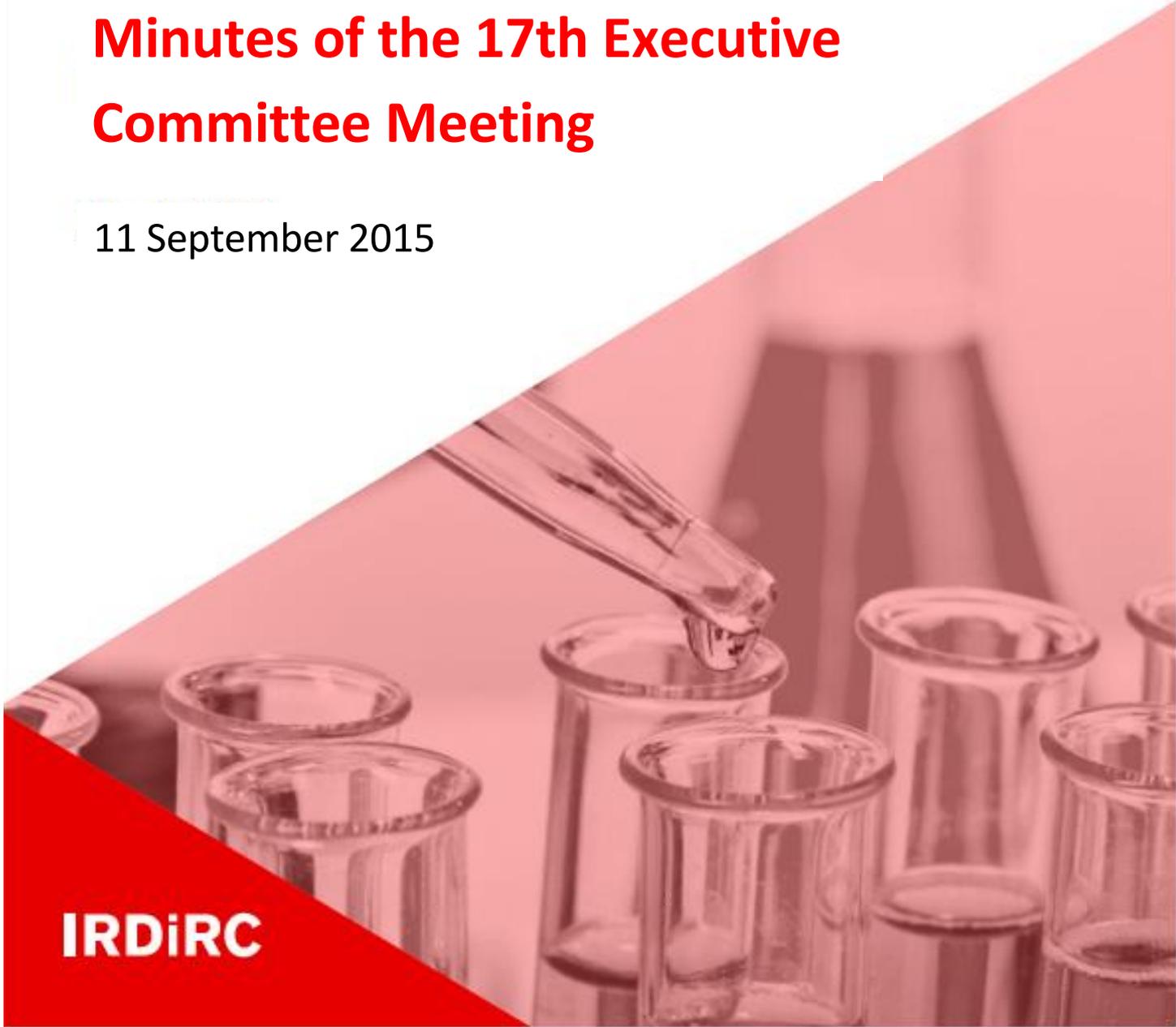




**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 17th Executive
Committee Meeting**

11 September 2015



IRDIRC

EXECUTIVE SUMMARY

- ▶ The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 11 September 2015 in Montreal, Canada. The seventeenth meeting of the Exec Comm was attended by 25 participants, representing 18 member organisations, the three Scientific Committees (Sci Comms) and the Scientific Secretariat (Sci Sec).
- ▶ This was the first meeting attended by new executive members from Pfizer and Japan Agency for Medical Research and Development (AMED); the new members presented in detail their activities. Further to this, it was also announced that the National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN) from Japan have also joined IRDiRC. Membership discussions with other industry partners are currently underway, facilitated by help from the Chairs of the Interdisciplinary (ISC) and Therapies Sci Comms (TSC).
- ▶ The voluntary membership fee account: expenditures thus far had been travel support of patient organisation representatives to Exec Comm meetings. IRDiRC also agreed to a sponsorship commitment for 6-8 speakers to travel to RE(ACT) Congress in Barcelona, March 2016. Members are asked to suggest uses of membership fees collected in the next teleconference of the Exec Comm.
- ▶ The IRDiRC governance was re-discussed to as to clarify the role played by the Coordinator of Sci Sec in relation to the Exec Comm. The Exec Comm agreed to the following changes to the Governance document: *“The Scientific Secretariat is represented at Executive Committee meetings, except in matters in which it has, or could reasonably be perceived to have, a conflict of interest. The Scientific Secretariat representative does not have voting rights at meetings of the Executive Committee.”*
- ▶ The organogram in the Governance document requires further consideration, with the role and responsibility of each body in the Governance clearly stated and reflects the evolution of IRDiRC’s operation not yet captured, for example, the changing role of Sci Comms following the completion of the actions of Working Groups and transition to the actions under the new Task Forces. A revised draft Governance document, with a new organogram, will be prepared and presented for discussion at the next Exec Comm meeting.
- ▶ The current term for IRDiRC’s Chair ends in December 2015, and the next term will run from January 2016 to December 2018. IRDiRC Chair “could be nominated for re-election to a further term but not to exceed two consecutive terms.” The Exec Comm will be contacted at the end of September to submit candidate names, which will be put through an electronic vote. The handover will take place at the next face-to-face Exec Comm meeting.
- ▶ An Operating Committee was established. It consists of Chair and Vice-Chair of Exec Comm, the Chairs of the Sci Comms and the Sci Sec. The Operating Committee has the mandate to prepare and advance IRDiRC activities.

