



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 17th
Consortium Assembly**

Paris, November 21-22, 2019



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on November 21-22, 2019 in Paris, France. It was attended by 33 participants in person and 4 via teleconference, representing 30 Member organizations, the Chairs/Vice Chairs of the Scientific Committees (SCs) and the Scientific Secretariat (Sci Sec).

1. Welcome

- ▶ IRDiRC Consortium Assembly (CA) members were welcomed by the Chair Lucia Monaco to the 17th CA meeting in Paris, France.
- ▶ The meeting started with a talk by Dr Catherine Nguyen, who presented the French National Institute of Health and Medical Research (Inserm), which recently joined IRDiRC as Funder member and hosted the meeting.

2. Rare Diseases State of Play and IRDiRC Achievements

- ▶ The Sci Sec presented the major policy and research initiatives in the RD field that took place in the last couple of years and provided an overview of the achievements made by IRDiRC.

3. New members presentation

- ▶ New IRDiRC member representatives (Yposkesi and APARDO) illustrated their organizations to CA members

4. Round Table

- ▶ IRDiRC members presented the 2-3 key actions and events that happened in the past six months at their respective organization relating to the IRDiRC goals (reported in the Annex).

5. Scientific Committees updates

- ▶ The Chairs of the Scientific Committees presented updates on their current and proposed activities.
 - Diagnostic Scientific Committee (DSC)
 - Currently 15 members
 - Task Force (TF) *Indigenous Population* ongoing
 - 2 proposals for the 2020 Roadmap
 - Interdisciplinary Scientific Committee (ISC)
 - Currently 12 members

- TF *Clinical Research Networks for RDs* ongoing
 - TF *Natural History Studies* currently on hold
 - 3 proposals for the 2020 Roadmap
 - Therapies Scientific Committee (TSC)
 - Currently 13 members
 - TF *Orphan Drug Development Guidebook* expected to be accomplished by end of year
 - 3 manuscripts under-preparation
 - 4 proposals for the 2020 roadmap
6. Report from Constituent Committees breakout sessions
- ▶ The IRDiRC Constituent Committees reported back from their breakout sessions, and presented updates on their current and proposed activities.
 - Patient Advocates Constituent Committee (PACC)
 - Currently 14 members
 - PACC members discussed membership criteria and their role in IRDiRC
 - TF *Barriers to Patient Participation in Research* now became a working group
 - The Committee did not put forward proposals for the 2020 roadmap but some actions proposed by other committees were identified as interesting for PACC
 - Funder Constituent Committee (FCC) / Companies Constituent Committee (CCC)
 - Currently 33 members
 - FCC presented the progress on Activity A, the database of funded research projects which should soon be operational
 - FCC submitted one TF proposal for the 2020 roadmap
 - The outcomes of workshop on Social and Human Sciences (SHS) to set up a funding call focused on this topic area within the EJP RD were presented
7. Engagement with external members
- ▶ The SciSec sent a survey to each member of the constituency and the scientific committees in order to map the different initiatives engaged by IRDiRC members with external partners. The SciSec received 34 responses (out of 98 total) and mapped a total of 129 engagements. A summary of the survey outcome was presented for discussion and identification of effective collaborative models.
8. Validation of the 2020 roadmap of activities
- ▶ Prior to the face-to-face meeting, The CA had been invited to rank and prioritize the activities proposed for year 2020. Based on this vote and other criteria (Sci Sec work-

capacity and budget), the Sci Sec proposed a Gantt chart of activities that was validated by the CA.

- ▶ Four activities were selected and prioritized in the 2020 roadmap of activities
 - *Shared Molecular Etiologies*
 - *Rare Diseases Treatment Access*
 - *Chrysalis Project*
 - *New Technologies for the Diagnosis of Rare Diseases*
- ▶ Two activities that were not prioritized could benefit from collaborative work and could therefore be launched in 2020
 - *Galaxy Guide 2* (collaboration with EJP RD)
 - *Machine Readable and Automated Consent* (collaboration with EJP RD and GA4GH)
- ▶ Four activities were not prioritized for year 2020
 - *Primary Care*
 - *Alternative Business Models*
 - *Big Bang Project/Drug Repurposing Guidebook*
 - *Pluto Project/Disregarded Rare Diseases*
 - Task Force proposers have the possibility to re-propose these activities for year 2021 or identify alternative source of funding to launch them.

9. Communication and outreach

- ▶ The Sci Sec highlighted the list of IRDiRC publications in 2019 and presented its new communication material
- ▶ The organization of the IRDiRC conference (March 11-14, 2020) and the IRDiRC CA-SC meeting (March 14-15, 2020), taking place back-to-back in Berlin, was advertised

10. TSC membership renewal and nomination

- ▶ Michela Gabaldo membership was renewed for a 3-year period
- ▶ Anneliene Jonker, Janet Maynard and Daniel O'Connor were appointed as new members of the TSC for a 3-year period

11. Update from the Sci Sec

- ▶ The Sci Sec presented an update of its activities in 2019 and described the budget used in 2019 and foreseen for 2020.

REPORT

1. Welcome

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

INSERM presentation – Dr Catherine Nguyen

Inserm, a leading European academic biomedical research institution, and the coordinating institution of the European Joint Programme on Rare Diseases (EJP RD) is a new IRDiRC member and hosted this CA meeting.

