

# IRDIRC DECEMBER 2017 UPDATE

---



---

## Editorial

The year 2017 was a very rich and productive one for IRDiRC: 9 new members, 8 scientific publications, 6 new IRDiRC Recognized Resources, the official start of 3 Constituent Committees, and most significantly, the unveiling of 3 new goals supporting IRDiRC's new vision for the next decade.

In February, IRDiRC organized its 3rd conference in Paris, France, to celebrate the considerable progress on its original goals in the past 6 years: the goal to deliver 200 new therapies was achieved in early 2017 – three years earlier than expected – and the goal for diagnostics is within reach. It signaled the time to look ahead to new challenges and shape IRDiRC's vision for 2017-2027.

At the same time, the Constituent Committees started their activities with the mission to better coordinate, enhance information exchange and identify actions towards IRDiRC objectives. These committees are currently under the leaderships of:

- Daria Julkowska, representing the Agence Nationale pour la Recherche (ANR), for the Funders Constituent Committee,
- Sharon Terry, representing Genetic Alliance, for the Patients Advocates Constituent Committee, and
- Mathew Pletcher, representing Roche, for the Companies Constituent Committee.

In April, IRDiRC updated its governance to reflect the evolution of the Consortium and additionally empowered patient advocate groups to apply for IRDiRC membership. This call was heard as IRDiRC counts currently 6 additional patient advocacy organizations as members, in comparison to 3 in 2016.

In August, concomitantly with the publications of 3 vision and goal articles ([1](#), [2](#), [3](#)), IRDiRC announced its new vision and goals for the next decade via a press release that was widely disseminated to the members' network. Additionally, IRDiRC also published articles on its [Policies and Guidelines](#), [A Global Approach to Rare Diseases Research and Orphan Products Development](#), [International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases](#), and the [position statement of the Patient-Centered Outcome Measures Task Force](#), while contributed to some others, as on [the importance of international collaboration for rare diseases research](#). More publications are currently in preparation to further disseminate the activities of the Consortium.

With the new goals in place, the Constituent and Scientific Committees set out to conceive and propose actions to support and advance these objectives. The first steps in the creation of the new roadmap for IRDiRC were presented at the Consortium Assembly meeting that took place last November in Tokyo, Japan. The Consortium is currently in the process of analyzing and prioritizing the proposed actions, and aims to deliver an aspirational and pragmatic roadmap in early 2018.

Through 2017, IRDiRC welcomed a total 9 new members. Among the 54 members, 31 participate as funders, 14 for companies, and 9 patient advocacy organizations. Importantly, the global coverage of the Consortium also expanded as IRDiRC welcomed its first members in India and South Africa while strengthening its presence in Australia, Canada, China, Japan, UK and USA. IRDiRC looks forward to welcoming more members, in particular from regions that are currently under-represented, ensuring their vital voice, experience and input are integrated into global rare diseases research agenda, bringing diagnostics and therapies to patients globally.

---

## Spotlight on the position statement of the Patient-Centered Outcome Measures Task Force

In the *Orphanet Journal of Rare Diseases*, Thomas Morel and Stefan Cano published a position statement based on the work and discussions of the IRDiRC Task Force on Patient-Centered Outcome Measures (PCOMs). The article *has since been accessed over 2500 times. Positively received, it was featured several times: for instance by [Rare Disease Report](#), [Charcot-Marie-Tooth News](#), [Angelman Syndrome News](#) and [Alport Syndrome News](#). More recently, Daniel S. Levine [interviewed Thomas Morel for RARECast](#), a Global Genes podcast. Additionally, *Global Genes* also published news on the article entitled "[Lost in Translation](#)".*

---

## Featured article

### Data Sharing from Clinical Trials – A research Funder’s perspective

An article by Kiley et al. in the *New England Journal of Medicine* presents the specific vision of research funders (Wellcome Trust, Medical Research Council, Cancer Research UK, and Bill and Melinda Gates Foundation) for optimizing the value of data produced in studies, especially clinical trials, that they funded. As the need in medical research is always greater than the resources, the authors emphasize on the importance of “collaboration and cooperation among members of the global research community to maximize the effect of funded research”. However, to support this vision, funders of research have to actively incentivize and support data sharing.