- ▶ A State of Play Report for the period July 2014 to June 2015 was produced based on published articles and press releases. Responses received following dissemination of the first draft were very positive. It is ready for wider dissemination after suggested minor changes are incorporated.
- ▶ The Chairs of Diagnostics Sci Comm (DSC) and ISC will be renewed for another three years. The Chairs of Sci Comms will also identify the gaps in each of their committees (e.g. lack of representatives from regulatory agencies in the TSC) and inform the Exec Comm so strategic nominations can be sought.
- ▶ The recommendations drafted by the TSC for consideration by the Exec Comm were reviewed. Sections 1 and 2 of the document were considered appropriate for a wider dissemination as a document recommended by the Sci Comms.
- ▶ The current status of the Task Forces (TFs) activities was reviewed and judged promising. TFs are entitled to publish their conclusions, while Sci Comms identify points that can be recommended as potential IRDiRC policies. All documents/recommendations produced will be discussed and synthesised by the Sci Comms, publicised as documents/recommendations from the Sci Comms and TFs, and be opened to public consultations.
- ▶ Four proposals were put forward for consideration of formation of future IRDiRC Task Forces: “Participant Unique Identifiers”; “Case-based Matching for Gene Discovery, i.e. MMEv2”; “Strategies for Diagnostic Translation”; and “Patient Engagement in Rare Disease Research and Health Product Development”. Further developments are needed prior to decisions on their implementation.
- ▶ Two resources have been approved for “IRDiRC Recommended” to date: the International Charter of Principles for Sharing Bio-Specimens and Data, and Orphanet. Some agencies/institutes indicated that “IRDiRC Recommended” approval by the Exec Comm endangers the impartiality of their organisations. The Exec Comm decided to delegate this initiative to the Sci Comms.
- ▶ The Exec Comm was presented with two options for the next face-to-face meeting, and decided it will take place on 14-15 March 2016 in Lyon, hosted by AFM-Téléthon. The Sci Comm meetings will also take place in Lyon on 14 March 2016.
- ▶ The 13th ICHG will take place in Kyoto, Japan, on 3-7 April 2016. A 90-minute session in the conference has been allocated to IRDiRC on 6 April. The Exec Comm also agreed be Associate Partner to the ECRD 2016 which will take place in Edinburgh, Scotland, on 26-28 May 2016.
- ▶ The possibility of organisation of a third IRDiRC Conference in 2016 or 2017, focusing on therapy development, was discussed but no conclusion was reached. The Chair of the Exec Comm called for interested members to get involved in the organising committee.

REPORT

1. Chair's activity report

The Chair of the Exec Comm welcomed all in attendance of this meeting to Montreal, Canada. The Chair extended a warm welcome to the new executive members from Pfizer and Japan Agency for Medical Research and Development (AMED) to their first meeting, and informed the Exec Comm that the National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN) – also from Japan – have also joined IRDiRC. Membership discussions with other industry partners are currently underway, facilitated by help from the Chairs of the ISC and TSC.

