INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 14th Executive Committee Meeting

16 March 2015

IRDiRC

EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 16 March 2015 in Madrid, Spain. The fourteenth meeting of the Exec Comm was attended by **24 participants**, representing 18 member organisations.

The Chair presented his activities report, and members EORTC and Genomics England also presented their current initiatives.

The "**State-of-Play of research** in the field of rare diseases" 2014 report will be made publicly available on the website after minor changes and a letter to the editor of a journal about it will be submitted as soon as possible.

The production of the next edition of this report will follow a modified scheme:

- Preparation of the Report by the Scientific Secretariat (Sci Sec), including a review of published articles in the past year, an analysis of the areas covered by funded projects, and a review of indicators of success as defined on the IRDiRC website;
- Submission of the Report for review by the Exec and Sci Comm members, to include edition of text and comments on the trends, breakthroughs and gaps;
- Finalisation of the Report and publication on the IRDiRC website;
- Submission of an article to a peer-reviewed journal.

Task Forces membership, based on received nominations, was reviewed. A few organisations, which should be represented on the Task Forces for "Patient Relevant/Reported Outcome Measures" and "Small Population Clinical Trials", were selected. These core representatives are tasked to invite other experts to join the Task Forces, taking into consideration the nominations received but not bound to select solely from the list provided. Only one representative will be selected to represent the three Sci Comms to each Task Force to ensure a good liaison. Two other Task Forces ("Matchmaker Exchange" and "Computable Consent Forms") will be joined by Task Teams of Global Alliance for Genomics and Health (GA4GH) already in place. A fifth Task Force will be constituted on "Repurposing and Data Mining".

The **representation of Patient Organisations** at the Exec Comm level will not be modified. **No associated status** will be created for organisations not fulfilling membership criteria.

The **IRDiRC website** will be substantially modified to better highlight the current activities.

REPORT

Welcome from the Director General of Institute of Health Carlos III

The Director General of the Institute of Health Carlos III, Jesús Fernández Crespo, welcomed the Exec Comm to the Institute and to Madrid, Spain.

Chair's activity report

No new member joined IRDiRC since the last face-to-face meeting in Shenzhen, China. A number of discussions with potential industry members were revived, thanks to the Chair of the Therapies Sci Comm.

The Chair would like to thank members who contributed towards the voluntary membership fees. The money will be primarily used to support the travel of patient organisation representatives to the Exec Comm meetings. The Chair also welcomed ideas for outreach activities that could be supported by these funds.

The Chair represented IRDiRC in a few public lectures and meetings. He also gave a print interview to International Innovation, issue 172, 18 February 2015, which highlighted some of IRDiRC achievements (e.g. approval of therapies developed by member companies) and discussed IRDiRC overall goals and upcoming activities.

Presentation: EORTC's SPECTArare

EORTC's Screening Patients for Efficient Clinical Trial Access (SPECTA) programme was presented to the Exec Comm, in particular, SPECTArare which pertains to rare tumours. SPECTArare was set up and is dedicated to setting up collaborative prospective clinical trials with programmes that are optimised for access to patients by utilising academic capture of biological sub-groups, coupled with technological expertise, oriented for full clinical trials, and based on industry cooperation for drug development.

SPECTA was initially built for frequent tumours to get the concept running, but faced many challenges, including lack of access for patients to treatment, request for patients' consent with no promises of treatment that is also acceptable to the ethics committee, and high cost of protocol development. These were overcome and hundreds of patients across Europe now participate in SPECTA. The system developed enables EORTC to structure and create new subgroups, built in consultation with patients, industry and regulators.

SPECTA takes into account the interests and needs of all stakeholders through multiple approaches, including breaking the silo approach of drug development, streamlining screening programmes, rapid identification of patients with specific genotypes, possibility of calling back patients, setting up of central biobanks that is audit compliant, and providing systematic NGS for all patients.