There is currently a consensus that clinical trials results should be shared in a timely manner, with consideration of participants’ privacy and confidentiality; this is supported by a [joint statement of the World Health Organization](#). The authors, as funders, identified three main challenges to data sharing: resources, equity and incentives.

As data sharing is not a cost-free process, funders need to support technical solutions to help researchers to access and (re)use data. Additionally, funders are increasingly asking researchers to include data management plans in their research proposals and the costs of these plans in the funding budget. To tackle concerns on inequity of data sharing for low- or middle-income countries, the authors suggest inclusion in data-access governance specific terms that “require users to contribute to training and capacity development or to share the resulting outcomes”.

The main challenge to data sharing remains that many researchers believe sharing can be disadvantageous for them. As such, funders must demonstrate the value of data sharing as well as other

outputs (e.g. material or software) by taking them into account in the grant review process. Additionally, to understand the significance of a study in its field, the scientific content of an article should be re-valued compared to its publication metrics. The idea is somehow to redefine criteria used for the grant application process but also maybe to ask applicants to demonstrate how they support the values of open research. Finally, the authors support the idea of “data authorship” where researchers would be cited when their data are (re)used.

These lines of thought are essential to ensure the use of research data to their fullest potential and even more so in the case of rare disease where data are more scattered.

For details, please read the [full article](#).

---

## Spotlights on IRDiRC Member Organizations

- EURORDIS–Rare Diseases Europe launched a new position paper calling [for urgent change to ensure patients' full and fast access to rare disease therapies in Europe](#).
- The Food and Drug Administration (FDA) [recommends new, more efficient approach to drug development for rare pediatric diseases](#).
- FDA advisors gave, for the first time, positive reviews for a [gene therapy treatment to target mutation in RPE65 causing hereditary blindness](#).
- NORD issues [statement in response to Senate vote on the Orphan Drug Tax Credit](#).
- Ned Sharpless, the new Director of the National Cancer Institute (NCI), expects [big data to revolutionize cancer research, care](#).
- The European Commission published its [P4P report \(project for policy\) about rare disease](#) demonstrating how the results of recent research and innovation (R&I) projects funded by the EU contribute to policy challenges related to rare diseases.
- The European Commission has [defined a uniform standard for the collection of data on rare diseases in Europe](#), as a first step towards ensuring that European data registries is comparable and interoperable.
- Cydan II announced [a new round of financing, raising a committed total of \\$34 million to advance the company's efforts to create new and innovative therapies for patients living with rare genetic diseases](#).
- Recursion Pharmaceuticals has been featured in the [AI 100, a list of 100 companies that are using artificial intelligence to redefine their industries](#).
- Recursion Pharmaceuticals was listed as [Top 15 Fierce Medtech for 2017](#).
- [ONCASPAR®, from Shire, received Marketing Authorization in Europe](#) for patients with Acute Lymphoblastic Leukemia (ALL).
- Ionis Pharma evaluated the action of [antisense oligonucleotides targeting PMP22 and showed promising results for the treatment of Charcot-Marie-Tooth Disease](#).

## Rare Diseases Research Highlights

- The European data for life science infrastructure, ELIXIR, collaborates with the Global Alliance for Genomics and Health (GA4GH) to [develop and promote standards and frameworks for the responsible sharing and reuse of genomics data](#).
- A study published in *Trials* highlights the [applicable opportunities to improve evidence-based clinical practice for rare diseases](#).
- The European Reference Networks and RD-ACTION are tackling challenges concerning [Clinical Practice Guidelines](#).
- [Clinical Patient Management System \(CPMS\)](#), a web-based application where healthcare professionals from the European Reference Networks (ERNs) will be able to discuss real patient cases, has just been launched by the European Commission (DG SANTE).
- An article published in the *Journal of American Medical Association* aims to show that [precision medicine diagnosis and risk assessment is moving medicine towards a deeper understanding of health and disease](#).
- ThinkGenetic with FDNA's Face2Gene combines [artificial intelligence diagnostic aid to help undiagnosed patients find answers](#).