The Chair of the Exec Comm most recently represented IRDiRC at the Asia-Pacific Undiagnosed Disease Programme (UDP) meeting in Perth, Australia, in August 2015, and will next attend the Global Genomic Medicine Collaborative (G2MC) meeting in Singapore in November 2015 as well as the International Undiagnosed Disease Program meeting in Vienna, Austria, in February 2016 on behalf of IRDiRC.

The Chair of the Exec Comm also provided an update on the voluntary membership fee account. Expenditures thus far had been travel support of patient organisation representatives to Exec Comm meetings. IRDiRC also agreed to a sponsorship commitment for 6-8 speakers to travel to RE(AC)T Congress in Barcelona, 9-12 March 2016.

By contributing to conferences, IRDiRC brings together people in rare diseases research and builds collaboration, while increasing its visibility at an international level. At the present rate of contribution, IRDiRC has sufficient funding to support 3-4 meetings per year. Alternatively, the membership fee could go towards organisation of an IRDiRC conference. Members were also asked to suggest uses of membership fees in the next teleconference of the Exec Comm. Letters will be sent out to members asking for their 2016 contribution of voluntary membership fee by the end of September.

2. Presentation: Pfizer

Katherine Beaverson, Patient Advocacy Lead of Pfizer, gave an overview presentation of Pfizer's efforts in the field of rare diseases. Pfizer owns a portfolio with over 20 products with orphan designation for haematology, neurology, metabolic, pulmonology, oncology and other conditions; it ranked number 3 in terms of orphan drug sales in 2014. Pfizer is strategically focused, and its high priority disease areas include sickle cell, haemophilia, Duchenne muscular dystrophy, cystic fibrosis, pulmonary hypertension, Gaucher and growth hormone deficiency. A snapshot of pipeline showed robust development programmes from preclinical stage to commercialisation. In its clinical phase, therapy of sickle cell is currently most advanced, with Rivipansel IV in Phase III trial and PDE9i in Phase I trial.

Extreme heterogeneity, global and scattered expertise, access to breaking science and new models for drug developments are the challenges faced in rare disease. Pfizer established a Rare Disease Consortium to build academic network with six institutions in the UK on the principles of complementary

skill sets, and sharing of both risk and rewards. Through this initiative, academic experts are given access to drug development capability of Pfizer and business models are built based on milestones.

An emerging interest for Pfizer lies in gene therapy, and it applies two-prong approach to treat rare diseases: disease management in the near term and disease correction in the long term. Pfizer combines the following strategies to maximise success: rapid early entry “buy” approach, and establish “build” know-how, IP estate and next generation assets, an effort led by Professor Michael Linden. Major challenges in gene therapy include vector design, manufacturing and standardisation, and it is hoped through various initiatives in place, Pfizer could create sustainable pipeline with focus on collaborations, best and brightest early science, scalability, and ultimately feed into commercialisation.

3. Presentation: AMED

Shigeki Kuzuhara from the Japan Agency for Medical Research and Development (AMED) introduced this newly-founded agency and rare diseases research projects in Japan. AMED was formed on 1 April 2015 through integration of medical research and development funding from 3 different ministries: Ministry of Economy, Trade and Industry (METI), Ministry of Education, Culture, Sports, Science and Technology (MEXT), and Ministry of Health, Labour and Welfare (MHLW). AMED aims to promote coherent R&D and application from basic science to clinical uses, comprehensively and effectively implement medical R&D, and supports and trains young researchers while improving their work conditions. Ultimately, its goal is to fast track medical R&D to directly benefits the people, not only “life” as biological existence but also quality of life.

Dr Kuzuhara is the Program Director of the “Nan-Byo” Project – “Nan-Byo” being the Japanese medical term referring to rare and intractable diseases – who is assisted by 3 Program Officers and overseeing more than 150 projects, which include basic research, research on induced pluripotent (iPS) cells, clinical trials, and registries and biobanks.

“Nan-Byo” was defined as diseases that are rare (less than 0.1% of population), with unclarified causes and pathogenesis, lacks effective treatment, and has slow progression without recovery. “Nan-Byo” leads to severe disability in patients while thrusting heavy, long-term burden on their families and caregivers. Japan’s “Nan-Byo” Program was founded in 1972 to accelerate research to clarify the cause of disease and to support patients medically and economically. In January 2015, the “Nan-Byo” Act became effective. As of 1 July 2015, 306 diseases were included in AMED’s programme, estimated to affect approximately 1.5M registered patients. An estimated budget of US\$ 1.53B (of which US\$ 765M from the Government) is needed for 2015 for medical support and care for these patients.

