International Rare Disease Research Consortium (IRDiRC)

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Research Areas

- **Biomarker Research**
- **Basic Research**
- **Product Development**

At a Glance

- Status: **Active Consortium**
- Year Launched: **2009**
- Initiating Organization: **Rare Diseases Platform**
- Initiator Type: **Third-party organization**
- **Rare** disease
- Location: **International**

Abstract

The International Rare Diseases Research Consortium (IRDiRC) teams up researchers and organizations investing in rare diseases research in order to achieve two main objectives by the year 2020, namely to deliver 200 new therapies for rare diseases and the means to diagnose most rare diseases.

Mission

IRDiRC addresses a number of grand challenges through collaborative actions to reach its 2020 goals, such as the following:

Consortium History

Individual researchers and consortia have been studying rare diseases for several decades. However, the rare disease research community was very fragmented, mainly because of the large heterogeneity of rare diseases. Indeed, for many diseases, the numbers of researchers and resources are very limited. Concerted efforts to organize the rare disease research community and funding were emerging in several countries, but international coordination was rather limited.

The idea to establish IRDiRC arose during a meeting between Ruxandra Draghia-Akli (European Commission) and Francis Collins (National Institutes of Health) in 2009. Their main objective was to accelerate medical breakthroughs for people affected by rare diseases by establishing a network of research funders.

In October 2010, the European Commission and the U.S. National Institutes of Health (NIH) announced during the first preparatory workshop in Reykjavik, Iceland, their intention to join forces on rare diseases research. The two institutions planned to coordinate their research funding on rare diseases and to make major investments in this research field in the years to come.

In April 2011, IRDiRC was officially established and launched during the second preparatory workshop in Bethesda, Md. The group of funding agency representatives agreed to have an Interim Executive Committee until the end of 2012. The group chose Ruxandra Draghia-Akli as interim chair.
In October 2011, in Montreal, Canada, the third preparatory workshop gathered 100 participants representing public and private funding organizations, scientists, regulators, industry, and patient groups. It focused on continuous efforts to develop common scientific and policy frameworks to guide the activities of the participating IRDiRC members.

In September 2012, Paul Lasko, scientific director of the Canadian Institutes of Health Research (CIHR) Institute of Genetics, was selected as the next chair of the Executive Committee, starting in 2013.
In April 2013, the first IRDiRC conference took place in Dublin, Ireland. Researchers, clinicians, patient groups, and representatives of public and private organizations met to assess IRDiRC’s three years of work.

Structure & Governance

IRDiRC is run by an Executive Committee composed of one representative per funding body, each group of funders (for the small funders), representatives of patient umbrella organizations, and the chair of each of the three Scientific Committees.

IRDiRC is composed of three Scientific Committees:

Financing

Members are composed of funding bodies/organizations investing a minimum of US$10 million over five years in research projects/programs contributing toward IRDiRC objectives and invited patient advocacy group.

Intellectual Property

IRDiRC research results should be rapidly shared and made highly visible to the scientific, healthcare, patient, and pharmaceutical communities. Their utility must be clearly demonstrated, and potential users must have the opportunity to receive training in the techniques and tools developed. This includes negative results, which can be as important for the rare diseases field as are new scientific breakthroughs (in relation to data-sharing policies and guidelines). A high level of visibility in scientific meetings and through scientific publications is mandated. The scientific impact of IRDiRC research projects should be maximized by pursuing opportunities for publication. Online publication of research results after peer review will be pursued in full respect of international copyright law. Where the most suitable journal for the results offers the possibility of paying a publication fee to ensure free access to all, this option will be strongly encouraged. Some IRDiRC members already mandate open-access publication for all projects they fund, and some cover open-access fees where required. In addition, journals might allow the author to post an electronic copy of the publication on their website. Publications in lay journals may be prepared in order to attract maximum attention to rare diseases.

Intellectual property (IP) is an important factor for the public and the private sector, in particular to cover the significant cost of developing new therapies. Issues related to IP rights need to be assessed and handled in accordance with fundamental ethical rules and principles. Tools to handle IP issues may include exploitation and technological implementation plans, nonexclusive licensing, patenting, knowledge property rights, and preexisting know-how. In many instances, confidentiality agreements may be required between the parties involved.

Policy:

Research projects should publish their results in a timely manner in peer-reviewed scientific journals, preferably with open access.

Guidelines:

Patent Engagement

A number of actionable key areas has been identified by IRDiRC Scientific Committees to advance IRDiRC's two main objectives for the rare disease community: have a therapy for 2OO new rare diseases and have a diagnostic test for most genetic diseases by 2020.

Ad hoc Task Forces are constituted to accelerate policy change in areas contributing to these goals. Task Forces identify actions likely to contribute to speeding up research and development and organize the debate among the relevant stakeholders to reach agreement and ensure appropriation of decisions.

IRDiRC has a task force that focuses on the development and adoption of patient-relevant and -reported outcome measures as instrumental in accelerating research and development in rare diseases. Another task force focuses on small population clinical trials, a collaborative effort on adaptive design, statistical methods, and acceptability of new methods in small population clinical trials.

Data Sharing

To achieve the stated goals of IRDiRC, many types of resources and data will be generated and shared; this will facilitate discovery of genes and treatments while ensuring efficient utilization of resources. Resources may include, but are not limited to, patient and family material (extracted DNA, cell lines, pathological samples), technical protocols, informatics infrastructure, and analysis tools. Datasets may include, but are not limited to, phenotypes, genomic variants, other "omic" data (including transcriptomic, metabolomic, biomarkers), natural histories, and clinical trial data. Ultimately, it is critical to the overall success of IRDiRC that datasets obtained from one project will be directly comparable to datasets obtained from another project (even if generated using a different approach or technology).

Policies:

Impact/Accomplishment

Because of IRDiRC's objective to contribute to deliver 200 new therapies for rare diseases, IRDiRC monitors the cumulative number of medicinal products with an orphan designation and marketing approval for the treatment of rare diseases in the U.S. and/or Europe.

The number is calculated from the information available on European Medicines Agency (EMA) and U.S. Food and Drug Administration (FDA) websites. A same medicinal product approved in both the U.S. and Europe is only counted once. A same medicinal product is counted once for each indication it received a marketing approval. Any medicinal product losing orphan designation or marketing approval for an indication will be removed from the count. The indicator is updated monthly. As of June 2015, the count is at 142 new therapies for rare diseases.

Links/Social Media Feed

Homepage	http://www.irdirc.org/
Twitter	https://twitter.com/irdirc

Points of Contact

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