

# IRDiRC SEPTEMBER 2017 UPDATE

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## A new Toolkit for Patient-Focused Therapy Development



National Center  
for Advancing  
Translational Sciences

The [National Center for Advancing Translational Sciences \(NCATS\)](#) released its new [Toolkit for Patient-Focused Therapy Development](#) at a public meeting on the NIH campus on September 8. The toolkit is a centralized portal of the "how-to" resources and tools that patient support organizations can use to advance translation for their diseases, from discovery through clinical trials, to regulatory and industry interactions, to post-approval access issues. It guides patients and patient organizations by giving them the means to engage in research, drug development and post-approval issues themselves. While the resources are primarily focused on rare diseases, the tools are useful for all diseases.

This toolkit presents several advantages:

- Collaborative creation of a well-designed source for online educational and informational research resources and tools.
- Provision of a single online portal with resources that patient groups can readily access along with context.
- Improved coordination rather than re-creation of existing resources.
- Identification of gaps in online resources, and information dissemination to patient groups.
- Promotion of continuity across the lifecycle of the drug development process.

This toolkit was developed in close collaboration between the rare diseases patient advocacy community and the NCATS' Office of Rare Diseases Research. In this month's NCATS [Director's message](#), Dr Austin, Director of NCATS and Chair of the IRDiRC Consortium Assembly, explained that it was one of his first directives **"to involve patients in every project we do from the beginning, and to develop the science of patient and community engagement, with best practices that can be disseminated to all doing translational research."** This toolkit is a step towards that goal.

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## New IRDiRC member

IRDiRC welcomes the Indian Organization for Rare Diseases (I-ORD).

India counts more than 100 million patients with rare diseases. Unfortunately, the government has not yet developed public policy on rare disease or orphan drug. To change this situation, [Indian Organization for Rare Diseases \(I-ORD\)](#) was launched in 2005 with 3 major objectives:

- Raising the awareness of rare diseases
  - Advocating public policy
  - Encouraging pharma/biotech to develop orphan drugs
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## Special attention to Sharon Terry

Sharon Terry, Chair of the IRDiRC Patient Advocates Constituent Committee, is President and CEO of [Genetic Alliance](#), a network of more than 10,000 organizations (disease-specific organizations, universities, hospitals, policymakers, companies). She is also the founding CEO of [PXE International](#), a research advocacy organization for the



genetic condition pseudoxanthoma elasticum (PXE), which affects her two children. Last November, she gave an inspiring TED talk called "[Science did not understand my kids' rare disease until I decided to study it](#)". She explained how citizens can contribute to science and make a difference by changing biomedical research. She emphasized the importance of international communities of rare disease patients and data sharing. While it is a long and complicated path, Sharon affirmed **"If we are open to breathing into our fear and being vulnerable with those systems and people who challenge us, our power as changemakers grows exponentially"**.

Last week, the video of Sharon's talk **hit the million view mark**, demonstrating the interest in rare disease and citizen science.

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## Spotlights on IRDiRC Member Organizations

- The Food and Drug Administration announced its first approval of a [promising type of immunotherapy called CAR-T cell therapy for kids and young adults](#) with B-cell acute lymphoblastic leukemia (ALL).
- The Food and Drug Administration released a video to inform patients, patient advocates, caregivers, scientists and others who have an interest in rare diseases about [the importance of natural history studies](#).
- The RD-Connect [Genome-Phenome Analysis Platform](#) continues to evolve and improve, through the incorporation of new annotations, features, and functionalities.
- The European Commission opened a [public consultation on Transformation of Health and Care in the Digital Single Market](#). Deadline: October 12, 2017.
- EURORDIS-Rare Diseases Europe opened the [nominations for the EURORDIS Black Pearl Awards](#). Deadline: October 20, 2017.
- The AFM Telethon and its laboratory Genethon are launching their [first clinical trial for a gene therapy treatment of X-Linked Myotubular Myopathy](#).

## Rare Diseases Research Highlights

- An interesting annual review entitled "[Sharing Data to Build a Medical Information Commons: From Bermuda to the Global Alliance](#)" has just been published in the journal *Annual Review of Genomics and Human Genetics*.
  - As part of a new five-year award from the National Human Genome Research Institute, [RTI International will expand the PhenX Toolkit](#), a web-based catalog of recommended measures for phenotypes and exposures for use in biomedical research.
  - A study published in the *Orphanet Journal of Rare Diseases* examines [the factors affecting information and knowledge exchange and underlines the role that the European Reference Networks will play in this exchange](#) in the context of the Cross-Border Healthcare Directive (2011/24/EU).
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## IRDiRC-Related Funding Calls

The NIH has launched a call entitled "[Palliative Care Needs of Individuals with Rare Advanced Diseases and Their Family Caregivers](#)". This funding opportunity announcement seeks to expand knowledge and increase the evidence base for palliative care in advanced rare diseases, including rare cancers, and to improve physical and psychosocial well-being and quality of life among seriously ill individuals and their family caregivers. Application deadline: **October 16, 2017**.