A new programme called “Initiative on Rare and Undiagnosed Diseases” (IRUD) was established in 2015; it is an all-Japan consortium network of patients, doctors and researchers to provide diagnosis and treatment of undiagnosed diseases through genome analysis, in addition to clinical examination and laboratory and imaging studies. AMED has identified the need for global collaborations, and hopes its collaboration with IRDiRC would help forwards the goals of both AMED and IRDiRC and help individuals

suffering from rare diseases. To further strengthen Japan's relationship with IRDiRC, the National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN) is also joining IRDiRC.

4. Revision of IRDiRC Governance

The nomination in November 2014 of the Coordinator of Sci Sec as a member of the Exec Comm created confusion, and a proposal was made to reverse that decision.

The Exec Comm agreed to the following changes to the Governance document:

- ▶ Page 3: removal of mention of Coordinator of Sci Sec in the composition of the Exec Comm
- ▶ Page 4: with the following text
“The Scientific Secretariat is represented at Executive Committee meetings, except in matters in which it has, or could reasonably be perceived to have, a conflict of interest. The Scientific Secretariat representative does not have voting rights at meetings of the Executive Committee.”

The organogram in the Governance document requires further consideration, with the role and responsibility of each body in the Governance clearly stated and reflects the evolution of IRDiRC's operation not yet captured, for example, the changing role of Sci Comms following the completion of the actions of Working Groups and transition to the actions under the new Task Forces.

The appropriateness of voting rights of the Chairs of Sci Comm was also raised, given their advisory role to the Exec Comm. This issue will be discussed in the next Exec Comm meeting.

A revised draft Governance document, with a new organogram, will also be prepared by the Operating Committee and presented for discussion at the next Exec Comm meeting.

5. IRDiRC Chairmanship

The current term for IRDiRC's Chair ends in December 2015, and the next term will run from January 2016 to December 2018. The Governance document states “[t]he Chair is elected for a maximum period of three years.” The official handover of the chairmanship will take place at the next face-to-face Exec Comm meeting.

The Exec Comm was asked to consider if the current Chair may run for a re-election as this was not explicitly stated in the Governance document. Members agreed that re-election is an option. IRDiRC Chair “could be nominated for re-election to a further term but not to exceed two consecutive terms.” The Governance document will be updated to reflect this decision, and to clarify that the Vice Chair is not automatically Chair-elect.

The Exec Comm will be contacted at the end of September to submit candidate names. The Chair must be elected from among the members of the Exec Comm, and self-nomination is allowed. A short paragraph should be provided to give a rationale to this nomination, on what this nominee will bring to

IRDIRC. All eligible nominations received will be circulated out-of-session and the election will be achieved through an electronic vote.

6. Operating Committee

An Operating Committee was established. It consists of Chair and Vice-Chair of Exec Comm, the Chairs of the Sci Comms and the Sci Sec. This committee will conduct regular teleconference to prepare and advance IRDiRC activities. [*Post-meeting note: the Operating Committee will meet every month on the second Thursday for the next six months, after which the frequency may be revised.*]

7. State of Play Report 2015

8.1 State of Play Report – Part 1

A State of Play Report for the period July 2014 to June 2015 was produced based on published articles and press releases. It contains major policy decisions, results of previous major initiatives and overview of trends in research, and excludes initiatives to improve the organisation of healthcare system and public health in general. Responses received from Exec and Sci Comm members following dissemination of the first draft were very positive.

In response to feedback received:

- ▶ The report is EU/US-centric as it was prepared based on publication in English. Moreover, it is difficult to identify initiatives and trends in other regions due to language barrier and lack of contact points to obtain information on major changes.
- ▶ Following internal discussions, it was decided to not cover breakthroughs in diagnostics as, while beneficial, evolution of technology in genomics is not rare disease-specific. Additionally, it is also difficult to identify the most relevant publications to indicate breakthroughs and changes due to overwhelming amount of articles in this area.

Additional suggestions:

- ▶ Cross-referencing relevant sections (where applicable)
- ▶ Addition of an Executive Summary

The State of Play Report should be used more extensively, among others:

- ▶ Identify new Task Forces (TFs) based on trends that IRDiRC could address
- ▶ Integration into members' activities (e.g. adoption of policies, decision on projects to fund)
- ▶ Dissemination through members' networks and/or websites

[*Post-meeting note: The State of Play Report 2015 based on publications is now available on the IRDiRC's website.*]

8.2 State of Play Report – Part 2

An analysis of the areas covered by funded projects and a review of indicators of success were among other items requested for inclusion in the State of Play Report. A pilot report was produced, focused on research projects funded by the European Commission, E-RARE, and NIH members of IRDiRC.

