INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 12th Consortium Assembly Meeting

October 29, 2018

IRDiRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on October 29, 2018, via web/teleconference. It was attended by 42 participants representing 30 member organizations, the Scientific Committees (SC) and the Scientific Secretariat (Sci Sec).

1. Consortium Assembly
    - Chairmanship and election
        - Current CA Chair will not seek re-election at the end of his term (December 31, 2018).
        - 2 candidates, Lucia Monaco and David Pearce, presented themselves, their background and their vision as IRDiRC Chair, and answered all questions the members had.
    - Face-to-face meeting in Brussels
        - Face-to-face meeting will take place in Brussels, Belgium December 6-7, 2018 at the European Commission offices.
        - Meeting will last two days, and will include, among others, a roundtable discussion, parallel breakout sessions, and a CA wrap-up session.
        - Members are asked to submit topics they want to discuss in the face-to-face meeting, so they can be included in the agenda.

REPORT

1. Consortium Assembly

1.1 Chairmanship and Election

The current CA Chair, Christopher Austin, will not seek re-election, and therefore we are currently organizing elections, to pass the torch to the next Chair.

- IRDiRC is on an extremely productive path – with many continuing and upcoming activities planned; and protocols and procedures in place to sustain the overall IRDiRC research infrastructure.
- The next Chair will be tasked with continuing on our positive and fruitful trajectory, to continue and accelerate the current path.

CA Chair Candidate Lucia Monaco

- Vision
    - Start from vision we all helped to build together
        - Strong vision, especially because it convenes a concrete sense of urgency
        - IRDiRC currently a mature and well-organized community, all working together to reach the goals
        - Added a sense of accountability
    - Now the time to assess what are the outcomes and inputs of the different stakeholders
        - Current output are policies, guidelines, recommendations, platforms, standards that are recognized by the community
    - Now also time to increase or accelerate IRDiRC's impact by – also in our own day jobs:
        - Reaching out, to promote new membership, foster understanding, adoption and implementation of IRDiRC's principles and tools by the scientific community
        - Influencing to gain access to the relevant strategy-setting working groups and decision-making bodies and promoting plans, decisions, and actions.
        - Acting together to devise joint research programs among all IRDiRC member to reach shared goals
    - To make sure that we don't only work on producing outputs, but to make sure that the produced outputs work in our own organization, aka, that we all implement outputs, as well as external organizations/ people
- Background
    - 17 years' work in Fondazione Telethon

        - 10 years' leading role as CSO; experience in strategic planning and management of the research portfolio
        - Current role: research impact and strategic analysis
    - Representing Fondazione Telethon in IRDiRC since 2011
    - Collaboration with the research community and patient organizations
        - Privileged position to have worked with various stakeholders, among which different funders, researchers, industry and most importantly patient organizations
    - Leading role in international biobanking initiatives
    - Member of international advisory boards
    - 20+ years research experience with continued interest in genetic diseases
- Institution is Fondazione Telethon
    - Italian biomedical charity focused on rare genetic diseases research
    - Founded in 1990 by initiative of a group of patients
    - Supported through fundraising
    - Portfolio of diverse research investments in Italy
    - Research development up to available therapies
- Questions
    - Have you ever dreamt of global data sharing for rare diseases patients?
        - Yes, I have, and it has only become more and more relevant and important in recent years. There are many organizations that are working on this goal, and I have been involved in several of these, such as the international undiagnosed diseases network, RD-Connect and the European Joint Programme for Rare Diseases, where sharing data is at the core whereby patients can be connected and recognized. Both motivation, goodwill and design on one side, and new tools on the other side will hopefully expect to an ever greater level of data sharing in the next era of rare diseases research. We also need to work on the relationship with regulators, as this is the part where it has so far been difficult to put data sharing into practice. Therefore we need to become trusted influencers, in order to allow for this common sharing to take place.
    - What is the best approach to build trust?
        - Building trust and confidence is fundamental; and it needs the involvement of all stakeholders early on continuously throughout the different processes, so that we understand from one another what is happening behind the scenes, to address potential misconceptions.
    - What are you talking about when talking about education?
        - Education can happen at many different moments, such as scientific meetings, conferences, online presence. There are many moments to deliver information and thereby educate on the pathway that research

has to follow. IRDiRC can thereby emphasize efforts of individual members and addressing common themes.

- How can we tackle the issue that IRDiRC is often seen as "closed club'?
  - Currently, it is not always clear where IRDiRC is different from other organizations, and this is a part where all of us need to think about where we are different, and this can help us actively engaging with different stakeholders. Each of us needs to take the time to explain this, providing examples of successful activities that worked together with many different people, that awe have accomplished together.
- How can we work even better together rather than as individuals?
  - We are on the right track with different activities, and several of the current activities, such as activity A will help us with even more concrete ways of what we can do together, each of us within our capacity, but with a shared objective.

