INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 18th Consortium Assembly Meeting

March 12-13, 2020

IRDiRC

AGENDA AND NOTICE

The annual face-to-face meeting of the Consortium Assembly (CA) and Scientific Committees (SCs) was scheduled to take place in Berlin on March 14–15, 2020 following the RE(ACT) Congress and IRDiRC Conference planned to March 11–14, but it was cancelled because of the COVID-19 outbreak. Consequently, IRDiRC organized two conference calls in replacement of the in-person meeting.

Although all the meeting topics originally proposed could not be covered during these two conference calls, the discussions highlighted the following points:

- Presentation of ongoing activities: Progress & feedback discussion with the CA/SC.
- New Task Force implementation and progress: Update on Task Force proposal if any, revisions, required expertise, expected start time, Task Force progress if already started

The booking of the Melia Hotel in Berlin was shifted to January 12-16, 2021. This was the only opportunity offered by the Hotel to not lose the money already spent. Because the face-to-face meeting of the CA & SCs next year is planned to take place between April and May in the US, IRDiRC wants to organize a one-day meeting dedicated to task force workshops prior to the IRDiRC-RE(ACT) Conference

- January 12: Task Force F2F meeting(s)
- January 13-16: IRDiRC-RE(ACT) Conference

Thursday, March 12, 2020 (2.30-4 pm CET)

- General update from Committee Chairs/Vice Chairs
- Working Group on Goal 3
- Indigenous Populations
- Clinical Research Networks for Rare Diseases
- Chrysalis project
- Shared Molecular Etiologies

Friday, March 13, 2020 (2.30-4 pm CET)

- Demonstration IRDiRC database of funded projects
- Rare Disease Treatment Access
- New Technologies
- Strategic planning for year 2021
- Conclusion and planning of next meetings

REPORT

The Consortium Assembly (CA) and Scientific Committees of the International Rare Diseases Research Consortium (IRDiRC) met on March 12-13, 2020 via teleconference. The meetings were attended by 50 participants, representing 34 Member organizations and the Scientific Secretariat (Sci Sec).

1. General update from Committees' Chairs/Vice Chairs

Funders Constituent Committee (FCC)

- Currently 34 members:
  - Europe: 17
  - North America: 12
  - Asia: 4
  - Australia: 01

- Changes in leadership:
  - The mandate of Daria Julkowska as FCC Chair ended on February 29, 2020
  - Adam Hartman, former FCC Vice Chair, was elected new FCC Chair for a 3-years mandate starting on March 2020
  - FCC Vice Chair position is vacant

- Activities involving the FCC:
  - Activity A: Database of Research Projects and Analysis Tools
    - Developed in collaboration with Orphanet and MyScienceWork
    - Translation tool is currently tested
      - First translations in Dutch, German and Japanese were just finalized
    - Next steps include:
      - Import data from IRDiRC funders
      - Define access to IRDiRC members
      - Decide if the database will be publicly accessible
  - Clinical Research Networks for Rare Diseases (see ISC)
    - Cross-committee action with the ISC and TSC
    - Described in the below section
  - Chrysalis project (see Section 3)
    - The overall goal is to identify the key criteria that would make RDs more attractive for industrial development.
    - The call for candidates will be opened by the end of March

Companies Constituent Committee (CCC)

- Currently 10 members:
  - Europe: 5
  - North America: 4
  - Asia: 1

- Changes in leadership:

- Mathew Pletcher (Chair) and Madhu Natarajan (Vice Chair) resigned on February 2020
- New CCC Chair is in being identified among CCC members

- IRDiRC is inviting new companies to join the CCC. Invitations were already sent to:
  - Amicus
  - BioMarin
  - Novartis/AveXis
  - Regenxbio
  - Sanofi Genzyme
  - Spark Therapeutics
  - Takeda

- Activities involving the CCC:
  - Collaboration with the FCC on the Chrysalis project (see Section 2)

→ IRDiRC members are welcome to suggest companies to join the CCC
→ CCC will engage in activities proposed by other Constituencies

Patient Advocates Constituent Committee (PACC)

- Currently 16 members:
  - Asia/Australia: 6
  - North America: 4
  - Africa: 3
  - Europe: 1
  - Latin America: 1
  - International: 1