Dr Catherine Nguyen, Director of research of the Thematic Institute Multi-Organisms “Genetics, Genomics and Bioinformatics” at Inserm, was nominated as representative to the IRDiRC Consortium Assembly and the Funders Constituent Committee.

- ▶ Inserm is a leading European organization for health research from basic science to public health with a budget of 998 million euros.
- ▶ It operates under the joint authority of the French Ministries of Health and Research
- ▶ Scientific activities are organized around 9 thematic institutes, corresponding to the main fields of biomedical and health research.
- ▶ With about 12,000 publications per a year, Inserm is the 2nd in the world behind the NIH.
- ▶ Inserm is directly funding clinical research, e.g. pilot projects that were set up as part of the France Genomic Medicine 2025 plan.
- ▶ Inserm contributes to the establishment of the national research and innovation strategy in the biomedical field as member of Aviesan (National alliance for life and health).
- ▶ Today Inserm is one of the main actors in rare diseases research.
- ▶ Inserm's thematic Institute of Genetics, Genomics and Bioinformatics participated in the preparation of the third National Plan for Rare Disease (PNMR3), the goals of which are in line with those of IRDiRC.
- ▶ Inserm hosts the scientific secretariat of the IRDiRC.

2. Rare Diseases State of Play and IRDiRC Achievements

The meeting kicked off with a presentation by the Sci Sec on the major achievements made in the RD field in the last couple of years

State of Play report (download a copy of the report [here](#))

- ▶ Major policy initiatives for RD patient community
- ▶ Major initiatives in RD research
- ▶ Research projects and clinical trials for RDs
- ▶ Distribution of research projects by medical domain
- ▶ Distribution of research projects by disease coverage

IRDiRC achievements

- ▶ Progress made toward IRDiRC goals:
 - Goal 1: 413 new RDs and 101 new RD genes between Jan and Oct 2019
 - Goal 2: 62 new orphan drugs in 2019 for 50 new indications
- ▶ Task Forces and Working Groups
 - 11 Task Forces (TFs) created since the establishment of IRDiRC TFs in 2015, 3 per year in average, and 2 Working Groups (WG)
 - 5 publications this year resulting from IRDiRC TFs/WGs
 - 2 TFs and one WG ongoing
 - Indigenous Population
 - Clinical Research Networks
 - WG on Goal 3
 - IRDiRC Database of funded projects about to be accomplished
- ▶ IRDiRC Recognized Resources (IRR)
 - 24 resources successfully applied for this designation since its conception in 2016, including platforms (7), tools (4), databases (3), guidelines and standards (10)
 - One new resource approved this year: MARRVEL
- ▶ Membership
 - Currently 59 Member organizations in 3 constituencies:
 - Funders Constituent Committee (FCC) – 33 members
 - Companies Constituent Committee (CCC) – 12 members
 - Patient Advocacy Constituent Committee (PACC) – 14 members
 - Constant growth in terms of membership number for all constituencies but the CCC due to failure to comply with the IRDiRC engagement rules, resulting in membership termination

- Member organizations spread all over the 5 continents:
 - Americas (North America and Canada)
 - Europe
 - Africa
 - Asia
 - Australia
- Since the last face-to-face meeting, 4 new members: Inserm, Yposkesis, Eloxx Pharmaceuticals, and APARDO

3. New Members Presentation

Over the past six months since the last face-to-face meeting, four new members have joined IRDiRC: Inserm, Yposkesi, Eloxx Pharmaceuticals, and the Asia Pacific Alliance for Rare Diseases (APARDO). Inserm introduction was included in the opening welcome; the other new members that were present at the meeting presented themselves.

- ▶ Yposkesi, Jean-Pierre Gaspard (Executive Chairman)
 - First French company to develop and produce gene and cell therapy products for RDs and one of the European leaders in gene therapy manufacturing capacity
 - Launched in Nov 2016 as spin-off of Genethon (121 M € investment)
 - AFM-Telethon is the major shareholder
 - It invests significantly in innovation and bioprocessing and has already invested 6 M € to research projects relevant to IRDiRC scientific objectives
 - Three top pharma companies and one US leading biotech as clients
 - It aims to make the first treatments available to patients and market them at a fair and controlled price, in accordance with the commitment made by its founders
- ▶ APARDO, Ritu Jain (President)
 - It brings together patient advocate leaders from across the Asia Pacific region representing rare diseases and rare cancers to help improve treatment outcomes for patients
 - At present, 24 organisations from 14 countries across the Asia Pacific region
 - Some members are representatives of disease-specific groups, others from societies for RDs or rare cancers in general, and others representing patient group alliances or networks.
 - APARDO Summit Oct 2019 in Taiwan attended by 51 participants from 17 countries, including South American countries
 - Partnership with the Asia-Pacific Economic Cooperation (APEC) working group on RDs

4. Members update – roundtable

Representatives of IRDiRC member organizations were invited to present the key actions/events that happened in the past six months towards advancing IRDiRC goals 2017-2027 (please refer to the Annex at the end).

