

IRDiRC AUGUST 2017 UPDATE

IRDiRC is proud to announce its new vision and goals for 2017-2027

IRDiRC, officially launched in 2011, was originally conceived with two main goals: to contribute to the development of 200 new therapies and the means to diagnose most rare diseases by the year 2020. The last six years have seen considerable progress on these goals: 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. These accomplishments were celebrated at the 3rd IRDiRC Conference in Paris, France in February 2017.



In light of this, IRDiRC initiated a year-long collaborative process to devise a new set of global rare disease goals for the upcoming decade. IRDiRC aims to accelerate progress in the short-term with three goals for the Consortium, and ambitiously push the limits of what is currently possible in the longer term with an audacious vision for the field, all with rare disease patients' lives in mind.

The new vision: 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.

In order to work towards this bold and ambitious vision, IRDiRC has set three goals for the next decade:

1. All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

2. 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

3. Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.

The progress on the previous goals has shown that the international rare diseases research community is eager to share knowledge and experience, and work collaboratively across borders in order to bring diagnoses and therapies to patients. These new goals can only be achieved with fundamental changes to the way science is conducted, shared, and applied to the care of rare disease patients. IRDiRC members have committed to catalyze such changes and we hope that others will share and help with this commitment to action. It is time to build new bridges and raise the bar for rare diseases research worldwide.

For more detailed information on the past progress and future actions, please read three papers that have just been published by IRDiRC:

- [Nature Commentary](#)
- [CTS Past Perspective](#)
- [CTS Future Perspective](#)

We would also like to thank all of you, IRDiRC members, who helped us spreading the message!

Spotlights on IRDiRC Member Organizations

- Dr Christopher Austin, Director of the National Center for Advancing and Translational Sciences (NCATS), describes [NCATS' strategy to overcome the challenges to move stem cells therapies to a clinical phase](#)
- Dr Daria Julkowska, Scientific Coordinator on rare diseases at the French National Research Agency explains how [a new EU-wide approach to funding rare disease research could help patients secure access to new treatments](#)
- EURORDIS-Rare Diseases Europe has published the [results of the first European investigation on the social impact of rare diseases](#)

Rare Diseases Research Highlights

- For the first time in the USA, scientists of the Oregon Health and Science University [have edited gene with a deleterious mutation in human embryos](#)
 - The European Research Council supports the MUSCLEGUY research initiative to develop a [3D drug screening system to reveal new disease pathways and treatments found faster than by existing methods](#)
 - [Orphanet has just published its activity report for 2016.](#)
 - A new software called [Mendel. MD. has been described as a more accurate and agile method to investigate and diagnose Mendelian disorders](#)
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IRDiRC-Related Funding Calls

The French association [AFM - Téléthon has launched a scientific calls for postdocs grants](#). This international call for proposals, open to both french and foreign groups, aims to support research which will: A- Increase our understanding of neuromuscular system and B- Encourage the development of therapies for neuromuscular diseases and rare genetic diseases. Application deadline: **September 5, 2017.**

The ANR and the Network of Centres of Excellence in Neurodegeneration (CoEN) have launched [a Transnational Call aimed at supporting transnational collaborations in the field of neurodegenerative disease](#). Seven countries from the European Research Area and abroad are joining this call: Canada, France, Germany, Ireland, Italy, Spain and the United Kingdom. The objective of this call is to stimulate new and unconventional approaches and creative solutions to the challenges of neurodegeneration research by undertaking high-risk / high-payoff research. Application deadline: **September 18, 2017.**

The French Foundation for rare diseases has launched a [call for proposals dedicated to 'High throughput screening of therapeutic molecules' to support projects aimed at identifying molecules with potential benefit in the treatment of rare diseases](#). Projects will be based on a high throughput/content screening (HTS/HCS) approach using compound libraries, towards the discovery of active molecules 'hits' with therapeutic potential. A second step towards investing into the 'hit to lead' process to approach drug-like characteristics will be supported. Application deadline: **September 19, 2017.**

The Academy of Finland has launched [its autumn calls for grants funding](#). Grants can be allocated to principal investigators, researchers, postdoctoral researchers. The Academy Project funding scheme is designed to promote the quality and diversity of research, scientific impact and impact beyond academia as well as science self-renewal. The aim is to attain internationally as high a scientific standard of work as possible and to support scientific breakthroughs and top-tier international research collaboration. Application deadline: **September 26, 2017.**

