INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 9th Consortium Assembly Meeting

May 15-16, 2018

IRDiRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on May 15-16, 2018 in Vienna, Austria. It was attended by 43 participants in person and 2 via teleconference, representing 28 member organizations, the Scientific Committees (SCs) and the Scientific Secretariat (Sci Sec).

1. New Member Presentations
   - Members were introduced to Cydan II, European Organisation for Treatment & Research on Cancer (EORTC), and Rare Diseases International (RDI)

2. Round Table
   - IRDiRC members presented the 2-3 key things that happened in the past six months at their respective organization relating to the new IRDiRC goals.

3. IRDiRC Constituent Committees
   - The IRDiRC Constituent Committees reported back from their breakout sessions, and presented updates on their current and proposed activities.
   - Companies Constituent Committee (CCC)
     - Activity C: Natural History (NH)/Registry platform with real-world evidence (RWE)
       - Build a universal, broad-based platform that can be used by everyone based on existing case studies, covering different technologies and therapeutic areas
       - Initiative will be run by ISC, but with critical input from CCC
     - Activity H: "Penumbra" Project
       - Investigate the characteristics that make a rare disease "acceptable" or worth investigation at a company based on various risk tolerances, with the aim of expanding the list of rare diseases that companies are willing to work on.
   - Funder Constituent Committee (FCC)
     - Ethical, Legal and Social Implications (ELSI) of rare diseases research
       - The FCC discussed the outcome of a short survey on funding opportunities for ELSI research, including potential gaps to overcome in the future.
       - After the discussion of the results of the questionnaire, three next steps were identified:
         - Explore better existing ELSI recommendations worldwide
         - Address the lack of expertise in reviewing ELSI grants applications
         - Prioritize ELSI topics
     - Activity A: Establish a process to coordinate and prioritize research funding efforts
       - The FCC discussed the preliminary results of a preceding questionnaire for funding collaboration as a concurrent part of Activity A
       - Preliminary conclusions: collaborative activities are possible for the majority of funders, especially using well-established schemes. In order to set up

collaborative calls, topics need to be defined in advance and the review process needs to be well-defined prior to the launch of the call.

- First discussion on international collaboration with developing countries
  - Explore a better understanding of indigenous populations as starting points
  - Bring IRDiRC to "developing" countries to attract and include policy makers, and have a discussion at their home turf
  - Align and discuss with Patient Advocacy Constituent Committee(PACC) where there are already several members from developing countries

- Patient Advocates Constituent Committee (PACC)
  - Activity B: Leverage IRDiRC's stakeholder and geographic representation for complementary environmental scan
    - Multi-step project
      - PACC-led Task Force to define feasibility and implementation
      - Initiate PACC focus groups
      - Initiate Funders and Companies scan
      - Initiate Scientific Committee scan to represent academic researchers across jurisdictions
  - Activity F: Creation of a position statement including specific recommendations for how to apply the second new IRDiRC goal (1000 new therapies) internationally, thereby aimed at stimulating drug development equally worldwide and including patients' perspectives in that therapy development
    - Recommendations or position paper re: multi-stakeholder collaboration on regulatory and therapy development pathways, activity to be started after Activity B

4. IRDiRC Priority Actions and Strategies

- Two proposals were approved by vote of the CA to start immediately
  - Activity A: Establish a process to coordinate and prioritize research funding efforts
    - Proposal by FCC
    - Put in place the tool (database) allowing in-depth analysis of funded projects and the rare diseases research funding landscape at an international level, and establish processes whereby funders can coordinate and prioritize efforts on research and avoid unnecessary duplication.
  - Activity C: Galaxy Guide - guidebook for drug developers describing available tools/initiatives specific for RD and how to best use them
    - Proposal by TSC
    - The Galaxy Guide project aims at creating a simple guidebook for academic and industrial drug developers describing the available tools and initiatives specific for rare disease development and how to best use them.
- One proposal was discussed by the CA, but not put forward to vote, awaiting further refinement
  - Activity G proposal: Clinical Research Networks for Rare Diseases
    - Proposal by ISC, FCC, TSC; Activity to be headed by ISC