5. Updates from Scientific Committees

- ▶ Diagnostics Scientific Committee (DSC)
 - Currently 15 members
 - Ongoing activities
 - *Indigenous population* TF
 - 14 members
 - Kick off meeting Sep 25, 2019
 - Face to face meeting Mar 10, 2020 in Berlin
 - Future Activities
 - 2 TF proposals for the 2020 Roadmap:
 - *New Technology and Integrative OMICs* – focused on the integration of metabolomics and genomics, and specifically clinical translation and implementation
 - *Primary Care* – transversal, pending inputs from TSC and ISC
- ▶ Interdisciplinary Scientific Committee (ISC)
 - 2019 actions
 - TF *Clinical Research Networks for RDs* launched Oct 2019
 - Article resulting from TF *Model Consent Clauses for RD Research* published
 - Next steps
 - TF Natural History Studies
 - 3 proposals for the 2020 Roadmap:
 - *Shared Molecular Etiologies, SaME* (TF)
 - *Access to RD Treatment* (WG) – need 3-4 experts from other committees or external
 - *Automated and Computable Consent Models* (TF)
 - Committee composition/turnover
 - Chair elected in June 2019 in replacement of Domenica Taruscio (currently representing ISS in the FCC)
 - 5 membership terminations: Edmund Jessop, Hanns Lochmüller, Angel Carracedo, Petra Kaufmann, Ken Ishii

- Member transition: Domenica Taruscio from ISC to FCC
 - 5 new members: Takeya Adachi, Kate Baker, Marc Doms, Yllka Kodra, Mahsa Shabani
- ▶ Therapies Scientific Committee (TSC)
- 2019 actions
 - Galaxy Guide phase 1
 - TSC Chair presented the guidebook for orphan drug development in its current stage (i.e. 116 building blocks) and application in 3 case studies. The TF *Orphan Drug Development Guidebook* (ODDG) will be finalized by end of the year
 - Dissemination of results in congress already started (ESGCT, Telethon convention, WODC EU) and continue next year
 - Article resulting from TF *Data Mining and Repurposing* published
 - 3 manuscripts under preparation:
 - ODDG main paper in discussion with Nature RDD
 - Gap analysis and IRDiRC roadmap toward GOAL 2
 - New Drug Counter
 - Next steps
 - 4 TF proposals for the 2020 roadmap:
 - *Galaxy Guide Phase 2* foreseen collaboration with EJP RD
 - *Drug Repurposing Guidebook*
 - *Disregarded Rare Diseases (PLUTO Project)*
 - *Alternative Business Models for Drug Development*
 - Planning ODDG publication (beyond NRDD paper)
 - Committee composition/turnover
 - 5 membership terminations: Karin Rademaker, Josep Torrent I Farnell, Sandrine Marreaud, Yann Le Cam
 - Member transition: Anne Pariser from TSC to FCC
 - 2 new members: Sangeeta Jethwa and Thomas Morel
 - 1 membership renewal: Michela Gabaldo
 - 3 nominations: Anneliene Jonker (TechMed Centre, NL), Janet Maynard (FDA, USA), Daniel O'Connor (MHRA, UK)

6. Report back from Constituent Committee breakout sessions

▶ Feedback from the Patient Advocacy Constituent Committees (PACC)

○ Committees update

- Currently constituted of 14 umbrella patient organizations, including national, regional and international alliances in Canada, US, Europe, Africa and Asia Pacific region
 - New member: APARDO
- PACC members came to a consensus that the current IRDiRC criteria for the inclusion of new umbrella patient advocacy groups are valid
- PACC members discussed together with the Sci Sec the role of the Committee in IRDiRC
 - PACC are expected to ensure and provide patient perspective in all of its undertakings and as participating members of multi-stakeholder TFs and WGs
 - PACC should take advantage of their collective capacity to put forward actions on cross-cutting issues relating to RD research
- PACC members differentiate in terms of activities and outreach but there is considerable interaction through their membership in regional/international alliances
 - IRDiRC addresses barriers to RD research

○ Ongoing activities

- TF *Barriers to Patient Participation in Research* held 6 conference calls to date but several issues are holding back its progress (leadership change, scope changes, budget constraints). Since the TF approval was made in 2018 and the budget is already committed to the roadmap, decision was made to transform this action into a WG.

○ Future activities

- The Committee contributed to the 2020 proposed actions; the PACC Chair is a co-leader of the WG *RDs Treatment Access* and at the steering committee of the *Chrysalis* Project
- Existing opportunities for patient advocates' engagement in activities proposed by other constituencies were discussed and all the 4 prioritized activities have been identified as interesting for PACC.