- New members:
  - Iberoamerican Alliance for Rare Diseases (ALIBER)
  - Rare Disease Ghana Initiative (RDGI)

- Ongoing activities involving PACC members:
  - Task Force 'Indigenous Populations' (see section 2)
  - Working Group on Rare Diseases Treatment Access (see Section 5)

Diagnostics Scientific Committee (DSC)

- Currently 15 members:
  - Asia/Australia: 7
  - Europe: 5
  - North America: 2
  - Africa: 1

- Activities involving the DSC:
  - Task Force 'Indigenous Populations' (see section 2)
    - Composed of IRDiRC members (4 DSC, 1 ISC, 1 PACC, 1 FCC) and 12 externals experts
    - State of play report was drafted by the Sci Sec
    - The TF workshop planned in Berlin on March 10 was cancelled.
    - The group is currently investigating what are the next frontiers that need to be addressed to advance RD diagnosis in underrepresented populations.

- Integrating New Technologies for the Diagnosis of RD (see Section 5)
  - Expected start time in Q3 2020.
  - The goal of this activity is to identify evidence behind new technology approaches likely to increase the diagnostic rate for RD patients.
  - This activity will build on previous work from the DSC TF 'Solving the Unsolved'

- DSC Chair was appointed by IRDiRC as member in the Global Commission to End the Diagnostic Odyssey for Children with Rare Diseases to ensures connectivity to, and alignment between, IRDiRC and the Global Commission

Interdisciplinary Scientific Committee (ISC)

- Currently 11 members:
  - Europe: 6
  - North America: 5

- Changes in leadership:
  - End of mandate of ISC Chair Stephen Groft is on June 2020
  - A call for the next ISC Chair will be opened in Q2 2020

- Activities involving the ISC:
  - Clinical Research Networks for Rare Diseases (see Section 2)
    - The goal is to map and analyze the existing ecosystem of national/supranational clinical research networks to develop policy recommendations on guiding principles for an international framework of collaboration of these networks
    - TF started on October 2020 and counts with 22 experts, including 9 IRDiRC members (6 FCC, 2 ISC, 1 TSC)
    - 4 conference calls were already organized
    - It is currently developing a questionnaire to identify CRNs attributes
  - Shared Molecular Etiologies (see Section 3)
    - Expected start time Q3 2020
    - The goal is to identify strategies for grouping patients based on the shared molecular etiology of their RD in order to develop clinical trials involving a larger number of patients and evaluating the effect of drug targeting common molecular pathways

Therapies Scientific Committee (TSC)

- Currently 12 members:
  - Europe: 9
  - North America: 2
  - Asia: 1

- TSC membership:
  - End of membership: Seng Cheng, February 2020.
  - A call for experts will be opened in Q2 2020
  - TSC will develop a reserve list for the upcoming nominations

- Activities involving the TSC:
  - Orphan Drug Development Guidebook (ODDG)
    - 3-years long development process.
      - 2017: First gap analysis.

    - 2018: Establishment of the Guidebook principle, first steps of development and workshop in Dublin.
    - 2019: Refinement and expansion of the ODDG material, submission of the manuscript to Nature Review and Drug Discovery.
- ODDG manuscript was submitted to Nature Review and Drug Discovery and received a positive feedback from the editor.
    - The edited version of the manuscript was sent to the journal.
    - All the material (Building Blocks, check lists, etc) will be made available on the IRDiRC website.
- Start of the communication campaign.
    - A video webinar on how to use the guide will be registered.
    - The ODDG will be advertised on the IRDiRC newsletter, Twitter, YouTube channel.
- Development of an interactive tool for the guide.
    - The TSC will open a collaboration with Polytechnic University in Milan and develop an interactive tool of the ODDG.
    - The TSC is also exploring the ODDG integration within the EJPRD Innovation Management Toolbox.