- Activity aims to develop guiding principles for networks to facilitate international collaboration and promote community-based research and data provision using innovative approaches.
- Proposal will be refined to include recent developments including the European Reference Networks, and provide a more defined focus including specific outcomes

- The discussion was initiated with regards to which activities should be included in the IRDiRC 2019-2020 roadmap
  - Several of the 2018 activities will run into 2019
  - Time to have another look at the list of activities compiled late in 2017 to determine which of the other ideas (not previously selected), if any, should be pursued
  - Additional ideas are welcome, to add to this running list of potential activities

REPORT

1. Welcome

The Chair of the Consortium Assembly welcomed all participants to the Consortium Assembly (CA) meeting in Vienna, Austria, and introduced the agenda of the meeting.

2. New Member Presentations

Over the past six months, five new members have joined IRDiRC. The two new members that were present at the meeting, Cydan II and Rare Diseases International (RDI), presented themselves, joined by the relatively unknown member, the European Organisation for Research and Treatment of Cancer (EORTC).

2.1 Cydan II

- Cydan II
  - Mission statement: accelerating rare genetic disease therapies
    - Vision: Help bring drugs to patients with rare diseases
  - Company that goes through funding cycles of four years, now in the second funding cycle
    - Biotech accelerator, developing companies that in turn develop orphan drugs
    - Searches for partner with asset that has a strong scientific rational to bring a transformative therapy to rare disease patients
    - Develop a de-risking model
    - Found the company, populate the company, and try to make the drug move through development towards the clinic
  - Cydan II is a small team, with 5 team member, but a large number of consultants
  - First funding cycle: Cydan I
    - Looked at over 1000 opportunities
    - Did due diligence to 60 programs
    - De-risking for 12 programs
    - Launched 3 companies successfully
    - Lead to hope that Cydan II can have an even more successful output
  - Example of timeline: Vtesse
    - ID asset in Fall 2013
    - Diligence in September 2014
    - Company Launch in December 2014
    - Finalizing phase III in 2018

2.2 EORTC

- European Organisation for Research and Treatment of Cancer (EORTC)
  - Pan-European cancer clinical and translational research infrastructure
    - 55 years of experience

        - Private, non-profit non-governmental organization
        - Large focus on rare cancers, as most types of cancer are rare
        - Game changer, developed and influenced rare cancer treatment in many cancer types
        - Active in precision oncology
        - Founding body of the International Rare Cancer Initiative (IRCI)
        - Partner in European Reference Networks (ERN)
    - Network consists of over 5500 collaborators, 930 institutions, 27 countries, more than 80 papers
        - Network coordinated in the Brussels office
    - Mission: follow up throughout life, till death, with all patients, ensuring a long-term follow-up, understanding the mechanism of relapse
        - More than 25000 patients in follow-up currently
    - Rich pipeline in clinical trials:
        - 15 new clinical trials at any point in time open to patients entering
        - Over 50 ongoing studies
        - Studies in protocol development, protocol outline development and studies in regulatory activation
    - Large reach on cancer patients
        - 91771 patients in clinical trials or follow up worldwide
        - 82136 patients in European Union
        - 3774 patients in Europe outside the European Union
        - 5861 patients in the rest of the world, in partnerships with other organizations, such as NCI

2.3 RDI

- Rare Diseases International (RDI)
    - Global alliance of people living with a rare disease of all nationalities across all rare diseases
        - Brings together national and regional rare disease patient alliances from around the world as well as international rare disease-specific federations to create the global alliance of rare disease patients and families.
        - RDI has more than 50 member organizations from over 30 countries, that in turn represent rare disease patient groups in more than 100 countries worldwide
        - RDI is patient-centric, patient-driven and patient-led
    - Mission: to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities.
    - RDI encourages member organizations to be involved in different aspects of research, and is committed to the development of research policy, for the larger rare diseases community

3. Round Table

Representative of each member organization was asked to present 2-3 key things which happened in the past six months that this group could benefit from knowing and relevant to the IRDiRC goals 2017-2027.