▶ Feedback from the Funders and Companies Constituent Committees (FCC/CCC)

○ Ongoing activities

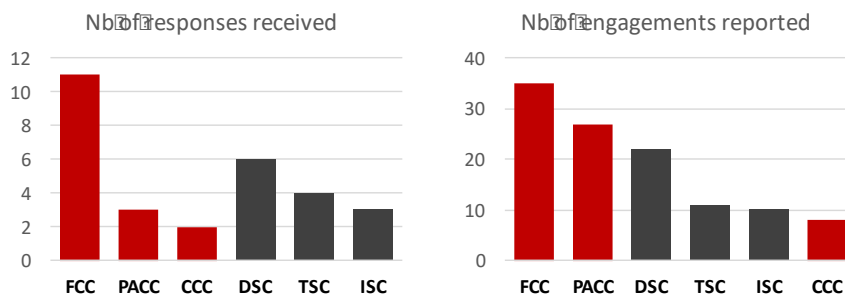
- Update on Activity A - IRDiRC database of funded RDs research projects

- Funders must submit information on access to their data
- Access to the Beta version of the platform can be given to members willing to test and provide feedback on the tool
- Ongoing developments: translation test (from JP, DE, NL to EN), curation details are finalized
- Results of the analysis can be utilized and exploited in publications and should appropriately acknowledge IRDiRC
- A data transfer agreement (DTA) should be signed by Funders on the data to be used and analyzed with the platform
- Need input/feedback in relation to:
 - Currency conversion (\$, €, etc.)
 - Next steps after the platform is operational – open it to all? Use as potential self-sustainable (payable) resource?
- Future activities
 - *Chrysalis* Project
 - Despite complementarities with the *PLUTO* Project, no consensus was reached with the applicants to merge the two
 - *PLUTO* may get some funding from outside IRDiRC but lead persons from *PLUTO* should be involved in the *Chrysalis*
 - *Chrysalis* will not start in Q1 2020 and needs to engage with CCC
 - Connection with *SaME* should be ensured
 - *Natural History Studies* (NHS)
 - The FDA draft guidance on NHS for drug development is open for comments by FCC members, after it could be integrated as an IRDiRC recognized resource
 - Idea is to start working on innovative NHS approaches based on good practices examples (eg. Duchenne) that could be expanded to larger scale
- Update on the E-Rare strategic workshop on Social and Human Sciences (SHS) provided by Sonja van Weely
 - E-Rare workshop took place in Gdansk, Poland on September 20, 2019 with the aim to identify topics in the SHS research area that may be suitable for the Joint Transnational Call 2021 in the European Joint Programme on Rare Diseases (EJP RD)
 - FCC members interested to get involved in the call
 - Topics identified to improve health care implementation and everyday life of people living with a rare disease and their families like psychological, social and health economic impact

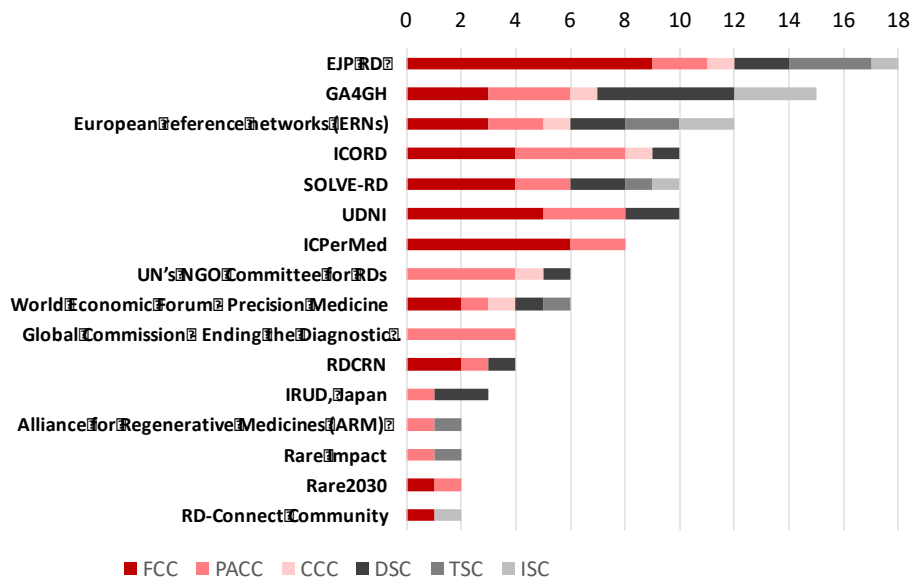
- Evaluation criteria need to be adapted to the specific stakeholders (including patients and families)

7. Engagement with external initiatives

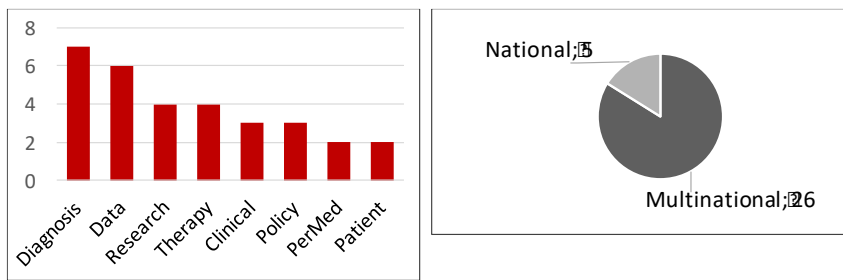
- ▶ The SciSec sent a survey to each member of the constituency and the scientific committees in order to map the different initiatives in which IRDiRC members are engaged with external partners
 - 34 responses received (out of 98 total)
 - 31 different national or multinational initiatives reported
 - Total of 129 engagements



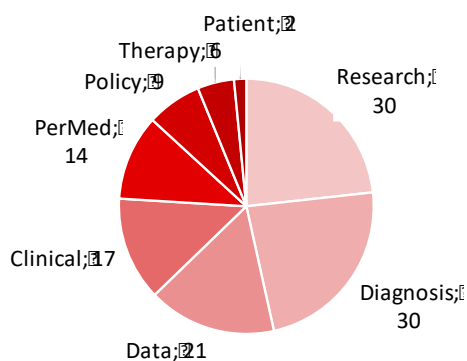
- ▶ The top initiatives were reported as follows:



- ▶ The different types of reported initiatives were Diagnosis (7), Data (6), Research (4), Therapy (4), Clinical (3), Policy (3), PerMed (2), Patient (2), and 84% (26/31) of the initiatives have multinational coverages.



