 - IRDiRC conference (open to all who register) will take place on 8-9 February 2017
 - Please save the dates and plan attendance to these meetings
 - Venue: Université Pierre et Marie Curie (UPMC) Jussieu, 75005 Paris
 - Organizers of potential satellite meeting(s) should contact the Sci Sec as soon as possible
 - Conference Committee in place, will put together conference agenda and work with their constituents to draft IRDiRC goals for the next 5 years
 - To be confirmed: if registration fees apply, if there will be call for abstracts and presentations
 - Ideas welcomed on program and speaker suggestions
5. Survey results
 - Members were asked to go through and consider the outcomes from recent surveys, in particular of IRDiRC impact and strategic priorities, for discussion at constituent calls.
 - The input on IRDiRC State-of-Play will be taken into consideration for the 2016 report.
6. Any other business
 - The Foundation for Biomedical Research and Innovation (FBRI) in Japan and the Academy of Finland are joining E-Rare.

REPORT

1. Updates by the Chair of Consortium Assembly (CA)

1.1 IRDiRC CA membership

- ▶ Roche joined IRDiRC as a member and will be represented by Dr Sangeeta Jethwa. Dr Omar Khwaja attended this call in her stead as representation of Roche, which is headquartered in Basel, Switzerland.
- ▶ AMED is now represented by its president, Dr Makoto Suematsu. Dr Takeya Adachi, who acted as Dr Suematsu's proxy, expressed their commitment to rare diseases research and IRDiRC.

1.2 Management of voluntary membership fees fund

- ▶ The voluntary membership fees have been used previously to fund travel support for patient organization representatives to IRDiRC meetings, as well as speakers of IRDiRC sessions in conferences.
- ▶ A mechanism is currently being sought to manage the voluntary membership fees account independent of the identity of the Chair of the CA
 - Still in the process of confirming feasibility and details.

2. IRDiRC governance and constituent committees

- ▶ A series of conference calls have been organized for the new IRDiRC constituent committees:
 - Patient advocates committee: 22 August, 2pm CEST/8am EDT
 - Funders committee: 22 August, 3pm CEST/9am EDT
 - Companies committee: 30 August, 2pm CEST/8am EDT

→ Any members who have not yet been contacted to participate in one of these committees should inform the Scientific Secretariat (Sci Sec).

- ▶ Purpose of the calls
 - To think together about priorities that are important to advance rare diseases research from the point of view of the general case of each committee
 - What's holding back progress
 - How to coordinate work and collaborate among members
 - How to inform to the degree that we can
 - To think:

“If I can wave a magic wand, 3 things that will happen in rare diseases that will dramatically accelerate my sector's ability to progress in research to get diagnostics and therapies to patients, what would they be?”
 - To elect Chair and point of contact for each group; self-nomination is encouraged

3. IRDiRC CA face-to-face meeting in Catania

3.1 Practical points

- ▶ The Chair of CA thanked E-Rare for hosting this meeting as well as the Joint IRDiRC – E-Rare meeting
- ▶ The CA face-to-face meeting will take place at the Grand Hotel Baia Verde in Catania, Sicily, Italy
- ▶ Practical travel information document has been circulated to all members

→ Members were also reminded to book their accommodation directly with the hotel (using the booking form provided – if do not have a copy, please contact the Sci Sec)

3.2 Program/agenda

- ▶ Program in a nutshell:
 - 22 September: Joint lunch
Joint IRDiRC – E-Rare meeting
Joint dinner
 - 23 September: All day IRDiRC CA meeting
- ▶ Agenda for the IRDiRC meeting will be circulated in due course
 - If CA members have items they'd like to include for discussion, please contact the Sci Sec
- ▶ E-Rare has put through a proposed agenda for the joint meeting, which would be discussed by the Op Comm on its call on 3 August

4. The 3rd IRDiRC conference in Paris

4.1 IRDiRC meetings vs IRDiRC conference

- ▶ Meetings for CA, constituent and scientific committees will take place on 6-7 February 2017: members only meetings
- ▶ IRDiRC conference will take place on 8-9 February 2017: open to all who register

→ Please save the dates in your diary and plan to attend these meetings!