Presentation: NIHR England and the 100K Genomes Project

The National Institute for Health Research (NIHR), supported by the National Health Service (NHS) England, launched the 100K Genomes Project in December 2012 and funded this project through Genomics England. The 100K Genomes Project focuses on patients with rare diseases and their families, and patients with cancer.

The projects itself received infrastructure investment by the NIHR to construct the National Logistics and Biosample Centre in Milton Keynes, to complement a similar centre in Great Ormond Street Hospital (GOSH) which manages 15M biological samples in a fully automated facility. Additionally investment was also received for the enhanced phenotyping side of the project, in particular to train young doctors and nurses in clinical genomics. In terms of data processing, the Medical Research Council awarded funding to implement platforms and computer power, and enable moving from in-house storage to a data security centre. Currently, large libraries have been set up to allow access of specific de-identified data to only authorised users; to maintain public trust, data will not be available to be extracted and integrated into external systems. The Wellcome Trust has also invested in a new sequencing hub in Cambridge.

The NHS estimated to deliver 40,000 diagnoses each year in the future. Health Innovation is a nationally accepted programme to reduce ethics and paperwork burden, and larger initiatives to bring new informatics to NIH hospitals will facilitate enrolment of patients and better serve the population. Connecting hospitals to a central hub furthers data capture in a systematic and controlled manner. A national tendering process ensures cost-effective and timely whole genome sequencing; other negotiated contract led to setting up a safe haven for data storage. The EBI are in charge of annotating the genome, and Genomics England is set up as a non-profit company, where surplus money will be reinvested. Data interpretation will be carried out by Genomics England Clinical Interpretation Partnership (GECIP).

The pilot projects have been essential to develop scalable systems at the national level for the 100K Genomes Project. International collaborations are enabled through early data sharing, and this innovation project is embedded in the NHS to achieve sustainability.

State-of-Play project: goal and methodology

The Support-IRDiRC contract called for the publication of an annual State-of-Play report to assist the work and discussion of IRDiRC members. It does not contain specific instructions as to the nature of the report, just that the report be based on the model of State-of-Art policy in Europe, produced by the European Union Committee of Experts on Rare Diseases (EUCERD). The description of work may be amended, with strong justification, if this report is deemed unhelpful and if the resources could be better used elsewhere.

The report was initially planned to include discussion on projects currently funded and trends in research funding by IRDiRC members. However, due to delay in data collection, such analyses have not yet been carried out. Data collection will soon be completed and the next State-of-Play will not only review the literature but also contain analyses as originally intended.

Notwithstanding these criteria, the Sci Sec drafted a pilot report based on analysis of published literature, thus excluded press releases and unpublished projects. The report gives a broad overview of rare diseases research and genomic efforts specifically targeted to rare diseases, not on individual genes, nor disease studies. The Sci Sec tried to be as comprehensive as possible but could not rule out the possibility that some information may have been missed. Given its preliminary state, this report was only distributed to the members of Exec Comm but not to members of Sci Comms.

Comments received from Exec members included the fact that what is published is outdated by essence, and there were gaps in the coverage of important projects given the restrictive scope based on only published literature during a specified period of time. However, the Exec Comm also acknowledged that it is very difficult, if not impossible, to completely cover the whole area pertinent to rare disease research.

A report based solely on the literature cannot provide relevant information if the goal is to identify potential gaps where funding should be concentrated for future funding calls. However, it is the right media to provide the public at large with a sense of the trends in rare diseases research and to communicate appropriately about the dynamics in this sector.