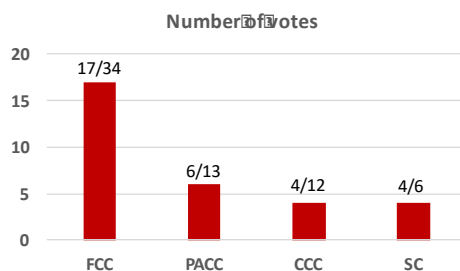
- ▶ The different types of reported engagement were Research (30), Diagnosis (30), Data (21), Clinical (17), PerMed (14), Policy (9), Therapy (6), Patient (2). The absence of engagement with regulatory agencies was highlighted as a point to address.



- ▶ Engagement with NGO Committee for Rare Diseases was discussed among the members. It was agreed that IRDiRC would be the right consortium to be present at the Committee to provide the research policy visions.

8. Validation of the 2020 roadmap of activities

- ▶ On November 13 2019, the SciSec sent a survey to the CA members and invited them to rank and prioritise the 10 activities previously identified during the CA-SC meeting in Leiden (May 22-24, 2019).
 - The SciSec received 31 completed responses



- A scoring coefficient based on the ranking was used to score each activity (project ranked 1st=10 points, ..., project ranked 10th=1 point). The final ranking was as follows:
 - 1- Shared Molecular Etiologies (SaME) (201 points)
 - 1- Rare Diseases Treatment Access (201 points)
 - 3- Chrysalis Project (199 points)
 - 4- New Technologies for the Diagnosis of Rare Diseases (186 points)
 - 5- Galaxy Guide 2 (168 points)
 - 6- Primary Care (164 points)
 - 7- Alternative Business Models (164 points)
 - 8- Big Bang Project/Drug Repurposing Guidebook (156 points)
 - 9- Pluto Project/Disregarded Rare Diseases (151 points)
 - 10- Machine Readable and Automated Consent (115 points)
- ▶ The activity selected for year 2020 were programmed as described in the following Gantt chart
 - The first four ranked activities (SaME, RD Treatment Access, Chrysalis and New Technologies) were prioritized (light blue). This selection was based on different criteria including the number of ongoing activities, the workload within the proposing Committees, the SciSec human resources and work capacity (2 Project Managers), and the SciSec budget (100,000 euros for the activities in 2020).
 - Two activities (green), namely “Galaxy Guide 2” and “Machine Readable and Computable Consent” were identified as activities benefiting from a collaborative action and could therefore be programmed for the year 2020. The Galaxy Guide 2 could be developed in collaboration with EJPRD-Pillar4 while Machine Readable and Computable Consent could be developed in collaboration with EJPRD-Pillar2 and GA4GH.
 - Four activities were not prioritized for 2020 (grey). However, the Task Force proposers have the possibility to re-propose them for the 2021 Roadmap of activities or identify alternative source of funding to launch them.

Activities	Proposers	2019				2020				2021			
		Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4
WG on Goal B	CA												
Indigenous Population	DSC												
Clinical Research Networks	ISC, TSC, FCC												
Natural History Studies	ISC, TSC, FCC												
RD Treatment Access	ISC, ACC												
Chrysalis	FCC, CCC												
New Technologies	DSC												
SaME	ISC												
Galaxy Guide 2 (+EJPRD)	TSC												
MRC Consent (+EJPRD/GA4GH)	ISC												
Primary Care	DSC												
Alternative Business Models	TSC												
Big Bang Project	TSC												
Pluto project	TSC												

	Ongoing and foreseen activities
	Activities prioritized for 2020
	Activities benefiting from collaborative action
	Activities not prioritized

- ▶ A plenary discussion followed the presentation of the survey outcome and of the proposed Roadmap. The Assembly expressed the following considerations for developing the 4 prioritized activities:
 - Shared Molecular Etiologies:
 - The regulatory aspect of this approach might be a roadblock. Reimbursement agencies will also need evidence for efficacy. It should be relevant to engage with companies using this approach in oncology and get their experience and perspectives.
 - The same molecular etiology in various diseases can lead to a spectrum of clinical manifestations and therefore, the definition of clinical trial endpoints will also be different among the diseases. This aspect will require the involvement of regulators and oncologists with experience in these types of clinical trials.
 - Rare Diseases Treatment Access:
 - It will be important for IRDiRC to engage with the UN and WHO. The Working Group should take advantage of the existing medicine lists (e.g. FDA and Orphanet lists) and also identify which diseases require priority actions in some countries. This will help in starting the work and engaging with partners.
 - IRDiRC should also be represented at the NGOs and WHO level, and be directly involved in their activities on rare diseases.
 - It will be important for the Working Group to identify initiatives moving towards the same goal (e.g. the Health Systems Strengthening Services from WHO).
 - Chrysalis Project:
 - The quality, type and reproducibility of data as well as the identification of relevant clinical endpoints are key elements in attracting industry and also bridging the translation phase from academic research to industry settings.
 - PAO engaged in drug development can work collectively with companies and support the development of research.
 - Attracting more companies to invest in RD or addressing neglected RD? The spirit that motivated this Task Force is the latter.
 - RD with very low prevalence are not interesting for the pharma industry because there is no business model that is working for them. There is a need to change the drug development model. It is important to remember how we can bring therapies to these patients and what are the incentives that can make a difference (e.g. review IMI work in speeding drug development and patient access to therapies, need to liaise with other kind of industries such as digital diagnostics).
 - Need to identify clinical champions and natural history data used by institutions/academics and then to go to the regulators and engage in phase-3