---

## Events

- [Rare Disease Day 2018](#) is taking place on February 28, 2018. The theme will once more be “Research”.
  - The [RE\(ACT\) Congress](#), co-organized by the Blackswan Foundation and E-Rare, will take place on March 7-10, 2018 in Bologna, Italy. New abstract deadline: **January 22, 2018**
  - The [Genomics of Rare Diseases \(GRD\)](#) Conference 2018, organized by the Wellcome Genome Campus, will take place on March 26-28, 2018 in Cambridge, UK. Abstract deadline: **January 30, 2018**
  - The [European Conference on Rare Diseases and Orphan Products \(ECRD\)](#), organized by EURORDIS-Rare Diseases Europe, co-organized with Develop Innovate Advance (DIA) and Orphanet, is taking place in Vienna, Austria on May 10-12, 2018. Abstract deadline: **January 31, 2018**
- 

## IRDIRC-Related Funding Calls

The NIH has launched several calls for “[Ethical, Legal, and Social Implications \(ELSI\) of Genomics Research Project Grant Program](#)”. This Funding Opportunity Announcement is to support studies of the ethical, legal and social implications (ELSI) of human genome research.

- Call R01 is dedicated projects using either single or mixed methods. Proposed approaches may include but are not limited to data-generating qualitative and quantitative approaches, legal, economic and normative analyses, and other types of analytical and conceptual research methodologies, such as those involving the direct engagement of stakeholders. Application deadline: **February 5, 2018**
- Call R03 is dedicated to small, self-contained research projects, such as those that involve single investigators. Of particular interest are projects that propose normative or conceptual analyses, including focused legal, economic, philosophical, anthropological, or historical analyses of new or emerging issues. Application deadline: **February 16, 2018**
- Call R21 is dedicated to projects using single or mixed methods studies that break new ground, extend previous discoveries in new directions or develop preliminary data in preparation for larger studies.. Application deadline: **February 16, 2018**

E-Rare has launched its [10th Joint Call for European Research Projects on Rare Diseases](#). The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear translational research approach. Application deadlines: **February 6, 2018** for the pre-proposals, and **June 19, 2018** for selected full proposals

The AFM Telethon has launched a call for [scientific proposals for PhD fellowships](#). This international call for proposals, open to both French and foreign groups, aims to support research which will increase understanding of neuromuscular system, and encourage the development of therapies for neuromuscular and rare genetic diseases. Application deadline: April 24, 2018

For more IRDiRC-related calls, please consult [this page](#).

---

## Upcoming Teleconferences and Meetings

- January 10, 2017 – Operating Committee – Teleconference
  - January 16, 2017 – Consortium Assembly Meeting – Teleconference
  - March 25, 2018 – Solving the Unsolved Task Force Workshop – Hinxton, UK
  - May 14-15, 2018 – Scientific Committees Meeting – Vienna, Austria
  - May 15-16, 2018 – Consortium Assembly Meeting – Vienna, Austria
-

If you are presenting in a meeting or a conference, and you would like to include some information about IRDiRC, standard slides are available on the IRDiRC private website; additional slides can be made available upon request.

Please also email the Scientific Secretariat when and where you will be presenting, so we can keep track of "IRDiRC presence at conferences."

---

*Marlène Jagut, Communication Manager*

*IRDiRC Scientific Secretariat, IRDiRC, Plateforme Maladies Rares / Rare Diseases Platform, 96 rue Didot, 75014 Paris, France, Tel: +33 1 56 53 81 37, Fax: +33 1 56 53 81 38*

*For more information on IRDiRC and its activities, please visit the [IRDiRC website](#). Stay up to date with news regarding IRDiRC and the rare diseases research community by following [@IRDiRC](#) on Twitter.*