Suggestions to improve the State-of-Play report:

- Circulation of the report for inclusion of members' contribution, esp. from Sci Comms
- Systematic crowdsourcing of information, although compliance may be poor
- Soliciting key players for comments and principle analysis of interests
- Inclusion of a statement on the limitations of the report
- Limit analysis to rare diseases with higher rates but currently without diagnostics

- Exec Comm and Sci Comms to generate questions they are searching answers to and that are forward looking, which the Sci Sec could try to address
- Members to create content on their websites related to activities with IRDiRC goals which could be easily forwarded to or mined by the Sci Sec during report preparation

Nonetheless, many members of the Exec Comm found the report to be useful and informative, and felt it should be made widely available to the community. The pilot report will be published on the IRDiRC website after minor changes are made in light of the discussion, with inclusion of a statement about its limitations. A modified methodology will be developed for the next State-of-Play report, with greater involvement from the Sci Comms.

Members of the Exec Comm interested in pursuing the idea of developing specific questions to be addressed by the State-of-Play report should give it further thought and these questions may be raised in the next Exec Comm meeting.

Meetings of the Sci Comms

The Sci Comms will be meeting face-to-face in Glasgow, Scotland, ahead of the ESHG 2015:

- Diagnostics Sci Comm: full day meeting on 5 June 2015
- Interdisciplinary Sci Comm: full day meeting on 5 June 2015
- Therapies Sci Comm: half day meeting on 5 June 2015 (afternoon)
- Joint meeting of all Sci Comms: half day meeting on 6 June 2015 (morning)

Exec Comm members who are in the area and/or will be attending the ESHG are encouraged to participate in the meetings of the Sci Comms.

Updates from the Scientific Committees

Diagnostics Sci Comm

The Diagnostics Sci Comm was represented by a committee member. Members of the Working Groups (WGs) reporting to the Diagnostics Sci Comm produced a number of tools (e.g. Matchmaker Exchange) and datasets (e.g. the European Variation Archive, EVA, implemented at the European Bioinformatics Institute, EBI), some of which are now ready to transition as Task Force projects or for application of the "IRDiRC Recommended" label.

In the upcoming meeting in Glasgow, the Diagnostics Sci Comm intends to review the work of this Sci Comm and its WGs to date, to perform a gap analysis for 2016-2020, to identify new

Task Forces for actionable projects, to discuss publication plans in disseminating the work of this Sci Comm and its WGs, and to develop/review the outline/draft of planned publications.

Interdisciplinary Sci Comm

The report of the Interdisciplinary Sci Comm was given *in absentia*. The members of this Sci Comm and its WGs were informed of IRDiRC restructuring, and nominations for the Task Forces were put forward. The Sci Comm is keen to establish a Task Force on "machine readable consent", a topic which is also worked on by an active group from GA4GH. A joint proposal will be submitted to the Exec Comm for consideration, with GA4GH taking the lead and the Sci Sec co-funding the workshop; such an alignment will avoid duplication of work.

The Interdisciplinary Sci Comm will discuss further activities, in particular objectives, Task Forces, as well as meetings and publications with the other two Sci Comms in Glasgow. It is also engaged in the discussion of scopes and objectives of additional Task Forces over the coming weeks with potential meeting dates in 2016.

Therapies Sci Comm

The report of the Therapies Sci Comm was given *in absentia*. The members of this Sci Comm and its WGs were also been informed of IRDiRC restructuring, and nominations for the Task Forces were put forward.

The Therapies Sci Comm plans to discuss the following in Glasgow: the update on Task Forces related to "patient related/reported outcome measures" and "small population clinical trials" in terms of their concept papers and workshop programmes; the Task Force on "repurposing" and information gathering for a background paper; and based on the Therapies Sci Comm Recommendation, the State-of-Play Report 2014, and an email sent in advance, to identify most critical next actions to speed up therapy development and increase success rates.

Task Force Formations

A number of key topics were selected for IRDiRC projects to ensure IRDiRC meets its objectives for the rare diseases community. IRDiRC, when in leading position, will produce a background document to ensure everyone attending the discussion has the same starting point and agrees on the main concept and objectives for discussion.