clinical trials on ultra-rare diseases. This point highlights the importance to integrate the proposers of the Pluto project in the development of the Chrysalis Task Force.

- **New Technologies for the Diagnosis of Rare Diseases:**
 - Different technologies are applicable to different diseases. It is expected to be a flourishing area in personalized medicines setting.
 - The transcriptomics technology is more advanced than metabolomics.
 - A list of precision medicine specialists could be used and companies should be involved.
 - Bioinformatics approaches, modelling and simulation need to be added.
 - The diagnostic paradigm should be reversed from companion diagnostics (diagnostic test used as a companion to a therapeutic drug to determine its applicability) to companion therapeutics (e.g. targeting a treatment according to the gene and phenotype). This could help in devising the pathway and type of diagnostics. This is a joint diagnostic approach to be considered.
- ▶ **Considerations for the activities that may benefit from a collaborative action**
 - **Galaxy Guide 2**
 - This guide needs progressive updates. It will be self-maintained by the community (provision of project management template that could be uploaded to project management software).
 - The guide will need curators.
 - The extreme potential of this tool must be preserved. The connection with EJP RD is being discussed regarding its maintenance and the development of an interactive version.
 - A transition plan is being designed with EATRIS as well.
 - **Machine Readable and Computable Consent:**
 - This activity can benefit from a collaboration with GA4GH and EJP RD.
 - The work will continue to advance although some points need to be clarified.
- ▶ From a project management perspective, it was mentioned that Task Force leaders should clearly define the objectives, identify the expertise, deliverables, milestones and timeline. Identifying the time for the in-person meeting and planning the budget in advance may generate some savings to inject in other activities (e.g. Pluto project).

9. Communication and outreach

- ▶ The work of IRDiRC members and Task Forces led to the publications of 6 articles in 2019
 - **Privacy-Preserving Linkage of Genomic and Clinical Data Sets.** Dixie B. Baker, Bartha M. Knoppers , Mark Phillips , David van Enckevort , Petra Kaufmann, Hanns

- Lochmuller, and Domenica Taruscio. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2019
- Ethical, legal, and social issues (ELSI) in rare diseases: a landscape analysis from funders. Adam L. Hartman, Anneliene Hechtelt Jonker, Melissa A. Parisi, Daria Julkowska, Nicole Lockhart and Rosario Isasi. European Journal of Human Genetics, 2019
 - The use or generation of biomedical data and existing medicines to discover and establish new treatments for patients with rare diseases – recommendations of the IRDiRC Data Mining and Repurposing Task Force. Noel T. Southall, Madhusudan Natarajan, Lilian PL. Lau, et al. Orphanet Journal of Rare Diseases, 2019
 - Model consent clauses for rare disease research. Minh Thu Nguyen, Jack Goldblatt, Rosario Isasi, et al. BMC Medical Ethics, 2019
 - A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Kym M. Boycott, Taila Hartley, Leslie G. Biesecker, et al. Cell, March 2019
 - International collaborative actions and transparency to understand, diagnose, and develop therapies for rare diseases. Kym M Boycott, Lilian PL LAU, Christine M Cutillo, Christopher P Austin. EMBO Molecular Medicine, April 2019
- ▶ To disseminate the results of IRDiRC activities more effectively, the organisation of webinar series should be planned once the activities are completed and the publication accepted. Each webinar should be complemented with statements from a person from each of the constituency that would provide context and make it more interactive. The manuscript could also be shared with the European Commission communication team in advance of publication so that they can prepare a communication material and relay the news.
 - ▶ The new IRDiRC flyer and poster have been developed and are available upon request to the SciSec. The twitter account is being maintained.
 - ▶ 383 contacts are registered to the IRDiRC newsletter. Its structure is as follows:
 - New members
 - Recognized Resources
 - Publications
 - Calls for Task Force opening or Scientific Committee appointment
 - Events
 - Task Force outcomes (articles, videos, etc)
 - Others
 - ▶ IRDiRC was presented in more than 15 conferences in 2019. Members who are expecting to present IRDiRC in 2020 are invited to inform the SciSec.
 - ▶ The joint RE(ACT) Congress and IRDiRC conference will be organised from March 11-14, 2020 in Berlin. IRDiRC members will benefit from a special registration fee.