Members were interested to see inclusion of data from other funding agencies, while acknowledging this would require active involvement from funding agencies. Moreover, information and updates from the roundtable discussion at the start of Exec Comm would be an interesting component to include.

However, caution was made on the utility of this report, considering the extensive work required. An overall funding trend analysis would require obtaining information not only from IRDiRC members but broader coverage of funding agencies on the whole.

A useful output could be a commentary on what IRDiRC is actively engaged in as a consortium, what is its collective resource (not granular information), and how the resource is invested. This enables reporting success of IRDiRC as initiative, but it requires all members to issue a short document on its investment and outcomes, based on a harmonised structure so same types of information are provided.

A technical comment was made: it is more appropriate to categorise animal model creation/study under “Basic research” instead of “Pre-clinical research”.

8. Scientific Committee Updates

9.1 Report from the Chair of the Diagnostics Sci Comm (DSC)

When the DSC met in Glasgow, it identified a number of Task Forces for future implementation and started drafting a commentary on the work of the DSC and the ISC towards IRDiRC’s diagnostic goal. The commentary has been circulated to a few individuals and contained about 3,000 words, with 3-4 images, and aimed to be co-published in American Journal of Human Genetics and European Journal of Human Genetics. The DSC/ISC were trying to build a pie chart to show contribution of research funding in terms of origin (e.g. public, industry, private via donation), geography (e.g. US, Europe, Asia), and amount. However, such information was not easily obtained, and industry members cannot disclose their real numbers. A chart may be built based on solely public funding instead.

9.2 Report from the Chair of the Interdisciplinary Sci Comm (ISC)

Since the ISC meeting in Glasgow, many members have been actively engaged in reviewing “IRDiRC Recommended” applications, developing ideas for future Task Forces, co-writing a commentary with the DSC, assisting partners’ recruitment to IRDiRC, and expressing high interest in contributing to the programming of an upcoming IRDiRC conference if one is planned. The Chair of the ISC also urged members to keep in mind the big picture of what IRDiRC wishes to achieve, with clear outcomes and products, and work towards a growing consortium.

9.3 Report from the Chair of the Therapies Sci Comm (TSC)

During the TSC meeting in Glasgow, members recognised the acceleration of development of orphan drugs in the EU and the USA, so the target of 200 new therapies will be reached by the end of 2016. This achievement could be used as leverage to put forward new models of development based on IRDiRC recommendation and beyond, through areas identified and to work on by Task Forces. The next IRDiRC conference can also be shaped around these approaches. The members of TSC also requested a count of therapies approved for rare diseases but without orphan designation. A recommendation was also developed by the TSC a few months ago, and a paper is currently being drafted based on the recommendation which will be circulated before submission for feedback.

9.4 Support of the Sci Comms by the Sci Sec

The Sci Sec will organise 1 face-to-face meeting per year for the Sci Comms, with 2-3 teleconferences in between so members could discuss and synthesise works; the aim is for more productive meetings. The Sci Sec will also assist Sci Comms in disseminating their outputs and coordinate reviews of “IRDiRC Recommended”.

Each Sci Comm has different needs so each committee should discuss individually how they wish to be supported by the Sci Sec.

9.5 Sci Comm memberships/renewal

Chairs of Sci Comms reported the need for engaged committee members and funders should think strategically when nominating an individual to the Sci Comm – someone who could commit, act and drive the activities (e.g. develop Task Forces) – and potentially grow into the role of Chairs.

A committee needs a good mix of experienced senior scientists and forward-looking actors. Chairs of Sci Comm could assist Exec Comm when looking for new members to participate in the committees by identifying the profiles needed and making recommendations. To attract the right individuals, it was also suggested that information on workload and expertise required should be provided.

The Chairs of DSC and ISC will be renewed for another three years. They will also consult members up for renewal if they wish to continue their participation. Other decisions:

- ▶ DSC: BGI will be contacted by the Sci Sec to suggest a substitute
- ▶ ISC: A replacement of patient voice is needed
- ▶ TSC: Ellen Welch will be removed from the TSC as she now serves in the Exec Comm

The Chairs of Sci Comm will also identify the gaps in each of their committees (e.g. lack of representatives from regulatory agencies in the TSC) and inform the Exec Comm so nominations can be sought. A list consisting of current Sci Comm members and its nominating organisation will be prepared and provided so that Exec Comm members who have not yet nominated Sci Comm members could contribute to the process.