4. IRDiRC Constituent Committees

4.1 Companies Constituent Committee (CCC)

- CCC is composed of 14 members, representing companies from North America, Europe and Asia
- Mission of the CCC is to bring more and better therapies to the market. In order to do so, the CCC focuses on:
  - Identifying common roadblocks to efficient execution of rare diseases research in the company space
  - Promoting concerted efforts and collaborations on pre-competitive aspects of R&D
- This should lead to the creation of a more amenable therapeutic development environment and enable increased output
- CCC presented the actions it plans to work on:
  - Activity D: Natural history and registry (NH/R) platform with real-world evidence (RWE)
    - Build a universal, broad-based platform for use by all stakeholders to drive decisions. Goals of the activity:
      - Establish best practices
      - Define standards for collection of high quality and interoperable data
      - Build infrastructure backbone
      - Build knowledge of diseases currently without available therapies
      - Ideally based on existing case studies, covering different technologies and therapeutic areas
    - Will be run by the ISC but CCC has critical input for creation of appropriate data and outputs relevant to patient fee-for-service, regulatory needs, and HTA/payer needs
  - Activity H: “Penumbra” Project
    - Investigate the characteristics that make a rare disease “acceptable” or worth investigation at a company based on various risk tolerances, with the aim of expanding the list of rare diseases that companies are willing to work on.
      - Generate knowledge away from the 10-15 core diseases currently worked on and address those with little/no activities
      - Preparatory phase in 2018 for more extensive work at a later stage
    - Focus CCC’s efforts on those approaches that will move diseases from outer to inner orbits, thereby not focusing on the 100 “core” diseases but on a broader set
- Some other ideas were discussed, but need to be better developed, in order to be able to be included in the 2019-2020 roadmap

- Classification on basis of biology rather than traditional disease names
- Tools to enable multiplex sharing of genotype and phenotype data to facilitate use of this data
  - Potential joint effort with DSC
- Use IRDiRC bully pulpit (e.g., position papers) to make new approaches with potential for logarithmic improvement viewed as less risky/more acceptable

- In addition, the CCC is also thinking about better criteria for new company members to join IRDiRC
  - Conceptualizing differently from initial "spend-only to join" criteria, similar to the PACC currently re-defining criteria
  - Create the basis for a dedicated set of members

4.2 Funders Constituent Committee (FCC)

- FCC is composed of 31 members, representing funders from North America, Europe, Asia and Australia
- The FCC had a discussion on Ethical, Legal and Social Implications (ELSI) of rare diseases research
  - ELSI are aspects of rare diseases research that have recently received increased attention.
  - The FCC discussed the outcome of a short survey on funding opportunities for ELSI research including potential gaps to overcome in the future.
    - What are current ELSI funding opportunities? Which topics should be covered by ELSI funding calls? What are common ELSI-related issues and questions? What are potential opportunities for international collaboration?
    - Questionnaire resulted in a list of topics that should be covered by ELSI funding
      - Data collection
      - Data sharing
      - Burden of rare disease
      - Consent and patient information
      - Patient engagement
  - After discussion of the questionnaire's results, three next steps were identified:
    - Explore existing ELSI recommendations worldwide
      - Better integration and communication of existing efforts
      - Report on existing ELSI funding and recommendations
    - Address the lack of expertise in reviewing ELSI grants applications
    - Prioritize ELSI topics
      - Discuss with other committees to better align across all of IRDiRC
      - Organize a workshop (together with E-Rare) to discuss and address gaps?
      - What are the main ELSI issues that prevent us from reaching the IRDiRC Goals?
      - Topic to be developed further in order to be included in IRDiRC 2019-2020 roadmap