- ▶ The IRDiRC conference will be followed by the IRDiRC CA-SC meeting on March 14-15. The costs (travel, accommodation) associated with this meeting for the Scientific Committees and the PACC will be taken in charge by the SciSec. A detailed agenda will be circulated early in 2020.

	Wed 11 March	Thu 12 March	Fri 13 March	Sat 14 March	Sun 15 March
Morning		Session B Diagnostic, WGS, artificial intelligence, new technologies	Session D Molecular etiology of RD, innovative clinical trials, precision medicine	Session F Patient engagement in drug development and clinical trials	IRDiRC committee meeting CA-SC common session
Afternoon	Session A (workshop) (1) Methodologies to assess the effect of diagnoses and therapies on RD patients (2) Presentation of the Galaxy Guide	Session C Advanced therapies: gene editing, cell therapy	Session E Access to diagnosis and drugs for all	IRDiRC committee meeting Breakout sessions PACC CA&SC	IRDiRC committee meeting CA session
Evening	Opening Ceremony	Speakers Dinner	Social event (if sponsored)		

10. TSC membership renewal and nominations

- ▶ The membership of Michela Gabaldo was renewed for 3 years (October 2019 – October 2022)
- ▶ Anneliene Jonker (TechMed Centre, NL), Janet Maynard (FDA, USA) and Daniel O'Connor (MHRA, UK) have been appointed as new members of the TSC for a 3-year period (November 2019 – November 2022).

11. Update from the SciSec

- ▶ The SciSec participated in the implementation and the development of 3 new activities in 2019:
 - Working Group on Goal 3: Started in June 2019, 5 teleconference calls were organized, F2F meeting is planned on February 19-20, 2020
 - Clinical Research Networks for Rare Diseases: Started on October 2019, 2 teleconference calls were organized.
 - Indigenous Populations: Started on September 2019, 1 teleconference call was organized.
- ▶ Meeting organized between January and November 2019:
 - Scientific Support team: 16 teleconferences, 2 in person (Paris, Milan)
 - Operating Committee: 10 teleconferences (Note: Chair and Vice Chair need to identify a substitute if neither of them can attend the call)
 - Consortium Assembly: 2 teleconferences, 2 in person (Leiden, Paris)
 - Task Force/Working Group: 14 teleconferences

- Committees: 6 teleconferences (Note: These meetings should be organized more frequently. A meeting calendar will be set up for the year 2020)
 - Hosted representatives from the Shanghai Center for Clinical Laboratory: F2F in Paris
- ▶ Meeting attended in 2019:
- Mary Wang: China Rare Disease Summit, Shenzhen, September 2019
 - Carla D’Angelo and Daria Julkowska: Fondazione Telethon Scientific Convention, Riva del Garda, October 2019
 - Galliano Zanello: Annual meeting of the French National Healthcare Network for Rare Endocrine Diseases, Paris, December 2019
- ▶ SciSec budget
- The SciSec had a budget of 180,500 euros for the year 2019. The funds were used as described in the table below:

Expense 2019	TOTAL €
CA-SC meeting, Leiden, May 22-24, 2019	10000,00 €
CA meeting, Paris, November 21-22, 2019	10000,00 €
SciSec travel	200,00 €
TSC Task Force meeting	300,00 €
Communication	2400,00 €
Other direct costs	700,00 €
Total	180500,00 €
Reserved budget	
Task Force (x4 F/year): 25000,00 €	100000,00 €
Total for 2019	180500,00 €

- ▶ The total SciSec budget for the year 2020 is 245,000 euros and it will be used as follows:

Missions Travel	TOTAL €
SC meeting (once a year): 35000 € total/year	35000,00 €
PACC meeting (twice a year): 15000 €	30000,00 €
Sous total	65000,00 €
Other direct costs	
CA meeting (for the year): 10000 €	10000,00 €
Task Force (x4 F/year): 25000 €	100000,00 €
IRDiRC congress (1 every 2 years): 65000 €	65000,00 €
Communication (for the year): 5000 €	5000,00 €
Sous total	180000,00 €
TOTAL	245000,00 €

- ▶ Voluntary membership fund
- IRDiRC reinstated a voluntary membership fee to provide the organization with additional funds to use for outreach activities and support the travel of patient organization representatives to IRDiRC meetings. The payment of this fee is voluntary and set at different levels depending on the nature of the member organization. As of November 2019, only one member organization contributed to the VMF. Therefore, IRDiRC is calling for additional contributions from its members

- The recommended annual contribution from each membership type is as follows, subject to discretion of each member organization with regard to the amount and capacity to contribute:
 - Companies \$5,000
 - Funders \$2,000
 - Patient advocates \$1,000

12. Next meetings in 2020

- ▶ Consortium Assembly
 - Joint RE(ACT)-IRDiRC congress, March 11-14, Berlin
 - IRDiRC CA-SC in person meeting, March 14-15, Berlin
 - CA teleconference in January, June, September
 - CA fall in person meeting October-November 2020
 - Regular Committee teleconferences (x3/year)
- ▶ Scientific Committees
 - Joint RE(ACT)-IRDiRC congress, March 11-14, Berlin
 - IRDiRC CA-SC in person meeting, March 14-15, Berlin
 - TF on Indigenous population, in person meeting, March, Berlin
 - TF on Clinical Research Networks, in person meeting on Q2 (TBD)
 - Launch of the activities prioritized in the roadmap, regular set of calls
 - Regular Committee teleconferences (x3/year)
- ▶ Other activities
 - Working Group on Goal 3: In person meeting, February 19-20, Paris