[Post-meeting note: IRDiRC Governance states “The mandate of a Scientific Committee member can be terminated for reason of non-participation at the discretion of the Chair of the Scientific Committee. In that case, in consultation with the Chair of the Scientific Committee, the Executive Committee will decide whether a replacement appointment is necessary, and a call for nomination will be made.”]

9.6 TSC Recommendations to the Exec Comm

Last year, the TSC drafted a recommendation document for consideration by the Exec Comm but the final approval was delayed. The core of the document lay in sections 1 and 2, and they pertain to policy recommendations to funding agencies. The document was also intended to be basis of drafting a paper for publication.

The Exec Comm discussed the appropriateness of the committee to endorse this document and disseminate it as an IRDiRC recommendation, as the language may be perceived to obligate member organisations beyond what was intended. Furthermore, to incorporate changes in the text, should each organisation be consulted independently, the process would be onerous and time consuming.

The Exec Comm agreed on the dissemination of the TSC Recommendation as is, coming from the TSC which is a group of experts and scientists. In response to the recommendation, IRDiRC will modify its policy. A sub-committee of the Exec Comm is needed to rewrite the policy document; interested members were asked to contact the Sci Sec.

9. Updates of current Task Forces (TFs)

9.1 Matchmaker Exchange (MME)

Two workshops will be held in Baltimore, USA, on 6 and 7 October 2015.

- ▶ 6 Oct: invited workshop of 3 sessions, about 40 people, output is position paper on what the experts think needs to be done in expanding MME’s functionality
- ▶ 7 Oct: community engagement session, 150-200 attendees, consists of short presentations and use cases, and for community to ask questions

A special MME issue of Human Mutation is in print, and 1,000 copies have been ordered for dissemination at the ASHG and other future events. The articles contained in the issue include introduction of MME, manuscripts from each of the matchmaking databases, and examples of use with reported gene discovery.

9.2 Patient-Centred Outcome Measures (PCOM)

Of the IRDiRC-managed Task Forces, the PCOM TF is the most advanced in its work. The Steering Committee members met once as a group, followed by one-on-one discussions with the Sci Sec to put through their ideas so the background documents can be modified and improved. Another group

teleconference will take place to finalise the agenda of the workshop, which will take place on 30 November 2015 in Paris.

The one-on-one discussion model has been a productive strategy even though it requires more preparative work in order to have effective exchanges. The purpose of the workshop was identified and the specific objectives were defined; these will also be validated in the upcoming group teleconference. The groups working on patient reported outcomes (PROs) aim to keep their work international and be culturally sensitive, and there is an opportunity for IRDiRC to build on the PCOM at international level. There may be a need for follow-up work after this TF holds its upcoming workshop.

9.3 Small Population Clinical Trials (SPCT)

This is a joint TF with the EMA, of which the Steering Committee members will soon meet for the first group teleconference before one-on-one discussions take place. Following an initial call with the EMA, it was envisaged that a workshop will take place in the Q1 of 2016, hosted by the EMA, in London, UK.

A FDA guideline on common issues in drug development for rare diseases was recently published for consultation, so it is timely to address this topic and combine it with knowledge in Europe. At present, however, the Steering Committee of the SPCT TF consists of mainly European experts. The Exec Comm was asked to nominate experts in the USA and in the industry to participate in this TF.

9.4 Data Mining / Repurposing (DMR)

A Steering Committee for the DMR TF has been assembled and the Sci Sec has commenced work on its background document. Once the draft background document is ready, it will be circulated to the Steering Committee and these experts' feedback will be sought through group and one-on-one teleconferences. The Exec Comm was encouraged to nominate additional experts to this TF.

9.5 Machine Readable Consent (MRC)

This is an initiative initially set up by GA4GH's MRC Task Team, to which IRDiRC joined in and provide support for the experts to meet and work. A teleconference of this TF took place recently, and a workshop will take place in Paris on 9-10 November 2015.

9.6 Financial support of the Task Forces

The workshops are currently supported financially by the Sci Sec, which is funded through a contract awarded by the European Commission. The resource available is therefore limited. Additional support can be made available through the membership fee.

9.7 Task Force outputs

The workshops are working workshops, and the attendees will be invited participants based on their expertise in relevant topics. Task Forces are entitled to publish their conclusions, while Sci Comms identify points that can be recommended as potential IRDiRC policies.

All documents/recommendations produced will be discussed and synthesised by the Sci Comms, publicised as documents/recommendations from the Sci Comms and Task Forces, and be opened to public consultations.

10. Proposals for future IRDiRC Task Forces

Four proposals were put forward for consideration of formation of future IRDiRC Task Forces. The preparatory documents related to these proposals are available on the private website.