The French Foundation for Rare Diseases has launched call for proposals dedicated to "[Preclinical Research in Rare Diseases: translational steps in large animals](#)" to support the development of experimental models and the evaluation of therapeutic strategies. The call for proposals aims to directly support intermediary key steps towards clinical development for patients. Application deadline: **October 24, 2017 at 5pm (Paris time)**.

The ANR is launching its [generic call for proposals for the year 2018](#). It is open to all scientific disciplines and all types of research, from the most fundamental projects to applied research conducted as part of a partnership with a company, especially SMEs and very small businesses. This call corresponds to the principal "Research and innovation" component of the Work Programme 2018, which has been simplified and is now structured by research theme. Application deadline: **October 26, 2017 at 1pm (Paris time)**.

The NIH has launched a call entitled "[Clinical Observational Studies in Musculoskeletal, Rheumatic, and Skin Diseases](#)". This call is to encourage research project grant applications to pursue clinical observational studies to obtain data necessary for designing clinical trials for musculoskeletal, rheumatic, or skin diseases or conditions. Application deadline: **November 1, 2017**.

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

## Featured Article

### Treating rare disorders: time to act on unfair prices

This month, the *Lancet Neurology* published an editorial on drug pricing, citing nusinersen, the first disease-modifying treatment for spinal muscular atrophy (SMA) as an example. Even if this treatment has been approved by both the US Food and Drug Administration and the European Medicines Agency, nusinersen is far from being widely used by patients.

Nusinersen is an antisense oligonucleotide and costs US\$125,000 per injection. Patients following this treatment need six injections the first year and three injections per subsequent year. The price for nusinersen is in line with the prices of other orphan drugs such as the US\$702,000 per year therapeutic for Batten disease or the US\$300,000-US\$750,000 per year therapeutic for Duchenne muscular dystrophy. Even if prices may vary depending on countries, it is clear that **the treatment is unaffordable for most patients**.

Nusinersen is now available worldwide via the manufacturer's Expanded Access Program (EAP). It provides "investigational therapies to patients who are not eligible or able to participate in a clinical trial – but who have a serious or life-threatening diseases for which their physician has determined there are no appropriate treatment options". With regards to SMA, only patients with an infantile onset (type 1) are eligible to be part of the EAP. In addition, not all health insurance will cover the treatment costs for SMA patients with late onset since the results of the clinical trials were less promising.

No matter if a drug is intended to treat hundreds or thousands of patients, its development is always expensive. However, as more and more molecules are developed or modified to receive an orphan drug designation, the authors stressed the importance of discussing fair drug pricing issues.



The development of drug for rare diseases is a major public health issue as most rare diseases still do not have a treatment. Last March, in its statement position, **the American Academy of Neurology (AAN) called for "price negotiations, transparency in drug pricing, and reimportation of high-quality drugs from Canada when these are cheaper than in the USA"**. The authors mentioned that the current prices for the treatment of rare diseases would be impossible to bear by any health care system and called for all stakeholders (health insurance, companies, policymaker) to coordinate their efforts to make affordable health care a priority.

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

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## Upcoming Teleconferences and Meetings

- October 6, 2017 - Consortium Assembly - Teleconference
  - October 12, 2017 - Patient Advocates Constituent Committee - Teleconference
  - October 12, 2017 - Diagnostics Scientific Committee - Teleconference
  - October 26, 2017 - Therapies Scientific Committee - Teleconference
  - November 1, 2017 - Operating Committee - Teleconference
  - November 6, 2017 - Interdisciplinary Scientific Committee - Teleconference
  - November 10-11, 2017 - Consortium Assembly - Face-to-face meeting in Tokyo, Japan
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## Others news

Dynamically supporting IRDiRC is the key mission of the Scientific Secretariat. As the Consortium grows and sets its new vision and goals, the Secretariat also evolves in its footsteps.

A number of organizational changes were recently made to better serve IRDiRC members and committees:

- Anneliene Jonker takes on the role of Project Manager and administers the SUPPORT-IRDiRC contract
- Marlène Jagut joins the team as Communication Manager to widely disseminate the work of the Consortium
- Driss el Moustaine remains the Data Manager in charge of research data collection and analyses
- Recruitment is also underway to further reinforce the support team

Additionally, Lilian Lau, in tandem with Christine Cutillo, assists the Chair on partnership and political development of the Consortium, and continues to work closely with IRDiRC and the Scientific Secretariat on specific projects.

Together, the team aims to diligently advance the actions of the Consortium and help achieve its aspirational goals.

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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."

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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.*