

# IRDIRC APRIL 2017 UPDATE

## A European perspective highlights the value of multi-national collaborations

In a recent review published in the *Gene Therapy*, Julkowska *et al.* called attention to the accelerated advances made in rare diseases research when multi-country stakeholders pool together resources and expertise. The many EU-centric collaborative actions described (including scientific endeavors, publications, patent applications and spin-off companies) also illustrate models for building international networks of researchers that enable cross-border education and training, foster the pooling of scarce and valuable resources, and support infrastructures that facilitate research and development in the field of rare diseases, all the way from bench to bedside. Gene therapy is one of the many areas that has reaped the benefits of transnational collaborations, bringing vital therapeutic options to rare disease patients. Beyond Europe, participation in IRDiRC brought forward additional centralization of efforts that aim to maximize the impact from the investments made at the international level.



### The importance of international collaboration for rare diseases research - a European perspective

Julkowska *et al.*, 2017 - Gene Therapy [article](#); PMID: [28440796](#)

## Spotlights on IRDiRC and its Members

- European Commission's DG SANTE wins the [EU Ombudsman Award](#) for EU collaboration on rare diseases
- New step towards the treatment of [myotubular myopathy](#): gene therapy restores strength and prolongs lives in affected dogs
- [Advancing Rare Diseases R&D: A Mission Update](#) - Dr Christopher Austin interviewed by the PharmExec Magazine
- Raising awareness: April 29th is [Undiagnosed Day](#)

## Rare Diseases Research Highlights

- Risky business? Rare disease patients and caregivers [willing to endure certain side-effects](#) for better drug response
- [Rare disease policy in 11 countries](#): an analysis
- [Tell OECD how health systems can improve sustainable access](#) to innovative pharmaceutical therapies
- European Medicines Agency releases framework and action plan for [increased collaboration with academia](#)

## New IRDiRC Documents

- Consortium Assembly - [Executive Summary](#) of the 3rd Consortium Assembly meeting - February 6, 2017, Paris, France
- Joint Scientific Committees - [Report](#) of the 3rd Joint Scientific Committees meeting - February 7 (AM), 2017, Paris, France
- Joint Scientific Committees and Consortium Assembly - [Executive Summary](#) of the 2nd Joint Scientific Committees-Consortium Assembly meeting - February 7 (PM), 2017, Paris, France

## Upcoming Teleconferences and Meetings

- May 3, 2017 - Operating Committee - Teleconference
- May 22, 2017 - Interdisciplinary Scientific Committee - Teleconference
- May 22, 2017 - Therapies Scientific Committee - Teleconference
- May 29, 2017 - Privacy-Preserving Record Linkage Task Force - Teleconference
- June 7, 2017 - Operating Committee - Teleconference
- June 12, 2017 - Consortium Assembly - Teleconference
- November, 2017 - Consortium Assembly - Face-to-face meeting in Tokyo, Japan

## IRDiRC-2017 Conference Update

### Presentations of the 3rd IRDiRC Conference are online

The 3rd IRDiRC Conference took place in Paris, France, on February 8-9, 2017. The powerpoint presentations of the speakers, organized by session, are now available [online](#). The conference report will also be made available shortly.

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## IRDiRC-related calls

AFM-Téléthon has launched their annual call for [Research and Trampoline Grants](#). This international call for proposals aims to support research which will increase our understanding of the neuromuscular system and encourages the development of therapies for neuromuscular diseases and rare genetic diseases. Application deadline: July 4, 2017

The National Institute of Arthritis and Musculoskeletal and Skin Diseases has launched a funding opportunity announcement entitled: [Small Business Innovation Research on Rare Musculoskeletal, Rheumatic and Skin Diseases](#). This announcement solicits Small Business Innovation Research grant applications from small business concerns that propose preclinical studies to advance the development of biomarkers or treatments for rare musculoskeletal, rheumatic or skin diseases. Letter of intent due date: June 10, 2017.

The National Organisation for Rare Disorders (NORD) has announced the availability of seven new [research grants](#) to study rare diseases. Grants are available for the study of alveolar capillary dysplasia with misalignment of the pulmonary veins, appendix cancer and pseudomyxoma peritonea, cat eye syndrome, malonic aciduria and post-orgasmic illness syndrome. Application deadline: June 23, 2017

## Featured Summary Report

### An opportunity: old drugs for rare diseases

In addressing the topic of drug repurposing for rare diseases, IRDiRC recently collaborated with B.Debate to discuss the importance of repositioning molecules thus increasing the number of therapies available to rare disease patients. The summary of the 2-day meeting is [now available](#), highlighting the gap where 90% are currently without approved treatment, which could be partially addressed through drug repurposing that typically requires shorter development time at lesser costs; some success stories were presented. However, the issues of (reduced) incentives, heterogeneity of rare diseases and geographical spread of patients hamper the interest of investments; a potential strategy to overcome the latter is through the use of networks of patients, for patients, cultivated through social media.

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## Other news

If you are presenting in a meeting or a conference, and you would like to show 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."

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*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.*