INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 3rd Consortium Assembly Meeting

6 February 2017

IRDiRC

EXECUTIVE SUMMARY

- The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on 6 February 2017 in Paris, France. It was attended by 37 participants in person and 1 via teleconference, representing 22 member organisations, the Scientific Committees (SCs) and the Scientific Secretariat (Sci Sec).

- Four changes of representatives of IRDiRC member organizations were announced.

- An announcement was also made of the outcomes from recent election of Chair and Vice Chair of the Funders, Companies, and Patient Advocates Constituent Committees (FCC, CCC, and PACC).

- Members have been actively involved in the funding of research, clinical studies, and undiagnosed diseases programmes (UDP). Increasing funding and/or research collaborations between funders and/or researchers with patient organisations were also shown, and enthusiasm was expressed towards research potential of the European Reference Networks (ERNs) for rare diseases (RD). In future, funding initiatives and RD research activities will be better captured through annual information submission to form a report.

- Members were introduced to the Saudi Human Genome Project (SHGP), in terms of its mission, objectives, team and facilities, activities, and next steps. The CA Chair welcomed the SHGP's involvement in IRDiRC, and hope this is the first step towards expansion in the MENA region.

- Changes to the SCs were also approved: 1 new nomination, 1 renewal, and 1 end-of-mandate acknowledgement. Additional announcements: TSC has a new Chair and a retirement, and ISC has a new Chair and Vice Chair.

- A number of governance changes were proposed to update outdated information and to add new terms that ensure efficient running of the consortium. A few changes were voted upon, including information exchange between CA and Operating Committee (OC), definition of SC membership, and the removal of limitation to participate in both CA and SC; others – including Funders definition, ineligible organisations, patient advocacy membership and member definition, and termination of inactive members – warranted further discussion, consideration and language change, and the essence would be captured in a document to be circulated prior to an electronic vote.

- Several breakout meetings followed the main CA meeting over the course of the next 1.5 days, i.e. the afternoon of 6 February and the full day of 7 February. Topics of discussions included IRDiRC Task Forces, IRDiRC goals and metrics, and draft activities for 2017-2027.

Annex - List of participants

Members	**Representative**
National Center for Advancing Translational Sciences, NIH/NCATS, USA	Christopher Austin, Christine Cutillo
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
E-RARE-2 Consortium, EU	Daria Julkowska
European Commission, DG Research and Innovation, EU	Iiro Eerola, Irene Norstedt
European Commission, DG SANTÉ, EU	Stefan Schreck
EURORDIS, Europe	Béatrice de Montleau
AFM - French Association against Myopathies, France	Marie-Christine Ouillade
Agence National de la Recherche, ANR, France	Catherine Dargemont
Federal Ministry of Education and Research, Germany	Ralph Schuster
Telethon Foundation, Italy	Lucia Monaco
Japan Agency for Medical Research and Development (AMED), Japan	Takeya Adachi, Ken Ishii, Noriaki Imanishi, Kenjiro Kosaki
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Akifumi Matsuyama, Hanayuki Okura
The Netherlands Organisation for Health Research and Development (ZonMw), the Netherlands	Sonja van Weely, Ineke Slaper-Cortenbach
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Food and Drug Administration, USA (by TC)	Katherine Needleman
Genetic Alliance, USA	Katherine Lambertson
National Cancer Institute, NIH/NCI, USA	Douglas Perrin, Jack Welch
National Human Genome Research Institute, NIH/NHGRI, USA	Lu Wang
National Institute of Neurological Disorders and Stroke, NIH/NINDS, USA	Adam Hartman
Pfizer, USA	Katherine Beaverson

Scientific Committees	
Diagnostics	Kym Boycott, Gareth Baynam
Interdisciplinary	Hanns Lochmüller, Petra Kaufmann
Therapies	Diego Ardigò

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Anneliene Jonker, Lilian Lau, Ana Rath

Apologies

Members	**Representative**
Genome Canada	Cindy Bell
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	James Wu
Chinese Rare Diseases Research Consortium, China	Qing Wang
European Organisation for Treatment & Research on Cancer, EORTC	Denis Lacombe
Academy of Finland, Finland	Heikki Vilen
Fondation Maladies Rares, France	Jean-Louis Mandel
Lysogene, France	Karen Aiach
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Shire Pharmaceuticals, Ireland	David Thomson
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, Italy	Domenica Taruscio
Prosensa, The Netherlands	Scott Clarke
Korea National Institute of Health, South Korea	Hyun-Young Park
Roche, Switzerland	Sangeeta Jethwa
National Institute for Health Research, UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Ionis Pharmaceuticals, USA	Brett Monia
National Eye Institute, NIH/NEI, USA	Santa Tumminia
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIH/NIAMS, USA	Stephen Katz
National Institute of Child Health and Human Development, NIH/NICHD, USA	Melissa Parisi
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall
NKT Therapeutics, USA	Robert Mashal
Office of Rare Diseases Research, NIH/ORDR, USA	
PTC Therapeutics, USA	Ellen Welch
Sanford Research, USA	David Pearce

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