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

Featured Article

Japan's Initiative on Rare and Undiagnosed Diseases (IRUD): towards an end to the diagnostic odyssey

Japan has a long history of tackling hard to diagnose rare diseases (called Nan-Byo in Japanese for “difficult illness”, literally). The first public definition for Nan-Byo appeared in 1972. Since then the Ministry of Health, Labour and Welfare (MHLW) has played a key role in supporting Rare Diseases (RD)

research, enabling the number of designated Nan-Byo to rise from 4 in 1972 to 56 in 2014, and is expected to be 330 in 2017. Despite these efforts, a vast majority of Nan-Byo patients are still struggling to obtain a diagnosis and eventually a treatment; known metaphorically as “the diagnostic odyssey”. A network has now been created to ensure systematic diagnosis by medical experts through phenotypic and genetic data matching.

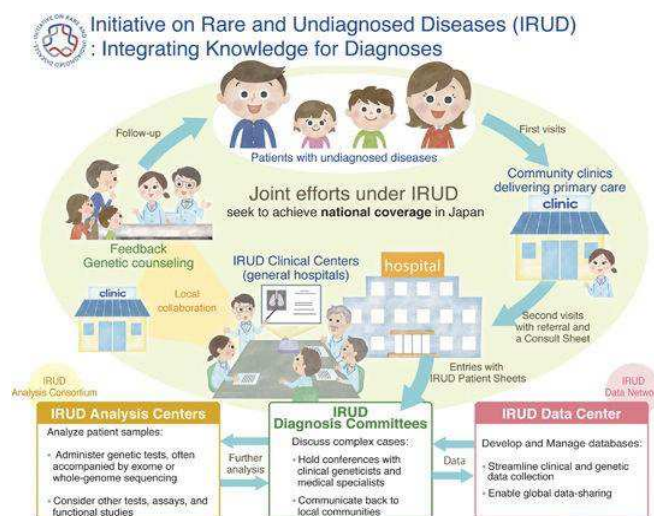
In collaboration with RD researchers and the Japanese universal healthcare system, the Japan Agency for Medical Research and Development (AMED) launched the **Initiative on Rare and Undiagnosed Diseases (IRUD) in 2015**. The rationale here is to accelerate the pioneering efforts made within the international community such as the Undiagnosed Diseases Program/Network (UDP/UDN) in the USA, the Finding of Rare Disease Genes (FORGE) in Canada, and the Deciphering Developmental Disorders (DDD) in UK. Japan also strongly supported for years the development of Next Generation Sequencing (NGS), thus facilitating the use of whole-exome and whole-genome analyses to diagnose RD patients.

For two years, IRUD has built (and will continue) a growing network composed of medical experts and the patients themselves. These efforts have led to the construction of a nationwide medical research consortium dedicated to helping these patients receive diagnoses. The network enables primary healthcare clinics to collaborate with more than 400 hospitals including **34 IRUD Clinical Centers**. Patients are referred to these IRUD Clinical Centers when the patient remains undiagnosed for 6 months AND shows a sign of either an affection of more than one organ or a disease with a genetic etiology. Complex cases can thus be reviewed by **multi-disciplinary IRUD Diagnosis Committees** made up of medical specialists and clinical geneticists. This process is supported by **4 IRUD Analysis Centers** which administer genetic tests, including whole-exome or whole-genome sequencing. After the committee's discussion, genetic counselors provide feedback to individual patients.

The **IRUD Data Center** is storing all clinical and genetic data gathered in for each patient in a globally compatible patient-matching system, enabling data to be exchanged, upon consent, with domestic and overseas medical organizations. As a consequence, the chances of successful diagnosis increase with the possibility to compare data of a broader pool of patients.

For the future, IRUD will continue to expand its nationwide network by recruiting more local collaborators and to increase patients' participation by disseminating widely its actions to hospitals, clinicians and municipalities. In addition, IRUD and AMED join forces to strengthen their collaboration with international organizations, for instance, through the IRDiRC consortium.

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



Upcoming Teleconferences and Meetings

- September 6, 2017 - Operating Committee - Teleconference
 - September 18, 2017 - Interdisciplinary Scientific Committee - Teleconference
 - September 25, 2017 - Solving the Unsolved Task Force - Teleconference
 - September 28, 2017 - Funders Constituent Committee - Teleconference
 - October 12, 2017 - Diagnostic Scientific Committee - Teleconference
 - November 10-11, 2017 - Consortium Assembly - Face-to-face meeting in Tokyo, Japan
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Others 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.