2. Updates on ongoing activities

Working Group on Goal 3

- In February 2017, IRDiRC members engaged in deep deliberations to define 3 new goals and the overall vision of the Consortium by the year 2027: *Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention*

- The Working Group on Goal 3 was established to identify what are the specific needs, metrics and tools for addressing the third goal of IRDiRC: ***Develop methodologies to assess the impact of diagnosis and therapies on rare disease patients***

- The members of the Working Group on Goal 3 met on February 19-20, 2020 in Paris to analyze how access to diagnostics and therapies impact the economy of health care systems and the socio-economic burden on rare disease patients and families

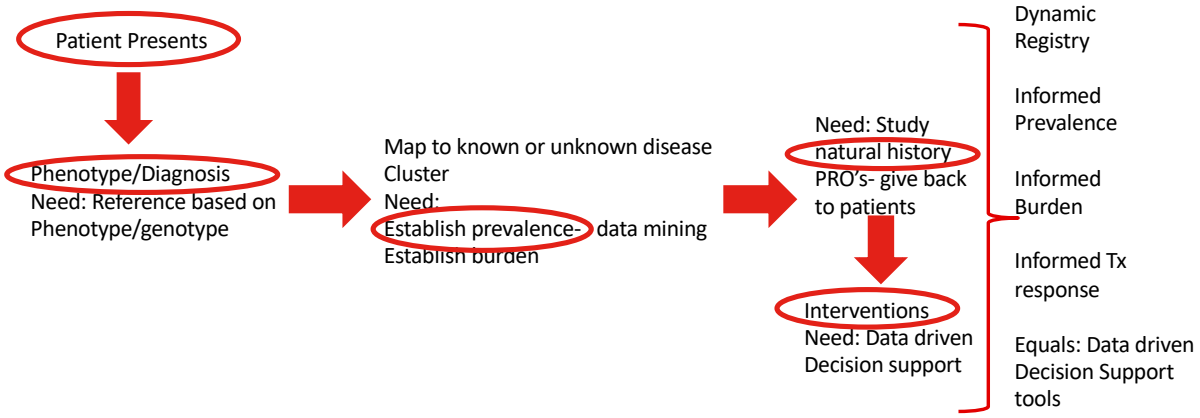
- The members of the WG3 have a wide background of expertise:
    - HTA and health economics
    - Patient advocacy
    - Genetics and diagnosis, health care intervention, population health
    - Drug evaluation and value measurement
    - Registries and epidemiology
    - Health policies and reimbursement policies

Graph 1 represents what is the patient perspective with respect to diagnosis and therapies. This schematic served as a template to what the Working Group expanded upon in term of the needs for:

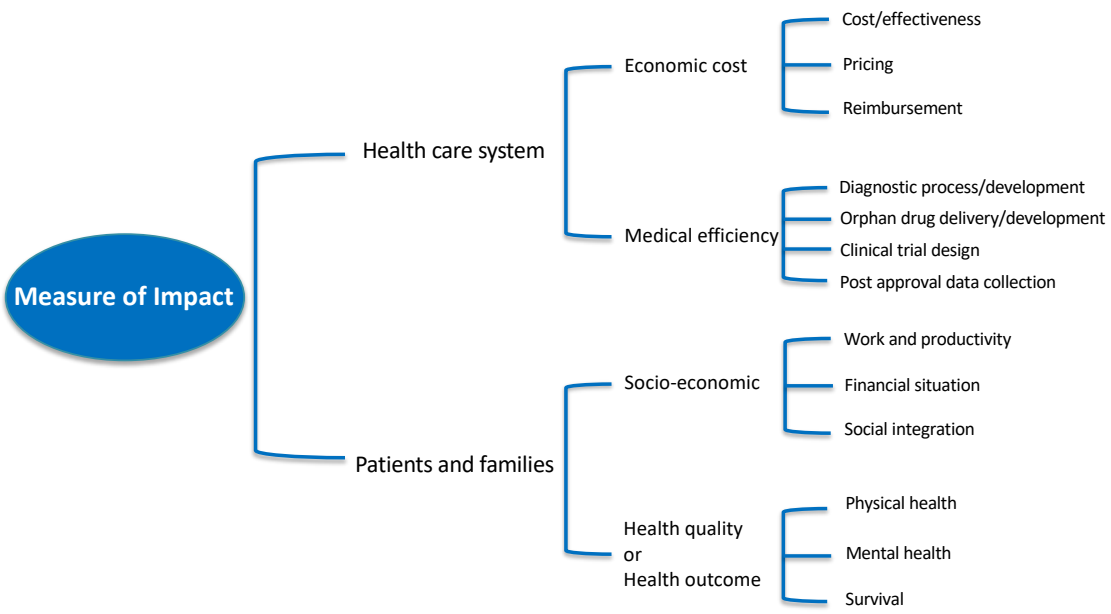
- Diagnosis
    - Need references based on phenotype/genotype.