10.1 Participant Unique Identifiers

The objective of this Task Force is to develop guiding principles for generation of participant-specific identifiers that enable data from the same individual to be connected across multiple projects without revealing the participant's identity. This project is challenging from legal and data protection aspects, and coordination across different jurisdiction is also non-trivial. However, if clear recommendations can be given on ways to ensure data interoperability, it will be a step forward. Additionally, the production of a state-of-play can be another way to push this issue in the right direction. A report on this issue is currently in preparation by Mats Hansson for RD-Connect, and parts of its preliminary results feed into this proposal. The Chair of the ISC will talk to representative(s) from GA4GH and explore the possibility of a collaborative effort, before identifying resource implication.

10.2 Case-based Matching for Gene Discovery, i.e. Matchmaker Exchange v2

The objective of this TF is to advance the efforts of the MME initiative to develop a sustainable solution for case-based matching for rare disease gene discovery. The Chair of the DSC/MME TF identified three phases of MME which do not transition but each has different use case.

- ▶ MMEv1: "2-sided hypothesis", currently implemented, matching gene to gene
- ▶ MMEv2: "single-sided hypothesis", e.g. find matches based on candidate gene
- ▶ MMEv3: "hypothesis free"

No immediate action was asked on this proposal, as it will be developed further following the MME workshop in Baltimore on 6 October 2015.

10.3 Strategies for Diagnostic Translation

The objective of this TF is to formulate recommendations for jurisdictions on strategic approach to the diagnosis of rare genetic diseases using next generation sequencing-based platform. Members of the DSC expressed interest in learning from different jurisdictions in relation to diagnostic translation, such as how health organisations make economic deliberations, and build cases that could aid other, small jurisdictions which do not have the same health technology assessment (HTA) resource. The TF will focus on process which can be adapted and it is a service product. This proposal will be further developed.

10.4 Patient Engagement in Rare Disease Research and Health Product Development

The objective of this TF is to promote the engagement of patients in rare disease research and health product development (diagnostic tools and therapies) based on sound, rational and good practices. A recent study by KPMG, and a trend noted in the State of Play Report 2015, show integration of patient involvement in healthcare industry. Leading academic teams and leaders of projects of good practices can come together with initiative such as Clinical Trials Transformation Initiative (CTTI), which has mapped landscape of engagement that many groups have adopted and mapped against, could come together in this TF and identify good practices to be IRDiRC recommendations. This proposal will be further developed.

11. “IRDiRC Recommended”

Two resources have been approved for “IRDiRC Recommended” to date – the International Charter of Principles for Sharing Bio-Specimens and Data, and Orphanet. However, a number of issues came to attention during the review process, and a guideline is needed to clearly specify inclusion/exclusion criteria so only eligible resources are applying and not to take up time unnecessarily from project investigators.

Some agencies/institutes indicated “IRDiRC Recommended” approval by the Exec Comm endangers the impartiality of their organisations. The Exec Comm decided to delegate this initiative, which will from now on be recommended by the Sci Comm; the Exec Comm will not be involved in this process but will be kept informed by the Sci Comms of its progress.

The Chairs of Sci Comms will work with Sci Sec to revise the scope of “IRDiRC Recommended” procedure and eligibility. Sci Sec will also ensure the review process is independent and check that reviewers have no conflict of interest. The revised documents and the way to decide on the seven applications awaiting outcomes will be discussed in the next Operating Committee meeting.

12. Upcoming meetings

12.1 Exec Comm Spring 2016 face-to-face meeting

The Exec Comm was presented with two options for the next face-to-face meeting, which will take place in March 2016. The majority voted for Lyon, and this meeting will be hosted by AFM-Téléthon.

12.2 Sci Comm 2016 face-to-face meetings

The Sci Comm wished to have individual meetings, followed by a joint meeting. A suggestion was made to also couple it to an Exec Comm meeting, to create tighter liaison between the Exec Comm and the Sci Comms. The Sci Comm meetings will therefore also take place in Lyon. The Sci Sec will work with AFM-Téléthon to organise the meetings and secure require rooms.

Post meeting note: the order of meetings following consultation with Operating Committee

- ▶ *Sci Comms: Monday 14 March, morning*
- ▶ *Joint Sci Comms: Monday 14 March, afternoon (Exec Comm welcomed to join)*
- ▶ *Exec Comm: Tuesday 15 March, all day*

12.3 IRDiRC partnership/sponsorship of RE(ACT) Congress 2016

IRDiRC has agreed to partner with E-Rare3 Consortium and Blackswan Foundation (Switzerland) in the organisation of the RE(ACT) Congress 2016, which will be held on 9-12 March 2016 in Barcelona, Spain. IRDiRC also agreed to sponsorship commitment for 6-8 speakers to travel to RE(ACT) Congress using funds collected from the voluntary membership fee. These speakers have been nominated by the Chairs of the Sci Comms.

