

IRDIRC MAY/JUNE 2017 UPDATE

Updated Governance Empowers Umbrella Patient Advocacy Groups to Join IRDiRC

The [IRDIRC Governance](#) has been updated, and among the changes, umbrella patient advocacy groups are now empowered to apply for IRDiRC membership. IRDiRC has long recognised the importance of the involvement of patient representatives in its programme and activities. Previously, umbrella patient advocacy groups were invited members.

IRDIRC welcomes application from umbrella organisations which represent the majority, if not all, rare diseases, across at least one country. Successful applicants will become a member of the Consortium Assembly and also participate in the Patient Advocates Constituent Committee, together with Rare Disease Europe – [EURORDIS](#), the [Genetic Alliance](#), The National Organisation for Rare Disease ([NORD](#)) and more recently the Chinese Organisation for Rare disease ([CORD](#)) and the Advocacy Service for Rare and Intractable Diseases ([ASrid](#)) from Japan.

Information on membership application can be found at [Become a Member](#) page.

The IRDiRC Governance documents, current and former, are available for consultation [here](#).



IRDIRC's New Members

IRDIRC welcomes the Chinese Organization for Rare Disorders (CORD) and the Japanese Advocacy Service for Rare and Intractable Diseases (ASrid) to the Consortium.

The Chinese Organization for Rare Disorders ([CORD](#)), founded by the China-Dolls Center for Rare Disorders in June 2013, is a non-profit organization focused on promoting exchange and cooperation among rare disease patients and organizations such as medical specialists, pharmaceutical companies and governmental agencies.

The Advocacy Service for Rare and Intractable Diseases ([ASrid](#)) is a non-profit organization in Japan for multi-stakeholders in rare and intractable diseases. ASrid is committed to providing valuable services for connecting various stakeholders. ASrid partners with the Japan Patients Association (JPA) and the Pediatric NANBYO Support Network to especially promote research, international relations and drug development. ASrid also collaborates with individual patient groups.



Spotlights on IRDiRC Member Organizations

- The FDA Commissioner Dr Scott Gottlieb unveils plan to [expedite the orphan drug designation process](#).
- Researchers at the NCATS and the University of Nevada have demonstrated that a drug originally targeted unsuccessfully to treat cancer may have new life as a [potential treatment for Duchenne muscular dystrophy](#).
- EURORDIS published a [new position paper on compassionate use](#). Improvement of compassionate use programs is a pre-condition of patients' early access to medicines.
- Fondation maladies rares, together with Eurobiomed, is hosting [Rare 2017](#) (5th edition) on November 20-21, 2017, in Paris, France - the registration of this meeting is now open.

Rare Diseases Research Highlights

- RD-Action publishes [guidelines](#) for implementation of the codification of rare diseases in health information systems.
- EMA publishes [reports on involving patients](#) in discussions on benefits and risks of medicines at the Committee for Medicinal Products for Human Use
- The capture-recapture method to [estimate surveillance data](#) for rare diseases
- An [analysis of orphan designations and authorisations](#) in Europe and United States

Upcoming Teleconferences and Meetings

- July 12, 2017 - Funders Constituent Committee - Teleconference
- August 2, 2017 - Operating Committee - Teleconference
- November 10-11, 2017 - Consortium Assembly - Face-to-face meeting in Tokyo, Japan

IRDIRC-Related Funding Calls

The National Institute of Neurological Disorders and Stroke has launched a funding opportunity announcement entitled: [NeuroNEXT Clinical Trials \(U01\)](#). This announcement encourages applications for exploratory clinical trials of investigational agents (drugs, biologics, surgical therapies or devices) that may contribute to the justification for and provide the data required for designing a future trial, for biomarker validation studies, or for proof of mechanism clinical studies. Application deadline: **August 2, 2017**.

The Canadian Institutes for Health Research has launched its recurrent funding opportunity entitled : [Project Grant Program](#). This announcement is designed to capture ideas with the greatest potential to advance health-related fundamental or applied knowledge, health research, health care, health systems, and/or health outcomes. It supports projects with a specific purpose and a defined endpoint. Letter of intent: **August 15, 2017**. Application deadline: September 15, 2017.

The National Institute of Neurological Disorders and Stroke has launched a funding opportunity announcement entitled: [Clinical Trial Readiness for Rare Neurological and Neuromuscular Diseases \(U01\)](#). This announcement is to support clinical studies that will fill gaps in the design of upcoming clinical trials in rare neurological or neuromuscular diseases by validating clinical outcome measures or biomarkers, or by characterizing cohorts of relevant patients. Application deadline: **August 17, 2017**.

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

Featured Article

International cooperation to enable the diagnosis of all rare genetic diseases

An article by Boycott *et al.* in the American Journal of Human Genetics details the current and future bottlenecks to gene discovery and suggests strategies for enabling progress in this regard.

The application of whole genome sequencing (WGS) and whole exome sequencing (WES) has increased the rate of discovery per year of new genes responsible for rare genetic diseases, enabling the identification of almost 3 times more genes compared than conventional methods. Additionally, the proportion of new disease-gene relations has also steadily increased, although a decline in the discovery rate has been observed in recent years.

There remain a non-trivial number of well-known rare diseases for which the causal genetic mechanism remains elusive despite the use of WES and WGS by multiple groups of investigators. The reasons that such discovery efforts fail most likely include both technical limitations and complex biology. The authors describe ideas to circumvent these issues such as the integration of genomic data into systems biology, the use of model system to facilitate gene discovery, improvement of computational and statistical models for variant identification, and the development of strategies for discovering causal genetic mechanisms.

Achieving the IRDiRC's goal of diagnosing all rare genetic diseases will require significant international cooperation and engagement of all relevant stakeholders at a scale the community has never seen before. Engaging clinical laboratories, researchers, and the patient community to share their data will be critical. The engagement of the research community, international coordination and funding of activities will be necessary.

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

Other news

Congratulations to Anneliene Jonker on the arrival of her bundle of joy! Both mother and child are doing well, and the Secretariat looks forward to a visit with the wee one.

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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.