- Mapping of the diseases (known or unknown)
  - Need to establish the prevalence and burden of the diseases.
  - Need to have access to natural history studies and patient reported outcomes.
- Intervention:
  - Need for data-driven decision support.

- And what could be the area for which data or tools are needed?
  - e.g. dynamic registries, informed prevalence, informed treatment response, data-driven decision support tools.



Graph 2 represents which measures of impact should be considered regarding impact on patients and families, and also health care systems.



- The check list represents the disease attributes that must be defined to measure a positive or negative impact on the metrics described above.

Diagnostic

- Age: Pediatric or adult
- Time to diagnose: Early (-1year) or late (+1year)
- Quality of diagnosis: No diagnosis, misdiagnosis, diagnosis

Prevalence

- Measure of prevalence at birth
- Total number of patients
- Age distribution
- Mortality

Natural History Studies

- Evaluation of the burden
- Disease progression models
- Disease registries
- Patient reported outcomes: Dynamic registries

Intervention

- PCOMs (IRDiRC Task Force)
- Standard of care
- Alternative/Innovative treatment
- Informed treatment response: Regulatory and effectiveness data

- The outcome of the Working Group work will be a white paper describing what are the specific needs, metrics and tools for measuring the impact of diagnosis and therapies on RD patients and health care systems. The manuscript should be oriented for policy makers/ regulatory agencies. The first draft should be ready in the summer 2020.

- The next steps will be a proposition to establish a Task Force that will use the recommendations developed by the WG3 to measure the impact of diagnosis and therapies in pre-selected rare diseases (e.g. well-characterized rare diseases such as cystic fibrosis, Duchenne muscular dystrophy, rare cancers, or poorly characterized ultra-rare diseases).

- Suggestions from IRDiRC members
  - While it is quite interesting to select a limited number of diseases and analyze them across the different items identified by the group, it would be also interesting to inversely select 1 or 2 items (e.g socio-economic aspect) and assess them transversally on different types of diseases.
  - It would be valuable to use some recommendations issued in the white paper to fit the 2021 EJP RD-funded research projects on Social and Human Sciences.

Indigenous Populations

The overarching goal of this activity is to improve RD diagnostics globally for indigenous populations and other underserved populations living with a rare disease by addressing existing barriers in access to diagnostic services. The output will be a review paper on the scope of the problem and a recommendation paper to deliver solutions that will help meeting the IRDiRC Goal 1.

- Task Force has a worldwide representation (20 members, 4 continents) with broad expertise:

- Clinical & medical geneticists
- Policy experts, research funders, patient advocates
- Researchers working with tribal communities, inclusiveness and indigenous aspects
- Aboriginal researchers
- Aboriginal medical students
- Data scientists
- Terminology and semantics
- Translation technology

- Task Force process and timeline:
  - The first TC was held on September 25 2019 with follow up meetings on December 19 2019 and February 26 2020
  - The face-to-face meeting of the Task Force was scheduled to March in Berlin but was cancelled
  - The State of Play document was delivered by the Sci Sec on February 2020
    - The general challenges/barriers to RD diagnostic for indigenous peoples are:
      - Lack of access to clinical genetic services
      - Remoteness of communities
      - Limited qualified health care providers in remote communities
      - Lack of genomic and phenomic reference databases
      - Under-representation in genomic research and biobanks
      - Indigenous specifics' ethical issues concerning genetic research
      - Heterogeneity in terms of language, culture and customs
    - Existing projects/initiatives addressing these barriers include:
      - Australian National Centre for Indigenous Genomics (NCIG)
      - Silent Genomes Project (Canada)
      - All of us Tribal Engagement (USA)
      - Ethical guidelines for genomic research
      - Culturally informed biobanks
      - Digital technologies for Indigenous languages (e.g. Lyfe Languages) and facial imaging (Pilbara Faces, Cliniface)
      - Genetic disorder databases for minority ethnic groups
      - Educational initiatives (e.g. SING Consortium, Songlines and weaving)
  - The next frontiers to RD diagnosis for Indigenous Peoples will involve proposing recommendations in areas for which there are opportunities for improvement, including the following:
    - Awareness and engagement
    - Historical basis and engagement
    - Indigenous patient identification
    - Primary care initiatives
    - Newborn screening
    - Congenital anomaly synergies
    - Culturally appropriate services and languages
    - Digital technologies and ontologies
    - Reference data
    - Framework and policies
    - Care principles, data sharing and sovereignty
    - Capacity building