CA Chair Candidate David Pearce

- Vision
  - Continue to build on the foundation that previous Chairs have laid
  - Listen to all team members, as IRDiRC is not just about one individual or geographic area, but about many different groups and varied stakeholders
  - Look for synergies between different Committees and groups
  - Emphasize that we are in a whole new realm of rare diseases research
    - Rare disease diagnosis has started a new chapter due to the development and availability of new technologies, such as WES and WGS
    - Gene therapy is a reality and therapy development is improving for rare diseases
    - This new realm requires new sets of data and natural history studies
    - Additionally, how do we pay for diagnosis and treatment for such patients?
- Background
  - PhD in Biochemistry at the University of Bath
  - Started with an interest in Batten disease. Quickly realized that clinical trials require extensive natural history, registry, and other sets of data. Has been a strong supporter such registries from the beginning.
  - Now, at Sanford health, established a research program and incorporated rare diseases program within it.
  - Precision medicine is the future of medicine -- this field needs and can learn much from rare diseases, further emphasizing the need for rare diseases research.
  - Extensive experience building teams and working with geographically diverse individuals and groups

        - Will be able to contribute to IRDiRC's mission, vision, and goals based on related experience and background
        - Has worked extensively with other researchers, pharma and patients
- Questions
    - Do you see IRDiRC moving into the issue of payment and access of rare diseases therapies?
        - Don't want to say that we are going to attack one particular issue, as IRDiRC should represent all constituents, but it is related to the access goal and thus the playing field of IRDiRC. Therefore, it should be included in our agenda, as part of a bigger plan, and have this be part of the CCC discussions on a philosophical basis.
    - How would you raise the profile of IRDiRC?
        - Would want to have the discussion with the larger group, but some low hanging fruit would be to raise our social media presence and have IRDiRC endorse and work with the high-level rare disease research conferences.
    - How do you relate the advocacy part of access to the research focus of IRDiRC?
        - The focus is not to investigate how to pay for each disease therapy; we need to bring the issue of access to the forefront of the agenda, and have it be part of the research pathway of drug development.
    - How would you envision your role within IRDiRC and the European Joint Progamme for Rare Diseases?
        - I have maintained relationships with different European partners, but it goes hand-in-hand with listening and building teams across geographic and sector barriers, and with the global approach to furthering rare disease research on a global scale.
    - What is the best approach to build trust?
        - Take your time, make people understand that there is no catch, and IRDiRC needs to make themselves even more accountable when moving forward.
    - What are you referring to when bringing up education?
        - We could set up various type of training modules to further education in and consequently move rare diseases research forward.
    - How do you envision IRDiRC galvanizing other medical disciplines, such as cancer research, by sharing data on a global scale?
        - Governance is the most important issue, in order to overcome the global data sharing problem, and we need to be collectively smarter, and have artificial intelligence help us with data formatting issues. Eventually, all data from rare diseases patients should be accessible in one common access spot, and we, as IRDiRC, need to advocate for this.
    - How can we tackle the issue that IRDiRC is often seen as a "closed club"?

        - We need to have a much more extensive social media presence, and improved overall communication, for example by presenting IRDiRC at different conferences.
    - How can we work even better together rather than as individuals?
        - Teamwork, aligning stakeholder interests, and making clear that no one has all the answers on their own.

Procedure to elect a new Chair

- Announced election: September 19
- Collection of nominations: early-mid October
    - Nomination includes bio and short statement of motivation from interested parties (via email)
- Fall teleconference: October 29
    - Each candidate will be asked to provide a very short (5 min) presentation stating why they are seeking Chairmanship and to answer any questions
- Electronic vote open: right after the teleconference
- Announce outcome: mid-late November
- Ceremonial hand-off: December 7, 2018 (Brussels F2F)
- Official start of new Chairmanship: January 1, 2019

→ Members are asked vote in the CA Chair poll

1.2 Face-to-face meeting in Brussels

The next CA face-to-face meeting will take place on December 6-7, 2018 in Brussels, Belgium, at the offices of the European Commission.

- Sci Sec has sent detailed logistics information following the call
    - Has included the address and hotel suggestions for members
    - CA members are responsible for booking their own accommodations
- For travel arrangements, please aim to:
    - Arrive prior to meeting start on Thursday, December 6 at 8 AM (recommend flights arriving day prior or by 6 AM on the same day)
    - Stay until evening of Friday, December 7 (recommend flights departing after 8 PM)

The provisional meeting format is the following:

- Thursday, December 6
    - Morning: CA meeting welcome
    - Mid-morning: EU activities in the field of rare diseases research & EJP celebratory session
    - Afternoon: CA meeting continued; parallel CC meetings
    - Evening: Informal CA dinner (7:00 PM)
- Friday, December 7

- Morning: CA meeting continued; priority actions and strategies discussion
- Afternoon: Roundtable; wrap-up CA meeting

Members are asked to submit topics they want to discuss at the face-to-face meeting, so they can be included in the agenda. These ideas for topics should be sent to the Sci Sec.

→ Members should book and arrange their own accommodation for the Brussels CA meeting
→Send in topics for inclusion in the agenda of the Brussels CA meeting

Actions and deliverables

- CA
  - Vote on CA Chair nominations via the electronic survey
  - Book and arrange own accommodation for the Brussels CA meeting
  - Send in topics for inclusion in the agenda of the Brussels CA meeting

Annex - List of participants

Members	**Representative**
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin (Chair)
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Genome Canada, Canada	Cindy Bell
BGI, China	Ning Li
WuXi NEXTCode	Christina
European Commission, DG Research and Innovation, EU	Iiro Eerola
European Commission, DG Health and Food Safety	Nicoline Tamsa
EURORDIS-Rare Diseases Europe, Europe	Virginie Bros-Facer
French Muscular Dystrophy Association (AFM-Téléthon), France	Alexandre Mejat
E-Rare Consortium, Europe and Agence National de Recherche, France	Daria Julkowska
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Istituto Superiore de Sanità, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
Japan Agency for Medical Research and Development (AMED), Japan	Makoto Suematsu, En Kimura
Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid), Japan	Yukiko Nishimura
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Rare Diseases International (RDI), Singapore	Ritu Jain, Paloma Tejada
Korea National Institute of Health, South Korea	Younjhin Ahn
National Institute of Health Carlos III, Spain	Manual Posada
Loulou Foundation, UK	Daniel Lavery
Genetic Alliance, USA	Sharon Terry
Global Genes, USA	Maureen McArthur Hart
National Eye Institute (NEI), USA	Santa Tumminia
National Institute of Dental and Craniofacial Research (NIDCR), USA	Jason Wan
National Human Genome Research Institute (NHGRI), USA	Lisa Chadwick
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
National Organization for Rare Diseases (NORD), USA	Vanessa Boulanger
Pfizer, USA	Katherine Beaverson

Recursion Pharmaceuticals Inc, USA	Tim Considine
Sanford Research, USA	David Pearce

Scientific Committees	
Diagnostics	Gareth Baynam, Kym Boycott
Interdisciplinary	Domenica Taruscio
Therapies	Diego Ardigo, Virginie Hivert

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Anneliene Jonker, Anne-Laure Pham-Hung d'Alexandry d'Orengiani, Ana Rath
NIH/NCATS	Christine Cutillo, Lilian Lau

Apologies

Members	**Representative**
Western Australian Department of Health, Australia	Hugh Dawkins
Rare Voices Australia, Australia	Nicole Millis
Botswana Organization for Rare Diseases (BORDIS), Botswana	Eda Selebatso
Canadian Institutes of Health Research (CIHR), Canada	Christopher McMaster
Canadian Organization for Rare Disorders (CORD), Canada	Durhane Wong-Rieger
Chinese Organization for Rare Disorders (CORD), China	Kevin Huang
WuXi AppTec Co. Ltd., China	James Wu
Academy of Finland, Finland	Heikki Vilen
French Foundation for Rare Diseases, France	Roseline Favresse
Lysogene, France	Karen Aiach
Organization for Rare Diseases India (ORDI), India	Prasanna Kumar Shirol
Indian Organization for Rare Diseases (I-ORD), India/USA	Ramaiah Muthyala
Shire Pharmaceuticals, Ireland	Madhu Natarajan
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Yoshihiro Yoneda
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Rare Diseases South Africa, South Africa	Kelly du Plessis
Roche, Switzerland	Mathew Pletcher

Ultragenyx, Switzerland	Tom Pulles
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Cydan II, USA	James McArthur
Food and Drug Administration (FDA), USA	Katherine Needleman
Genzyme, USA	Daniel Gruskin
Ionis Pharmaceuticals, USA	Brett Monia
National Cancer Institute (NCI), USA	Edward Trimble
National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), USA	Faye Chen
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
NKT Therapeutics, USA	Robert Mashal
PTC Therapeutics, USA	Ellen Welch

Scientific Committees	
Interdisciplinary	Dixie Baker

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