12.4 IRDiRC session at the 13th International Congress of Human Genetics (ICHG 2016)

The 13th ICHG will take place in Kyoto, Japan, on 3-7 April 2016. A session in the conference has been allocated to IRDiRC, initially on 5 April but now changed to 6 April.

A timeslot of 180-minute has been made available to IRDiRC and GA4GH, which can run as two separate 90-minute sessions or a combined 180-minute session. IRDiRC opted for two separate sessions, one after another, and will coordinate with GA4GH to work out a complementary program and identify invited speakers.

Professor Tsuji, a member of the ICHG 2016 Organising Committee suggested:

- ▶ Introduction of IRDiRC: its goals, activities and outcomes
- ▶ Overviews and prospects of rare diseases research programs
- ▶ AMED's Initiative on Rare and Undiagnosed Diseases (IRUD) Program
- ▶ Matchmaker Exchange: collaboration between IRDiRC and GA4GH

12.5 Associate Partnership of European Conference on Rare Diseases and Orphan Products (ECRD) 2016

The ECRD 2016 will take place in Edinburgh, Scotland, on 26-28 May 2016. IRDiRC has been invited to be its Associate Partner, contributing the following:

- ▶ Provide logo for dedicated conference website page
- ▶ Provide topic, speaker and/or session chair recommendations to Programme Committee
- ▶ Promote registrations to the conference
- ▶ Disseminate the Call for Posters

The Exec Comm agreed to this partnership, and accepted the suggested session on “Maintaining Europe’s position in a global collaborative research environment”.

12.6 Acknowledging IRDiRC at meetings

IRDiRC is getting listed in talks in symposium and congresses, but an acknowledgement policy has not yet been developed. This point will be discussed in a future meeting of the Exec Comm.

13. Future IRDiRC conference

IRDiRC had organised two successful conferences in the past and there has been calls for another IRDiRC conference. As the goal of 200 therapies will likely be reached at the end of 2016, the occasion could be a good opportunity to organise a meeting which focuses on drug development and its success stories, and to engage industry partners who help drive therapy development forward.

IRDiRC needs to deliberate in terms of budget and workload in organising a meeting/conference. IRDiRC could consider partnering with a group such as FasterCures, which is dynamic with positive profile, and bring the conference to the USA.

Some funding is available through the IRDiRC membership account, but additional sponsorship will be needed. Additionally, the Sci Sec does not have the scope to organise this conference, so resources to form the organising committee is required.

Any members of the Exec Comm interested in getting involved in the organising committee were asked to contact the Chair.

14. Any other business

- ▶ Anneliene Jonker joined the Sci Sec on 1 September 2015, replacing Antonia Mills.
- ▶ IRDiRC website is currently undergoing another revision.
- ▶ IRDiRC is now present on Twitter (@IRDiRC).

- ▶ The page of International Consortium of Human Phenotype Terminologies is live and the table of terminologies is available for download at www.irdirc.org/ichpt.

15. Next steps and actions

- ▶ Identify activities and suggest uses of membership fee
- ▶ Revise IRDiRC Governance and organogram
- ▶ Submit nominations for the position of chairmanship
- ▶ Organise a private electronic vote for the next Chair
- ▶ Get involved as IRDiRC conference organising committee
- ▶ Prepare a list of current Sci Comm members and nominating organisations
- ▶ Publish State of Play Report 2015
- ▶ Disseminate TSC Recommendations after document revision
- ▶ Further development of future Task Forces proposals
- ▶ Revise “IRDiRC Recommended” scope and procedure
- ▶ Organise standing teleconference of the Operating Comm
- ▶ Organise the next teleconference of the Exec Comm
- ▶ Organise the next face-to-face of the Exec Comm and Sci Comms in Mar 2016
- ▶ Collaborate with GA4GH on ICHG 2016 session programme

Acknowledgements to the host

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Annex - List of participants

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Lysogene, France	Karen Aiach
Shire Pharmaceuticals, Ireland	Albert Seymour
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Telethon Foundation, Italy	Lucia Monaco
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PTC Therapeutics, USA	Ellen Welch

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Genetic Alliance, USA	Sharon Terry

<u>Scientific Committees</u>	
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Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam

<u>IRDIRC Scientific Secretariat</u>	
SUPPORT-IRDIRC Project	Sékolène Aymé, Lilian Lau

Apologies

Members	Representative
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