4.2 Logistics

- ▶ Venue: Université Pierre et Marie Curie (UPMC) at Jussieu, 75005 Paris
 - Located centrally, by the River Seine, easily accessible with public transports
 - The conference center is new and will open in September 2016
 - The venue is also wheelchair accessible

→ Organizers of potential satellite meeting(s) should contact the Sci Sec as soon as possible to enable inclusive scheduling and ensure room availability

4.3 Conference Committee

- ▶ A conference committee with representatives across countries and constituent groups
 - Chris Austin, Hugh Dawkins, Kym Boycott, Hanns Lochmüller, Yann Le Cam, Ana Rath, Jeffery Schloss, Lu Wang, Ruxandra Draghia-Akli, Makoto Suematsu, Sharon Terry, Carlo Incerti, Daria Julkowska
- ▶ First call of this committee on 4 August to discuss the agenda of the conference
- ▶ Committee will also work with their constituents to draft IRDiRC goals of for the next 5 years

4.4 Other queries

- ▶ Registration fees
 - Decision will be made following discussion of the Conference Committee
 - Pending on budgetary availability and needs
 - The European Commission is seeking funding to support experts' travel
 - First conference in Dublin was funded by the European Commission, so it was free
 - Second conference in Shenzhen was fee paying, approximately \$300-400 for early registration and \$500-600 for normal and on-site registration
- ▶ Abstracts and presentations
 - This will also be decided by the Conference Committee
 - The Chair of CA is fully supportive of this component to the conference
 - This will be communicated as soon as possible to facilitate planning of researchers who wish to submit an abstract and present their work
- ▶ Program and speaker suggestions
 - Any members who have suggestions should email Sci Sec and cc the Chair
 - Ideas for posters and abstracts are also welcomed

5. Survey results

Outcomes from recent surveys were collated and circulated; members were asked to go through the responses and consider the points that were raised collectively.

5.1 Surveys on IRDiRC impact and strategic priorities/goals

- ▶ IRDiRC is a membership organization
 - Potential impact comes from joint efforts of its members
 - Goals must be those which excite the members
- ▶ Members should prepare to talk of his/her organization's priority on upcoming constituent calls
 - Think about point(s) which resonates and relevant to work of member/organization

- Think about thing(s) that will benefit from global coordination and cooperation

5.2 IRDiRC State-of-Play

- ▶ Trying to shape a report that will be useful to members and RD community
- ▶ The 2016 report will be written and modified based on input from members

Any other business

The Foundation for Biomedical Research and Innovation (FBRI) in Japan has joined E-Rare, opening avenue for future collaboration with Japan, and the Academy of Finland will also join E-Rare for the next joint call for proposals.

Actions and deliverables

- ▶ Contact Sci Sec if not yet contacted to participate in constituent committee
- ▶ Study survey inputs and prepare discussion points on constituent calls
- ▶ Send to Sci Sec item for agenda in Catania
- ▶ Review joint meeting agenda proposed by E-Rare
- ▶ Save the date for IRDiRC meetings and conference
- ▶ Contact Sci Sec if wish to organize satellite event at IRDiRC conference or if you have suggestions for the conference program or particular speakers

Annex - List of participants

<u>Members</u>	<u>Representative</u>
National Center for Advancing Translational Sciences, NCATS/NIH, USA	Christopher Austin
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
E-RARE 2 Consortium, Europe	Daria Julkowska
European Commission, DG Research and Innovation, EU	Diana Salmen
Agence National de la Recherche, ANR, France	Daria Julkowska
French Foundation for Rare Diseases, France	Christine Fetro
Children’s New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Telethon Foundation, Italy	Lucia Monaco, Leopoldo Robbio
Agency for Medical Research and Development (AMED), Japan	Takeya Adachi
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Korea National Institute of Health, South Korea	Hyun-Young Park
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Roche, Switzerland	Omar Khwaja
Food and Drug Administration, USA	Katherine Needleman
National Eye Institute, NEI/NIH, USA	Santa Tumminia
National Human Genome Research Institute, NHGRI/NIH, USA	Jeffery Schloss, Lu Wang
Pfizer, USA	Katherine Beaverson
PTC Therapeutics, USA	Ellen Welch

<u>Invited Patient Advocacy Groups</u>	
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall

<u>IRDIRC Scientific Secretariat</u>	
SUPPORT-IRDIRC Project	Lilian Lau, Anneliene Jonker

Apologies

Members	Representative
European Organisation for Treatment & Research on Cancer, EORTC, Belgium	Denis Lacombe
Genome Canada, Canada	Cindy Bell
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	Mao Mao
Academy of Finland, Finland	Heikki Vilen
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
Shire Pharmaceuticals, Ireland	Omar Francone
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, Italy	Gualtiero Ricciardi
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Yoshihiro Yoneda
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Ionis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NCI/NIH, USA	Edward Trimble
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS/NIH, USA	Stephen Katz
National Institute of Child Health and Human Development, NICHD/NIH, USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke, NINDS/NIH, USA	Danilo Tagle
Office of Rare Diseases Research, ORDR/NIH, USA	
Sanford Research, USA	David Pearce

Invited Patient Advocacy Groups	
EURORDIS, Europe	Béatrice de Montleau
Genetic Alliance, USA	Sharon Terry

Scientific Committees	
Diagnostics	Kym Boycott
Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam



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