Clinical Research Networks

- The objectives of the Task Force on Clinical Research Networks are threefold:
  - To map and analyze the existing ecosystem of national/supranational clinical research networks.
    - The output of this objective will be a definition and landscape paper on the different network initiatives and their characteristics
    - To develop policy recommendations on guiding principles for an international framework of collaboration of these networks in respect to best practices, interoperability, tools and common goals
    - To develop relevant recommendations for funders based on gaps identified through the mapping exercise.

- The Task Force group was assembled during the summer 2019
  - Chair: Steve Groft, NCATS, USA.
  - Co-Chair: Rima Nabbout, Necker Hospital, ERN EpiCare, France.

- The group includes 22 members representing 11 countries and 4 continents.

- The group gathers expertise in clinical and basic research, academia and industry, network development and governance, patient advocacy.

- Four conference calls were organized:
  - October 3, 2019
  - November 7, 2019
  - December 19, 2019
  - February 11, 2020

- The current task of the group is to identify the attributes of the different CRNs in order to map the network landscape. The group is developing a questionnaire that will be addressed to pre-identified CRNs.

- The Project Plan for year 2020 includes the following tasks:
  - Q1-Q2: Draft the questionnaire and identify the networks.
  - Q2-Q3: Conduct the survey, analyze the results and map the CRNs landscape.
  - Q3-Q4: F2F meeting (July 9-19, Brussels, if possible), validation of the CRN map, recommendations for policy makers and funders, final report and white paper.

- The next meetings of the Task Force group will be held on:
  - April 03 – Conference call
  - May 04 – Conference call
  - June 16 – Conference call
  - July 9-10 – F2F meeting in Brussels, if possible.

- Suggestions from IRDiRC members
  - It is important to synergize the discussion between the Working Group on Goal 3 and the Task Force on CRNs to provide recommendations regarding the importance to design effective clinical trials (e.g. development of international CTs including patient

perspectives and outcome measures).

3. Update on the new 2020 activities

Chrysalis project

Increasing the interest of industry in RD research is central in the development of new diagnosis and therapies for the benefit of patients.

- The objectives of the Chrysalis project are:
  - To identify key criteria that would make rare diseases research more attractive to industry for research and development (financial and non-financial).
  - To identify gaps in current funding opportunity landscape to develop criteria from #1.
- The output of this activity will be a white paper describing the identified key criteria and providing recommendations for funders.
- The identification of the Task Force members will start in March 2020.
  - A call for candidates will be published on the website and IRDiRC CA-SC members will be notified by email.
- The expected date for the first conference call with the whole group is May 2020.
- Candidates with the following expertise will be considered:
  - Funding rare disease research as a public or private entity
  - Involvement in strategic planning for rare disease research
  - Personal or family experience with rare diseases and its funding or advocacy
  - Research in rare diseases and corporate/finance experience
  - Translation of research results into clinical settings

The Chrysalis project is a cross committee initiative involving the FCC and CCC. The Task Force leadership is under modification while the CCC achieves its transition (i.e. identification of a new Chair and Vice Chair)

- Suggestions from IRDiRC members:
  - Engaging CCC members in this activity is a very good idea but the Task Force leaders could also identify members from specific companies that can contribute in the project. In addition, this could be an entry point for these companies to join IRDiRC.
  - Additional expert profile: People engaged in technology transfer offices having experience in interacting with companies.

Shared Molecular Etiologies

The identification of strategies for grouping patients based on the common molecular etiology of their RD will support the development of clinical trials involving a larger number of patients and evaluating the effect of drug targeting common molecular pathways.