- The FCC discussed the preliminary results of a preceding questionnaire for funding collaboration as a concurrent part of Activity A
  - Preliminary conclusions
    - Collaborative activities are possible for the majority of respondents
    - In order to establish a funding collaboration, the funding strategies that are most likely to be used are bi- & multinational collaborations or eventually mutual recognition, especially when using well-established schemes
    - Most of the funders have similar evaluation processes (peer review)
    - New funding opportunities need to be established well in advance
    - Most funding initiatives implemented at different institutions have the same duration (3-5 years)
    - Several topics were identified higher priority for potential collaborative funding efforts
  - Next steps
    - Get a better understanding of the funding collaboration landscape, so ask ALL funding members to fill out the survey
    - Better inform each other (FCC members) of upcoming funding opportunities
    - Interface with PACC to understand their recommendations on co-funding or topics
    - Better understand evaluation and review processes across member institutions
    - FCC members asked to interface with appropriate people at home institutions to better understand decision making process for potential future collaborative calls
    - Investigate several topics for first potential collaborative funding opportunity, but much more preparation is needed before the FCC can launch a collaborative call
- The FCC also initiated a discussion on international collaboration with developing countries, and exchanged preliminary ideas on how to start such collaborations
  - Explore as a starting point a better understanding of indigenous populations in "developed" countries to start research "back home", and share knowledge afterwards
  - Bring IRDiRC to "developing" countries to attract and include policy makers, and have a discussion on their home turf
  - Align and discuss with PACC in which there are already several members from developing countries
  - Organize a dedicated meeting in a "developing country" to organize exchange, and lower the barrier from both sides
  - Start discussions with policy makers, on both sides, to see what is possible

4.3 Patient Advocates Constituent Committee (PACC)

- PACC is composed of 13 members, representing patient advocates from North America, Europe, Africa, Asia and Australia

- PACC presented the actions it plans to work on:
  - Activity B:
    - Initial idea: Identification of barriers to patient participation in rare disease research and recommendations to remove them via multi-stakeholder survey
      - Initial suggested approach was determined to be complex and expensive -- resources, time, expertise, and access constraints would be significant -- and objectives could be accomplished within a more limited framework. Therefore the activity idea was refined.
    - New idea and goal: Leverage IRDiRC's stakeholder and geographic representation for complementary environmental scan. Steps included:
      - PACC-led Task Force to define implementation
      - Initiate PACC focus groups
      - Initiate Funders and Companies scan
      - Initiate Scientific Committee scan to represent academic researchers across jurisdictions
    - Potential outcomes:
      - Identify barriers from the perspective of all stakeholders within IRDiRC
      - Determine whether existing strategies align with barriers identified by stakeholders
      - Create list of the suite of methodologies/strategies used in patient engagement (from Funders/Companies perspective)
        - Developing countries can view the list; see which they potentially could use
      - Develop recommendations:
        - Facilitate better patient engagement across geographic areas with shared resources
        - Determine strategic areas for new funding initiatives
        - How do the findings inform future activities of IRDiRC
    - Initial timeline:
      - Refine and finalize proposal, and submit to vote by CA – by June 30
      - Initiate PACC-led Task Force – by September 2018
        - Include representative members from PACC, FCC, CCC, and SCs in addition to a methodologist (external), a qualitative data analysis expert (external), and a data protection expert (Dixie or external?). Similar to traditional TF, but with more internal members.
      - Start TF work to define implementation details
        - Define design and implementation details – via TC, Q3-4 2018
          - Determine questions to ask the varied stakeholders and instrument (e.g., interview only, inclusion of survey, digital?)

        - Develop detailed budget and reach of focus groups and scans – via TC and finalize at Dec 6-7 CA meeting
      - Initiate PACC focus groups and FCC, CCC, and SC scans – Q1 2019
      - Intermediate review of results from three streams (PACC focus groups, FCC/CCC scan, SCs scan) – Q2 2019 (take advantage of May 2019 CA F2F)
      - Analyze results and develop recommendations – Q3 2019
      - Publish/disseminate recommendations – Q4 2019
    - Points that should be kept in mind, in order to best focus this questionnaire are:
      - How to make the survey focused enough? Focus on clarifying distinct goals of the survey
        - Identification of the right questions will be performed by cross-disciplinary TF; TF members are integral
        - Adapt questions based on targeted stakeholder in order to get a full spectrum of responses and meaningful results
      - Look at similar or related efforts to tailoring this scan to the specific needs of IRDiRC
        - Learn from existing initiatives to avoid pitfalls and duplication
        - Examples : EUPATI, IMI efforts, PARADIGM
  - Activity F: Creation of a position statement including specific recommendations for how to apply the second new IRDiRC goal (1000 new therapies) internationally, thereby aimed at stimulating drug development equally worldwide and including patients' perspectives in that therapy development.
    - The expected outcome is a recommendations or position paper re: multi-stakeholder collaboration on regulatory and therapy development pathways
    - This action is scheduled to take place after Activity B
      - Potentially include some related questions within Activity B
- Other areas of need and potential ideas were discussed, but need to be further developed before included in the 2019-2020 roadmap
  - Data protection/ethics and research oversight
  - Prevention research
  - Capacity building re: patient involvement in research
    - Multi-sector/stakeholder (patient, researcher, institution)
    - E.g. train-the-trainer knowledge platforms
  - Access
    - Innovative instruments/technologies
      - For data collection (re: cost and access)
      - Language translation and/or pictorial representation