Actions and deliverables

- ▶ SciSec
 - Prepare the Task Force calls for nominations
 - Set up a meeting calendar for each Committee in 2020
- ▶ Committees
 - Task Force leaders to implement an operation plan for each new activity
 - Chairs and Vice Chairs are requested to identify a substitute if neither of them can participate in the Operating Committee meetings
 - IRDiRC members should be invited to the SciSec if they plan to present IRDiRC during a conference in 2020

Document history

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Version 2. Report edited by Carla D’Angelo and Galliano Zanello, December 20, 2019

Version 3. Report edited by Carla D’Angelo and Galliano Zanello, January 2, 2020

Circulated to members of the CA, xx xx, xx

Annex - List of participants

Members	Representative
Telethon Foundation, Italy	Lucia Monaco (Chair), Mary Wang
Sanford Research, USA	David Pearce (Vice Chair)
Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid)	Yukiko Nishimura
Agence Nationale de la Recherche (ANR), France	Dominique Dunon-Bluteau
Botswana Organization for Rare Diseases (BORDIS)	Eda Selebasto
Canadian Institutes for Health Research (CIHR)	Christopher McMaster
Canadian Organization for Rare Disorders (CORD)	Durhane Wong-Rieger
Chinese Organization for Rare Disorders (CORD)	Qi Sun, Rachel Yang
Cydan II	James McArthur
E-RARE Consortium, Europe	Florence Guillot
European Commission	Irene Norstedt, Christina Kyriakopoulou
Federal Ministry of Education and Research (BMBF), Germany	Ralph Schuster
Food and Drug Administration, Office of Orphan Products Development (FDA/OOPD)	Katherine Needleman (teleconference)
French Muscular Dystrophy Association, AFM-Téléthon	Marie-Christine Ouillade, Alexandre Mejat
Genetic Alliance	Katherine Lambertson (teleconference)
Global Genes	Maureen McArthur (teleconference)
Indian Organization for Rare Diseases (I-ORD)	Ramaiah Muthyala
Instituto de Salud Carlos III (ISCIII), Spain	Manuel Posada de la Paz
Istituto Superiore di Sanità (ISS)	Domenica Taruscio (teleconference)
Japan Agency for Medical Research and Development (AMED), Japan	Noriaki Imanishi
Korea National Institute of Health (KNIH), South Korea	Younjhin Ahn
National Center for Advancing Translational Sciences (NCATS), USA	Anne Pariser, PJ Brooks

National Institute of Health and Medical Research from France (INSERM), France	Catherine Nguyen, Daria Julkowska
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
National Human Genome Research Institute (NHGRI), USA	Lisa Chadwick
Pfizer	Katherine Beaverson
Rare Diseases International (RDI), APARDO	Ritu Jain
The Netherlands Organisation for Health Research and Development (ZonMw)	Sonja van Weely
Western Australian Department of Health, Australia	Gareth Baynam
YposKesi	Jean-Pierre Gaspard, Sophie Blondel
<u>Scientific Committees</u>	
Diagnostics	Gareth Baynam, Sarah Bowdin
Interdisciplinary	Stephen Groft, Dixie Baker
Therapies	Diego Ardigò
<u>IRDIRC Scientific Secretariat</u>	Carla D'Angelo Galliano Zanello Katerina Tzima Nadia Ibellaatti Juliane Halftermeyer Yanis Mimouni

Apologies

Members	Representative
Academy of Finland	Heikki Vilen
BGI	Ning Li
Chiesi Farmaceutici	Andrea Chiesi
Eloxx Pharmaceuticals	David Snow
European Organisation for Treatment & Research on Cancer (EORTC)	Denis Lacombe
EURORDIS-Rare Diseases Europe	Virginie Bros-Facer

French Foundation for Rare Diseases (FFRD), France	Daniel Scherman
Genome Canada, Canada	Cindy Bell
Genzyme	Daniel Gruskin
Georgian Foundation for Genetic and Rare Diseases (GeRaD)	Oleg Kvlividze
Loulou Foundation, UK	Daniel Lavery
Lysogene	Karen Aiach
National Institute for Health Research (NIHR), United Kingdom	Willem Ouwehand
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
National Cancer Institute (NCI), USA	Edward Trimble
National Eye Institute (NEI), USA	Santa Tumminia
National Institute of Dental and Craniofacial Research (NIDCR), USA	Jason Wan
National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), USA	Faye Chen
National Organization for Rare Disorders (NORD)	Vanessa Boulanger
National Rare Diseases Registry System of China (NRDRS), China	Zhang Shuyang
Organization for Rare Diseases India (ORDI)	Prasanna Kumar Shirol
Rare Diseases South Africa	Kelly du Plessis
Rare Voices Australia	Nicole Millis
Recursion Pharmaceuticals, Inc	Christopher Gibson
Roche	Mathew Pletcher
Saudi Human Genome Project	Sultan Turki Alsedairy
Takeda Pharmaceuticals	Madhu Natarajan
Ultragenyx	Tom Pulles



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