- The objectives of this Task Force are:
  - To assess the global landscape for development of drugs targeting shared molecular etiologies (SMEs).
  - To develop a framework for basket trials of drugs targeting SMEs underlying multiple rare diseases, based on the success of tissue-agnostic basket trials in oncology.
  - To identify potential regulatory roadblocks to such rare disease basket trials, and solutions to overcome them.
  - To explore strategies to identify patients that would be eligible to participate in SME clinical trials.

- The output of this Task Force will be a white paper describing the challenges in the conduct of clinical trials or drugs targeting SMEs underlying multiple RD, and potential recommendations to funders.

- The identification of the group members should start in April/May 2020. A call for candidates will be published on the website and IRDiRC CA-SC members will be notified by email. The expected date for the first conference call with the whole group is September 2020.

- Candidates with the following expertise will be considered:
  - Representatives from regulatory agencies.
  - Specialists in clinical trial design and data analytics.
  - Biostatisticians with expertise in designing small population CT, and basket trials, and trials involving products with shared molecular etiologies and pathways.

- Suggestions from IRDiRC members:
  - It would be valuable to engage experts who analyzed why some of the basket trials in oncology failed.
  - Some of the challenges in shared molecular etiologies drug targeting are upstream clinical development (e.g. assay development); it should be relevant to involve participants working on pre-clinical phase.
  - It would be interesting to explore how innovative therapies (e.g. gene therapies) affect the implementation and outcome of basket trials.

4. Database of funded projects

- Recap
  - FCC action (Activity A): To establish process to coordinate and prioritize research funding efforts
  - The main goals of this activity are:
    - To put in place the tool (database) allowing in depth analysis of funded projects and rare diseases research funding landscape at international level
    - To establish processes whereby funders can coordinate and prioritize efforts on research and avoid unnecessary duplication
  - Project timeline:

- Call for tenders launched in July 2018
- MyScienceWork (MSW) selected in Nov 2018
- First survey to identify methods of data collection sent out to FCC members in Feb 2019
- First demonstration of the tool provided at the FCC meeting in May 2019, Leiden
- Second survey to enable automated data capture sent out to selected FCC members in June 2019
- First translation test from Dutch, German and Japanese to English finalized in Mar 2020

- Presentation of the Database by Ana Rath (Orphanet) and Manuel Guzman (MSW)
  - Orphanet maintains a qualified, curated and quality-controlled database of RD research-related resources, including research projects and clinical trials (CTs) for the countries covered by the Orphanet network
  - As past coordinator of the IRDiRC Sci Sec, Orphanet also collects research projects and clinical trials data from IRDiRC funders located in countries for which there is no Orphanet national team
  - A collaboration between World Health Organization's ICTRP (International Clinical Trials Registry Platform) and Orphanet is in place for information on CTs
  - The database being developed by MSW will provide a web analysis interface to both Orphanet and IRDiRC members. The tool will enable to analyze data on RD research projects and clinical trials since 2010, and a comparison of data sets across projects and clinical trials (see below)

Live demonstration of the web interface for RD research landscape analysis:

- Analytics dashboard:
  - Research projects (N of validated projects in the database)
    - Filter options:
      - Project state (where in the validation process the project is)
      - IRDiRC projects (funded by IRDiRC members or not)
      - Type (e.g., multinational/national)
      - Funding institution
      - Country
      - Category (e.g., gene search, biomarker development)
      - Supra-category (e.g., basic research, clinical observational, pre-clinical)
      - Disorders
    - Results by:
      - Projects by type
      - IRDiRC funded vs non-IRDiRC funded projects
      - Proportion
      - Distribution of projects and disorders by medical domain (based on the Orphanet classification)
      - Categories of research within each supracategory
      - Research type (category and supracategory) by medical domain
      - Top 20 diseases based on project count (projects concerning with multiples disorders, each disease is counted once)
      - Number of projects over time by year of start since the 2010

→ the same type of analysis is available for Clinical Trials except that project categories are different (also include study phase and drugs tradenames & substances) and Crossed Research Projects & Clinical Trials