5. IRDiRC Priority Actions and Strategies

Three Activity proposals were set out for discussion, for which two were approved by vote.

5.1 Activity A proposal: Establish a process to coordinate and prioritize research funding efforts

- Proposal by FCC
- The main goals of this activity are:
  - Establish a tool (database) that allows for in-depth analysis of funded projects and the rare diseases research funding landscape at an international level
  - Establish processes whereby funders can coordinate and prioritize efforts on research and avoid unnecessary duplication
    - Explore existing tools/platforms that allow identification/comparison/analysis of current and previous funding; and then adapt and implement the most efficient tool
      - Call for tenders is in process of preparation for a subcontract to create the tool
    - Create real-time tracking database for upcoming calls
      - Preceding survey performed to better understand the processes of establishing funding opportunities and new collaborations by funders
    - Create "funders pipeline" to promote the "journey" of the project from basic to applied research
- The steps in this activity are:
  - Submit to CA Activity proposal for vote
  - Finalize and launch call for tenders
  - Discuss potential next steps based on the survey outcomes
    - Aim to have Funders agree on a format and mechanism through which collaborative funding calls could be made.
    - Need to get further information on: how confidential information could be collected, what timelines/deadlines would need to follow, what type of actions could result from the "database" of planned funding opportunities, and what metrics could be used to measure success.

Activity A proposal was approved by CA members

5.2 Activity C proposal: Guidebook for drug developers describing available tools/initiatives specific for RD and how to best use them

- Proposal by TSC
- This project aims to create a simple guidebook for academic and industrial drug developers that describes the available tools and initiatives specific for rare disease development and how to best use them.
  - Concept: there is a way to develop drugs for rare diseases that is different from the classic drug development pathway used for blockbuster drugs
  - The approach for Activity C is like a game, the framework being the game board that includes the typical steps of drug-development.

- The building blocks of the game are all single initiatives and tools. Each building block will have a description sheet that will enable developers, academic researchers, and others to decide which block to use, according to their need in the process of orphan drug-development

- The steps in this activity are:
  - Submit to CA Activity proposal for vote
  - Finalize consolidated draft list of tools
  - Brainstorm and prioritize Building Blocks list
  - Fill out Building blocks form
  - Workshop to consolidate and discuss information on December 12-13, 2018

Activity C proposal was approved by CA members

5.3 Activity G proposal: Clinical Research Networks for Rare Diseases

- Proposal by ISC, FCC, TSC; Activity to be headed by ISC
- Objective of the Task Force is two-fold:
  - First objective is to develop guiding principles for networks to facilitate international collaboration:
    - Landscape analysis of already existing efforts
    - Alignment of protocols for data collection, cost sharing
    - Identify barriers to collaboration among CRNs and methods for overcoming them (limitations and opportunities)
  - Second objective is to promote community-based research and data provision using innovative approaches:
    - Reaching research-naïve locales
    - Recommendations on innovative models and use of technologies (e.g., social media) for data provision and identification of patients
- The steps in this activity are:
  - Proposal drafted and initially presented in Tokyo
  - Discussed at joint Scientific Committee meetings for consensus
  - Fine tune the proposal with suggestions and comments
  - Present to the CA for vote by June 30
- Comments and suggestions:
  - Members suggested that the proposal, in its current form, does not include recent developments of the European Reference Networks
    - Task Force proposal needs to be re-scoped in order to take into account changes before brought to vote
    - Needs clear explanation of the outcomes and recommendations
    - What is the exact goal of this Task Force, and how does it want to contribute to the current landscape?
  - Proposal is too broad in its current format

- Focus on one of the two objectives
- Proposal will be refined to include recent developments including the European Reference Networks, and provide a more defined focus including specific outcomes

➔ Refine proposal for Activity G, prior to bringing the proposal to CA for vote

5.3 Potential additional thoughts for future activities

While many of the current activities will last into 2019, it is time to start thinking about activities that can be included in the IRDiRC 2019-2020 Roadmap.