The agreed method for the preparation of a background paper/workshop:

- Sci Sec to prepare the first draft of background paper

- Draft to be sent to Task Force and Sci Comm members for review and input
- Sci Sec to contact Task Force members individually for discussion, focusing on topics deserving an agreement and on points of action
- Sci Sec to amend the draft based on discussions and input received
- Sci Sec to elaborate with Task Force members the draft agenda for the workshop
- Sci Sec to produce preparatory documents for the workshop

The composition of Task Forces is very important; targeted participants should be key players on the selected topics (e.g. representatives from PCORI and ISPOR for the Task Force on outcome measures), even if they may not necessarily be familiar with the rare disease community. This strategy ensures that work is not started from scratch and could additionally encourage these external players to champion specific development for rare diseases.

Previously defined steps of Task Force nomination and participant selection require some practical adjustments in general, as follows:

- Seeking of nominations from the Exec and Sci Comms
- Selecting core group representations during an Exec Comm meeting
- Contacting selected individuals/key organisations for agreement to participate, and in the case of key organisations, to name suitable delegates to participate
- Providing names of non-selected proposed participants to the core group
- Inviting newly-identified experts to complete the membership of Task Forces, up to 12 people maximum, balanced for representation of different regions and backgrounds

Members of the Sci Comms will not be nominated for Task Forces as they will already be consulted to review the documents and may be invited to participate in the workshops. Only one member from one Sci Comm, identified as closest to the field of each Task Force, will be appointed to ensure a liaison between Sci Comms and Task Forces.

The Exec Comm would like the Task Forces for the 2015 projects to be constituted by the time Glasgow meetings take place. The names of nominees not selected as core members will be provided to constitute Task Forces and for consideration as attendees of the workshops.

The Sci Sec reported that the first draft of the background paper to “patient related/reported outcome measures” is now ready, and will soon commence work on the background paper for “small population clinical trials”. The Exec Comm would like to provide input into these papers before the Sci Comm meetings in Glasgow, in a teleconference to be held in May.

Patient relevant/reported outcome measures

This workshop will explore whether, how and to what extent the patient relevant/reported outcome measures initiatives in common disease studies could be expanded to target rare disease research.

Representatives from the following organisations will be invited as core participants:

- Core Outcome Measures in Effectiveness Trials (COMET)
- Patient-Centered Outcomes Research Institute (PCORI)
- International Consortium for Health Outcome Measurement (ICHOM)
- International Society for Pharmacoeconomics and Outcomes Research (ISPOR)
- Genzyme

[Post meeting note: the National Institute for Health and Care Excellence (NICE) UK has been suggested as potential participating organisation for input from health economics' perspective.]

Small population clinical trials

The FDA and the EMA are, respectively, developing and updating guidelines on clinical trials in small populations. Concurrently, the European Commission has invested in three projects specifically on this theme. This workshop will bring together these key players to discuss adaptive designs, statistical methods and acceptability of new methods.

Representatives from the following organisations will be invited as core participants:

- EMA
- FDA
- Integrated Design and Analysis of Small Population Group Trials (IDEAL)
- Innovative Methodology for Small Populations Research (INSPIRE)
- Advances in Small Trials Design for Regulatory Innovation and Excellence (ASTERIX)
- Pfizer

The Sci Sec will ask the above to identify major US initiatives on small population clinical trials, similar to that of IDEAL/INSPIRE/ASTERIX, for inclusion in this Task Force.

Matchmaker Exchange

This is an ongoing project, in which its active participants organised a workshop in January 2015. Therefore, a pre-determined core group is in place to ensure optimal collaboration, although additional participants are needed to balance its representation (e.g. from industry

and public health, other geographical regions). No background paper will be produced by the Sci Sec for this workshop.

Computable consent form

This workshop aims to establish elements of computerised consent forms to create scope of consent and clear records of patient agreement. This workshop will be coordinated with and led by the Task Team of GA4GH on Consent. Additional information will be provided to IRDiRC in due course on how to move this project forward.

Other proposals for Task Forces

A number of projects have been put forward for consideration, including "repurposing and data mining" and "ontology interoperability and translation".

Repurposing and data mining

This project aims to bring together key players with mature projects to compare the different approaches to identify new targets and repurpose drugs through data mining. Development of drugs through repurposing has been paradoxically difficult for rare diseases, and it is also difficult to incentivise due to the small market size. Data mining efforts in particular have shown convincing outcomes with proof of concept. It is time to link SMEs and academic groups that are working in these fields. A call for nomination will be launched and the core group selection procedure may be carried out in the next Exec Comm teleconference.

Ontology interoperability and translation

This project was suggested by the Ontologies WG which would like to move forward as a Task Force as there are still a number of practical things to work on. The project concept is currently still under development, and a proposal will be put forward to the Exec Comm in due course.

Functional analysis

Functional analysis of identified variants is an area which falls outside many funding opportunities which, from a diagnostics point of view, could help the community to improve discovery rates through the use of model systems. There is also a lack of mechanism for a model systems group to be more useful to the rare disease research community. However, the

question was raised concerning whether this could be conducted via a (sustainable) matchmaking approach. There is a need to reflect on what a Task Force in this area could do.

ICHPT Update

Some of the ICHPT terms are currently undergoing further validation and the list will be finalised shortly. The IRDiRC website will host specific pages, which are currently under preparation, to display the core terminologies agreed upon and the links of these terms with other international terminologies. This will be launched as soon as possible. There is also a plan to write this up as an article for a peer-reviewed journal.

"IRDiRC Recommended"

The "IRDiRC Recommended" initiative was introduced to the rare disease community at the annual RD-Connect/Neuromics meeting in Palma de Majorca, Spain, in March. This is a form of seal of approval to identify useful tools and guidelines contributing to IRDiRC goals, and to raise the profiles of projects and collaborations that lack visibility.

A page on the IRDiRC website (http://www.irdirc.org/activities/irdirc-recommended/) set out to explain its status as quality indicator, its criteria for eligibility, and contains a simple application form to be downloaded and completed. Commercial products are not eligible for the label, although questions were raised on the eligibility of, for example, commercial resources that are free to academic and patient groups but not to professional groups. The Exec Comm decided not to create a subset of definitions at present, and will evaluate each application on a case-by-case basis.

The "IRDiRC Recommended" initiative has been promoted by some members of the Exec and Sci Comms, either at meetings or through direct contacts. Further promotion will be carried out to ensure wider knowledge of this initiative:

- Poster at upcoming meetings
- Announcement in an upcoming OrphaNews
- A journal article (in preparation)
- Direct mail to members and IRDiRC conference participants

The use of the IRDiRC logo

The standard IRDiRC logo has been found on display on the website of several projects. A discussion on the guideline for the use of IRDiRC logo is needed to define what constitutes an appropriate use of the logo. This will be an item on the agenda of the next Exec Comm meeting.

IRDiRC outreach with patient organisations

Given the expansion of IRDiRC in different geographical regions, the vision in engaging patient organisations is to open up access to patients in places where IRDiRC has active presence to bring up particular issues which may be addressed through its activities. IRDiRC is a global organisation and it is desirable as a goal that it engages with patients from all over the world.

However, as the focus of the consortium is on funding and acceleration of research, any expansion should be backed up with the reason why and how it contributes to the consortium. Patient organisations vary in size, structure and activities. The addition of patient groups with research focus would be logical and more valuable in advancing IRDiRC's mandate.

It was suggested that IRDiRC forms a WG to define an engagement strategy with patient organisations. Both the Chair and the Vice Chair will participate on this WG and as engagement depends on the people involved, IRDiRC recognises that these organisations are dynamic with interest that may wax and wane; IRDiRC will adapt accordingly to these changes. Additionally, EURORDIS is open to contacts from other patient organisations to discuss its work in IRDiRC.

IRDiRC engagement with funding bodies and industry partners

There was expression of interest from a number of public funders and industry that are on-going and require follow-ups. The Exec Comm would like to see more industry partners around the table. One way to engage them is to show the value and the benefits of being part of IRDiRC. Industry-based members would be good spokespersons to encourage potential partners to join IRDiRC and help clarify points in terms that are more industry and less academic speak. Having members from industry in IRDiRC Task Forces would be another step in offering value to their membership.

There are multiple points of query regarding IRDiRC membership, and while the Exec Comm members involved exchange as much information as possible, appropriate follow-up method is needed. Members should copy the Sci Sec in these correspondences.

Associated status to IRDiRC

IRDiRC receives, from time to time, requests of associated status from organisations that have some kind of rare disease-focus mandate. However, defining attributes for different categories of associated members etc is too onerous and detracts the work of the consortium. All similar future requests will be dealt with by the Chair, who is empowered to respond at his discretion with appropriately worded communication, and will be reported back to the Exec Comm.

IRDiRC website

The IRDiRC website will undergo a reorganisation shortly and changes will be made:

- To better publicised key documents (e.g. policies and guidelines)
- To promote current and ongoing activities (e.g. Task Forces and workshops)
- To facilitate global communication (e.g. simplified site with clear navigation)

The Chair expressed a wish to see a graphical representation of progress on diagnostics tool, similar to that of the therapy count, and a focus on IRDiRC activities. The Chair is happy to provide further feedback when the reorganised site is ready.

The Chair wished the IRDiRC website to include achievements from its private sector members, and the effort to profile them may produce tangible benefits for more companies to join the consortium. The contacts opportunity is high. One example of such success stories is the collaboration between Sanford Research and PTC Therapeutics following contacts made through IRDiRC Exec Comm meeting.

Authorship attribution

This is a topic brought up previously by Chairs of Sci Comms but absent at this meeting. In principle, the authors will be individuals who are involved in the preparation and the writing of papers. Any report on IRDiRC activities should be presented to the Exec Comm for review to ensure that it is a fair representation of those activities.

Support-IRDiRC survey

The performance review is a contractual requirement under the contract of Support-IRDiRC and the members of IRDiRC constitute the external assessment board of the Sci Sec. A short summary of the survey outcome was circulated and a number of issues were discussed.

Exec Comm meetings

The Exec Comm will hold its next face-to-face meetings in Montreal, Canada, on 11 September 2015. The following were proposed for the face-to-face meetings in 2016:

- The UK – spring 2016 (tbc)
- The Netherlands – autumn/fall 2016 (tbc)

The Exec Comm agreed that a joint Exec/Sci Comms meeting can be organised.

Some comments received touched upon a lack of discussion of strategic issues, and the Exec Comm agreed to step up on this theme and have more valuable meeting; strategic discussions will be made prominent in future face-to-face meetings.

Teleconferences

The Exec Comm will schedule teleconferences, whenever possible, 2 months in advance.

There are expressions of dissatisfaction with GoToMeeting, and at some workplaces, this web service is not accessible. Members who are aware of other more universal tools should forward the information to the Sci Sec. The Sci Sec will also explore for an alternative tool, and to determine the feasibility of putting in place teleconference facility during face-to-face meetings.

Scientific Secretariat

The Sci Sec will ensure that all requests received be implemented when feasible.

Any other business

Letter to NIH agencies members of IRDiRC

Members of the Exec Comm were informed that research projects funded by IRDiRC members are listed in the Orphanet database and consent from the principal investigators are required. This information is also relevant to all members to note.

The letter is a request to NIH to inform project leaders of the consent issue, and to add a text in their funding call to clearly specify this consent clause. Withdrawal from the list is allowed. The Sci Sec has not received any response to this request so far.

Model organism databases

The National Human Genome Research Institute (NHGRI) funds important databases on model organisms (e.g. FlyBase, WormBase) that are actively used (e.g. by investigators studying the function of rare disease variants). Given the increasing share of funds needed to adequately support these important data resources, in an environment of flat (at best) funding, NHGRI is unlikely to be able to continue to support these resources as it has done in the past. NHGRI and others are studying options for sustaining their support.

Satellite meeting at the ESHG

The Office of Population Health Genomics, Department of Health Western Australia is organising a satellite meeting at the ESHG in Glasgow entitled "Whole-Population Preconception Carrier Screening" on 10 June 2015. For more information, please consult http://www.genomics.health.wa.gov.au/home/workshop.cfm

Innovation and Biomarkers in Cancer Drug Development

EORTC, together with National Cancer Institute and EMA, are organising a joint meeting in Brussels on 3-4 December 2015. More information is available http://www.eortc.org/ibcd2015/

Agenda of Exec Comm teleconference in May

The following topics were proposed for the next Exec Comm teleconference:

- State-of-Play report: specific questions to be addressed
- Feedback of background papers to "patient relevant/reported outcome measures" and "small population clinical trials"
- Selection of core participants to the Task Force for "Repurposing and Data Mining"
- "IRDiRC Recommended" review (standing item on all Exec Comm meeting)
- Guideline to the use of IRDiRC logo

[Post meeting note: Members of the Exec Comm who would like to propose item(s) to the agenda may do so by emailing the Chair of the Exec Comm, copying the Sci Sec.]

Acknowledgements to the host

The Exec Comm is very grateful to the Institute of Health Carlos III for hosting the meeting. The Chair and the IRDiRC Secretariat wish to thank the Institute of Health Carlos III for their generosity and hospitality.

Annex - List of participants

Members	**Representative**
Western Australian Department of Health, Australia	Hugh Dawkins
European Organisation for Treatment & Research on Cancer, EORTC	Denis Lacombe
Canadian Institutes of Health Research, Canada	Paul Lasko
BGI, China	Cong Yu
E-RARE-2 Consortium, EU & ANR, France	Natalia Martin
European Commission, DG Research and Innovation, EU	Iiro Eerola
European Commission, DG Health and Consumer Protection, EU	Stefan Schreck
Academy of Finland, Finland	Heikki Vilen
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Fondation Maladies Rares, France	Nicolas Lévy
Telethon Foundation, Italy	Lucia Monaco
The Netherlands Organisation for Health Research and Development	Janna de Boer
Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández, Teresa Chavarría Giménez, Ignacio Baanante
National Institute for Health Research, UK	Willem Ouwehand
PTC Therapeutics, USA	Diane Goetz
Sanford Research, USA	David Pearce

Invited Patient Advocacy Groups	
EURORDIS, Europe	Béatrice de Montleau
Genetic Alliance, USA	James O'Leary

Scientific Committees	
Diagnostics	Xavier Estivill

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Ségolène Aymé, Lilian Lau, Antonia Mills

Apologies

Members	**Representative**
Genome Canada	Pierre Meulien
WuXi AppTec Co. Ltd., China	Mao Mao
Chinese Rare Diseases Research Consortium, China	Qing Wang
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	Albert Seymour
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, Italy	Fabriozio Oleari
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Prosensa, The Netherlands	Luc Dochez
Korea National Institute of Health, South Korea	Hyun-Young Park
Food and Drug Administration, USA	Katherine Needleman
Genzyme, USA	Carlo Incerti
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute (NCI), USA	Edward Trimble
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin
National Eye Institute (NEI), USA	Santa Tumminia
National Human Genome Research Institute (NHGRI), USA	Jeffery Schloss
National Institute of Arthritis and Muskoloskeletal and Skin Diseases (NIAMS), USA	Stephen Katz
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke (NINDS), USA	Danilo Tagle
NKT Therapeutics, USA	Robert Mashal
Office of Rare Diseases, USA	Pamela McInnes

Scientific Committees	
Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam

Invited Patient Advocacy Groups	
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall

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