- New functionalities are under development:
  - Disease coverage (number of disorders covered by medical domain)
    - New annotation has been added to the Orphan database in order to not count projects that are general and relevant to the group as a whole but not for individual diseases
  - Prevalence
  - Introduction of non-validated projects (ongoing and unpublished research projects) and substances to clinical trials

- Next steps:
  - To gather data from all IRDiRC funders
  - To check all info in the Orphanet database for latest updates and missing data
  - Otherwise the analysis interface is ready to use

- Need feedback from FCC on the options provided for filtering data according to their use and needs, and specifically:
  - Disorders: as of now not possible to select disorders by groups/subtypes
  - Top diseases by project count: showing the top 20 diseases (categorization at the disorder level)
  - Funding costs: currency is given in US dollars
- CA feedback
  - Integration with data from Orphanet knowledge base → projects are tagged with metadata from the Orphanet knowledge base
  - Proposition to enable the selection of groups but also categories of diseases
  - Rare acute infectious diseases included? → yes
  - Proposition to include as filter (1) age of onset, (2) top hundred diseases, (3) analysis with and without oncology → data on oncology can be filtered out through the Add filter option
  - Methodology for collecting data in the Orphanet database? → Through official sources when possible, learned societies, big research institutes, researchers' self-declaration through the Orphanet registration tool
  - Not possible to know the percentage of projects covered by country
  - For CTs data is coming from WHO

5. Update on the new 2020 activities (Cont.)

Rare Disease Treatment Access
(Durhane Wong-Rieger, PACC Chair)

- Scope
  - Working group led by Bill Gahl (NHGRI/NIH), Durhane WR (PACC Chair), and Susanne Weissbaecker (Global Head, Access to Medicines, Takeda)
  - Expected to start Q1 2020

- Objectives:
  - To create a list of standards of care products for RDs (e.g., drug products including generic, off-patent, and repurposed drugs, diagnostic and medical devices, guidelines) and make this list available worldwide
- To identify barriers in making treatments available in different countries, particularly in low/medium income countries
- First output will be a White Paper on RDs treatment access to be issued by late 2020
- Deviation from the original proposal validated by the CA in Nov 2019 (Paris):
  - The work will be carried out in two phases and will focus first on the list of drugs and the identification of barriers to accessing drugs worldwide

- Ongoing or foreseen steps
  - Identification of experts between Feb & Mar 2020
  - 14 experts already identified by TF proposers or self-nomination
    - Letter of invitation is being sent along with:
      - Terms of Reference
      - WHO Essential Medicines Lists and EMA and FDA approved orphan drugs
      - Template for an annotated list of 'essential' products
  - First conference call with whole group expected in May 2020
  - Face to face was planned to take place in late spring 2019
  - Connect the project to RDI-WHO Access to Essential Medicines/Diagnosis Ad Hoc Group

- Expert profile:
  - Active in rare disease research and/or clinical practice in diagnosis, treatment access, including representation from low-and-middle income countries or regions
  - Personal or family experience and/or advocacy with accessing diagnosis and/or treatment
  - Experience in regulatory approval, health technology assessment and/or reimbursement (funding) of rare disease treatments
  - Deviation from the original project validated in November 2019, if any
  - Divide in two phases and focus on first: list of drugs and the identification of barriers to accessing drugs worldwide

Integrating New Technologies for the Diagnosis of RD
(Gareth Baynam, DSC Chair)

- Scope
  - Task Force led by DSC and chaired by Sarah Bowdin (DSC Vice Chair)
  - Will build on earlier work from the DSC Task Force Solving the Unsolved (STU)
  - Expected to start Q3 2020
  - Objectives:
    - To identify new technological approaches (metabolomics methods combined with genomics and AI) likely to increase the diagnostic rate for RD patients
    - To identify opportunities to enable widespread clinical adoption of the most effective combined diagnostic approach
  - Output will be a review paper on the current state of knowledge of metabolic disease diagnostics focused on areas for which the development of diagnostic standards would maximize patient benefit

- Deviation from the original proposal validated by the CA in Nov 2019 (Paris):
  - No deviations as there has been no further discussion on this action since the last CA meeting

- Ongoing or foreseen steps
  - Identification of experts should start in April/May 2020
  - Call for nominations on the website and email to the CA and SC
  - Expected date for the first conference call with the whole group September 2020

- Expert profile:
  - Scientists with expertise in techniques for rare disease diagnosis including metabolomics, genomics, artificial intelligence and machine learning experts, translational medicine, diagnostic evaluation scientists

- CA comments
  - (Daniel Scherman, FFRD) suggested Carlos Malpica, an industry expert in metabolomics
  - Dr Malpica is the CEO of MLP Vision Biotech S.L. and currently involved in establishing Valdia Health in Qatar. For the last seven years he was actively promoting global adoption of metabolomics technologies as Global Business Development Director at Metabolon Inc. (USA)
  - (CA Chair) Since the primary focus of the TF is the clinical translation and implementation of novel technologies, inputs from experts in health system implementation is crucial

6. Strategic planning for year 2021

- Four activities are going to start in the coming months and will run up to Q1-Q3 2021:
  - Working Group on Access to Medicines
  - Chrysalis Project
  - Shared Molecular Etiologies
  - New Technologies

- Proposed work plan for 2021:
  - To start planning the 2021 roadmap in Sept 2020
  - Activity proposals to be presented at the CA meeting in Milan (October 1 & 2)
  - Actions that were not prioritized for year 2020 can be resubmitted if still pertinent
    - Primary Care
    - Alternative Business Model
    - Big Bang Project
    - PLUTO Project
  - 2021 roadmap to be validated in Q1 2021

7. Conclusion and planning of next meetings

- Teleconference of the Consortium Assembly
  - July 2020 (date to be confirmed)

- Consortium Assembly F2F meeting
  - October 1 & 2, 2020, Milan, Italy

- IRDiRC-RE(ACT) Conference and IRDiRC Task Forces meeting
  - Task Forces workshops: January 12, 2021, Berlin
  - IRDiRC Conference: January 13-16, 2021, Berlin

- Consortium Assembly & Scientific Committees F2F meeting/IRDiRC 10th Anniversary special event
  - April/May 2021, USA (tbc)

→ No meeting of the Consortium Assembly is planned for January (Berlin)

Document history

Version 1. Report drafted by Carla D'Angelo and Galliano Zanello, April 6, 2020
Version 2. Reported circulated among CA & SC members April 7, 2020

Annex - List of participants

March 12, 2020

Lucia Monaco (CA Chair)	Telethon Foundation	Italy
David Pearce (CA Vice Chair)	Sanford Research	USA
Adam Hartman (FCC Chair)	National Institute of Neurological Disorders and Stroke (NINDS)	USA
Durhane Wong-Rieger (PACC Chair)	Canadian Organization for Rare Disorders (CORD)	Canada
Yukiko Nishimura (PACC Vice Chair)	Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid)	Japan
Gareth Baynam (DSC Chair)	Western Australian Department of Health	Australia
Stephen Groft (ISC Chair)	National Center for Advancing Translational Sciences (NCATS)	USA
Dixie Baker (ISC Vice Chair)	Martin, Blanck and Associates	USA
Virginie Hivert (TSC Vice Chair)	EURORDIS-Rare Diseases Europe	Europe
Alice Chen	National Center for Advancing Translational Sciences (NCATS)	USA
Alexandre Mejat	French Muscular Dystrophy Association, AFM-Téléthon	France
Anneliene Jonker	TechMed Centre, University of Twente Enschede	Netherlands
Catherine Nguyen	National Institute of Health and Medical Research from France (INSERM)	France
Christina Kyriakopoulou	European Commission	Europe
Cindy Bell	Genome Canada	Canada
Clara D.M. van Karnebeek	Emma Chilldren's Hospital, Amsterdam	Netherlands

Daniel O'Connor	Medicines and Healthcare products Regulatory Agency	United Kingdom
Daniel Scherman	French Foundation for Rare Diseases (FFRD)	France
Daria Julkowska	European Joint Programme on Rare Diseases (EJP RD), Inserm	France
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Heikki Vilen	Academy of Finland	Finland
Jean-Pierre Gaspard	Yposkesi	France
Karen Aiach	Lysogene	France
Madhu Natarajan	Takeda Pharmaceuticals	USA
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