- Several of the 2018 activities will run into 2019
- Many ideas that could be incorporated into the Roadmap pipeline were discussed throughout the three days of the meeting
- Time to get out the comprehensive, running list that was collected around the Tokyo meeting
  - IRDiRC is currently working on ~10 activities of 150 total proposed activities
  - Time to have another look, and select new priority actions
- Additional ideas are welcome, and can be submitted to the Sci Sec to be added to this running list of potential activities
  - For example, idea suggested around outstanding issues surrounding gene therapy

→Re-send running idea and activities list to all members for reflection
→Members that have additional ideas for potential future activities, send to Sci Sec

Actions and deliverables

- Members asked to submit ideas and suggestions for future activities to Sci Sec and review running list
- Refine proposal for Activity G prior to bringing the proposal to CA for vote
- Send out running list of ideas and potential activities to all members for reflection

Annex - List of participants

Members	**Representative**
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin (Chair)
Rare Voices Australia, Australia	Nicole Millis
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Canadian Institutes of Health Research (CIHR), Canada	Paul Lasko
Genome Canada, Canada	Cindy Bell
Chinese Organization for Rare Disorders, China	Qi Sun
WuXi Next Code Ltd., China (observer)	Christina Waters (observer)
E-Rare Consortium, Europe	Daria Julkowska
European Commission, DG Research and Innovation, EU	Iiro Eerola, Irene Norstedt
European Commission, DG Health and Food Safety, EU	Stefan Schreck
Academy of Finland, Finland	Heikki Vilen
EURORDIS-Rare Diseases Europe, France	Virginie Bros-Facer
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Federal Ministry of Education and Research, Germany	Ralph Schuster
Indian Organization for Rare Diseases, India/USA	Ramaiah Muthyala
Istituto Superiore di Sanità, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid), Japan	Yukiko Nishimura
Japan Agency for Medical Research and Development (AMED), Japan	Takeya Adachi, Yuko Endo, Aikichi Iwamoto, En Kimura
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Rare Diseases International, Singapore	Ritu Jain
National Institute of Health Carlos III, Spain	Manuel Posada
Cydan II, USA	Shi Yin Foo
Food and Drug Administration, Office of Orphan Products Development (FDA/OOPD)	Katherine Needleman (teleconference)
Genetic Alliance, USA	Katherine Lambertson
National Human Genome Research Institute (NHGRI), USA	Teri Manolio
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman (teleconference)
Pfizer, USA	Katherine Beaverson
Recursion Pharmaceuticals Inc, USA	Tim Considine

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

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Marlene Jagut, 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 (Vice Chair)
Botswana Organization for Rare Diseases (BORDIS), Botswana	Eda Selebasto
Canadian Organization for Rare Disorders, Canada	Durhane Wong-Rieger
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	James Wu
Agence Nationale de la Recherche	Dominique Dunon-Bluteau
French Foundation for Rare Diseases, France	Daniel Scherman
Lysogene, France	Karen Aiach
Children's New Hospitals Management Group, Georgia	Oleg Kvlividze
Organization for Rare Diseases India (ORD-I)	Prasanna Kumar Shirol
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, Saudi Arabia	Sultan Turki AlSedairy
Rare Diseases South Africa, South Africa	Kelly du Plessis
Korea National Institute of Health, South Korea	Hyun-Young Park
Roche, Switzerland	Mathew Pletcher
Ultragenyx, Switzerland	Tom Pulles

Loulou Foundation, UK	Daniel Lavery
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Global Genes, USA	Nicole Boice
Ionis Pharmaceuticals, USA	Brett Monia
National Cancer Institute (NCI), USA	Jack Welch
National Eye Institute (NEI), USA	Santa Tumminia
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
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
Sanford Research, USA	David Pearce

IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM