



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 4th Consortium
Assembly Meeting**

12 June 2017



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on 12 June 2017, via web/teleconference. It was attended by 34 participants representing 24 member organisations, the Scientific Committees (Sci Comms) and the Scientific Secretariat (Sci Sec).

1. Updates by the Chair of Consortium Assembly (CA)
 - The IRDiRC Governance document was updated and finalized, posted to the IRDiRC website
 - The IRDiRC goals were finalized and article writing to communicate IRDiRC progress and future goals in a concerted manner is currently underway
 - New dates of Tokyo face-to-face meetings: 10-11 November 2017 (instead of 13-14 November)
 - Two new IRDiRC members: Recursion Pharmaceuticals and Loulou Foundation
2. Members' commitment
 - High-level investment information to be collected through a survey to assure members remain committed as per membership criteria (of investing a minimum of 10M USD over 5 years)
 - Possibility to specify direct and/or indirect cost, therefore total investment in RD research
3. IRDiRC Constituent Committees
 - Patient Advocates Constituent Committee (PACC)
 - Currently engaging the community to apply to become IRDiRC members
 - Three applications received to date, hope to reach out to wider geographical areas
 - A teleconference will be organized in July to discuss charter and strategic goals
 - Funders Constituent Committee (FCC)
 - A document on goals and actions have been compiled and will be sent to FCC members
 - A teleconference will be organized in July to validate actions and priorities, and to establish timeline and deadlines thus kick off selected activities
 - Companies Constituent Committee (CCC)
 - A replacement Chair is needed to lead the CCC
 - The CA Chair will take over, in the interim, the role of identifying potential companies to engage and become IRDiRC members
4. IRDiRC Scientific Committees
 - Therapies Scientific Committee (TSC)
 - Looking to expand membership to improve representation (e.g. clinicians, regulators)
 - Patient-Centered Outcome Measures, Small Population Clinical Trials, and Data Mining and Repurposing Task Forces in various stages of publication of their recommendations
 - Patient Engagement Task Force kicking off with the set up of Steering Committee
 - Clinical Research Network Task Force will be shortly presented to the CA for approval
 - Ongoing discussions on metrics to measure therapy success and on strategic plan to contribute towards new IRDiRC therapy goals
 - Diagnostics Scientific Committee (DSC)
 - A commentary published in the AJHG on enabling diagnosis of rare genetic diseases

- Matchmaker Exchange going on independently and will next meet in June to discuss how to increase specificity of matches and how to quantify impact/successes
 - Solving the Unsolved Task Force just kicked off with experts identified and invited to join the Task Force and attend a workshop on 25 March 2018 in the UK
 - Clinical Data Sharing Task Force will kick off next and focus on clinical work flow where tools and resources are developed to facilitate data sharing among clinicians
 - An article on international cooperation to advance the diagnosis and treatment of rare genetic diseases is also anticipated
 - Interdisciplinary Scientific Committee (ISC)
 - Latest activities focused on consent language and data use agreements, and guiding principles of data standards
 - Automatable Discovery and Access Task Force has no further update since February meeting
 - Privacy-Preserving Record Linkage Task Force are updating their ethico-legal and technical primers, and working on the publication plan
 - SC memberships vote
 - Two new DSC members were approved following a recent online vote
 - Two new TSC nominations, one TSC renewal and one ISC renewal will be up for vote next
5. IRDiRC Scientific Secretariat
- The Sci Sec presented activities it was working on, including supporting teleconferences, submitting periodic report, updating Governance, article writings, etc
 - The Sci Sec will focus on, among other activities, State-of-Play report writing, final conference report, and Task Forces articles and background documents in the next quarter
 - The SUPPORT-IRDiRC contract will be amended to enable hiring of an additional staff to increase the manpower of the team to cope with the increased volume of activities
6. Any other business
- The other Constituent Committees and all Scientific Committees are encouraged to use the FCC documents on goals and actions as template to plan their actions

Annex - List of participants

<u>Members</u>	<u>Representative</u>
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin, Christine Cutillo
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research (CIHR), Canada	Paul Lasko
E-Rare Consortium, Europe	Daria Julkowska
European Commission, DG Research and Innovation, EU	Irene Norstedt, Iiro Eerola
Agence Nationale de la Recherche (ANR), France	Daria Julkowska
French Foundation for Rare Diseases, France	Roseline Favresse
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Telethon Foundation, Italy	Lucia Monaco
Japan Agency for Medical Research and Development (AMED), Japan	Makoto Suematsu, Takeya Adachi
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Roche, Switzerland	Sangeeta Jethwa
Loulou Foundation, UK	Ashley Winslow
Food and Drug Administration (FDA), USA	Katherine Needleman
Genetic Alliance, USA	Sharon Terry
Genzyme, USA	Carlo Incerti
National Cancer Institute (NCI), USA	Jack Welch
National Eye Institute (NEI), USA	Santa Tumminia
National Human Genome Research Institute (NHGRI), USA	Lu Wang, Teri Manolio
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
Pfizer, USA	Katherine Beaverson
Sanford Research, USA	David Pearce

<u>Scientific Committees</u>	
Diagnostics	Kym Boycott, Gareth Baynam
Interdisciplinary	Petra Kaufmann
Therapies	Diego Ardigò, Virginie Hivert

<u>IRDIRC Scientific Secretariat</u>	
SUPPORT-IRDIRC Project	Ana Rath, Lilian Lau

Apologies

<u>Members</u>	<u>Representative</u>
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Genome Canada, Canada	Cindy Bell
BGI, China	Ning Li
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
WuXi AppTec Co. Ltd., China	James Wu
Rare Diseases Europe-EURORDIS, Europe	Béatrice de Montleau
Academy of Finland, Finland	Heikki Vilen
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire Pharmaceuticals, Ireland	
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
Korea National Institute of Health, South Korea	Hyun-Young Park
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Ionis Pharmaceuticals, USA	Brett Monia
National Institute of Arthritis and Musculoskeletal and Skin Diseases, (NIAMS), USA	Stephen Katz
National Organization for Rare Diseases (NORD), USA	Peter Saltonstall
NKT Therapeutics, USA	Robert Mashal
PTC Therapeutics, USA	Ellen Welch
Recursion Pharmaceuticals Inc, USA	Chris Gibson



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