Progress in Rare Diseases Research 2010–2017: An IRDiRC Perspective

Jagut M[1], Jonker AH[1], Lau LPL[2], Cutillo CM[2], Rath A[1], Dawkins HJS[3], Austin CP[2] on behalf of IRDiRC

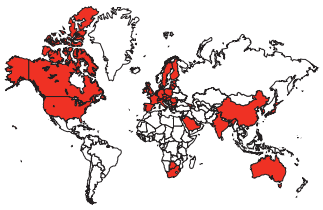
1) IRDiRC Scientific Secretariat, Inserm-US14, Paris, France, 2) NCATS, NIH, Bethesda, US, 3) Department of Health Western Australia, Perth, Australia

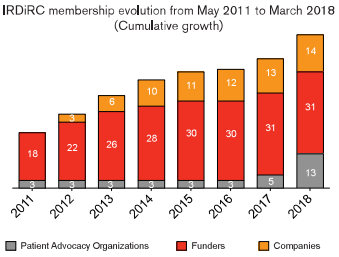
Contact: contact@irdirc.org - Website: www.irdirc.org

Introduction

The International Rare Diseases Research Consortium (IRDiRC) was launched in 2011, uniting public funding organizations, industry, scientific researchers and patient advocacy groups, with a mission to support and promote international collaboration in rare diseases research. The last 7 years have seen considerable progress on rare disease research: the original goal to deliver 200 new therapies was achieved in early 2017 – three years earlier than expected – and the goal for diagnostics is within reach. Nevertheless, the field of rare diseases research still faces many more challenges ahead. This spurs IRDiRC to work towards its 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***".

IRDiRC Membership



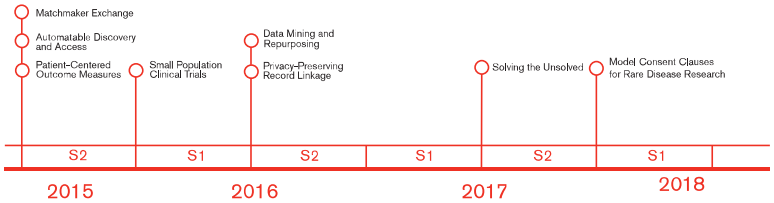


Policies and Guidelines

The IRDiRC Policies and Guidelines were set out to emphasize collaboration in rare diseases research, involvement of patients and their representatives in all relevant aspects of research, as well as the sharing of data and resources.

Task Forces

Task Forces review current barriers to efficient and effective rare disease research, and propose solutions through policy and/or technical recommendations. They are comprised mostly of external experts to ensure that different perspectives are cultivated to drive innovation and new perspectives.



Tools

Automatable Discovery and Access
Initiative set up to develop standardized computer-readable data used types in consent forms by aligning a user's permission against the common set of permitted data use types.
Website:http://www.irdirc.org/activities/task-forces/automatable-discovery-and-access/

Matchmaker Exchange
Initiative created to find genetic causes for patients with rare disease by connecting databases , which contain exome and genome sequencing data from patients.
Website: www.matchmakerexchange.org

Publications

Data Mining and Repurposing
This effort gathers expertise and identifies opportunities for collaborations to efficiently exploit data mining tools to identify new therapeutic targets and repurpose drugs. ***Recommendations on IRDiRC website and article in preparation***

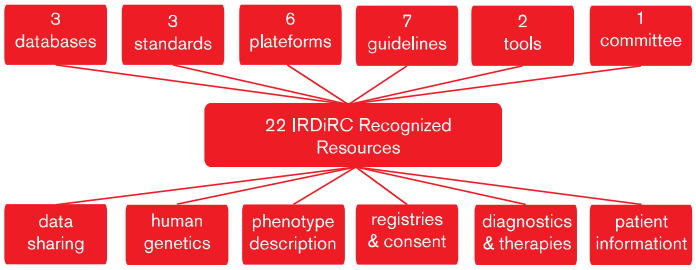
Patient-Centered Outcome Measures (PCOMs)
This work emphasizes on PCOMs being the most meaningful and interpretable measure of the patient benefit, especially in the case of rare diseases. ***Morel T. and Cano S., Orphanet Journal of Rare Diseases, Nov 2017, doi: 10.1186/s13023-017-0718-x***

Privacy-Preserving Record Linkage
This effort develops a guiding policy for the generation of participant-specific idnetifiers that enable data from the same individual to be connected across multiple projects without revealing its identity. ***Recommendations on IRDiRC website and article accepted in IEEE/ACM Transactions on Computational Biology and Bioinformatics Journal***

Small Population Clinical Trials
This effort explores adaptive design, statistical methods and acceptibility of new methods in small population clinical trials. ***Recommendations on IRDiRC website and article submitted***

IRDiRC Recognized Resources

IRDiRC Recognized Resources highlight publicly-available resources that researchers in the rare diseases community have found useful and, if were to be used more broadly, may accelerate the pace of translating discoveries into clinical applications.



Advisory Committee

TREAT-NMD Advisory Committee for Therapeutics (TACT)

Databases

LOVD, 5 YEARS OMIM, orphanet Inserm

Guidelines

- FAIR Guiding Principles document for Scientific Data Management and Stewardship
- Framework for Responsible Sharing of Genomic and Health-Related data
- Gene/Disease Specific Variant Database Quality Parameter Guidelines
- Guidelines for Diagnostic by Next-Generation Sequencing
- Guidelines for the Informed Consent Process in International Rare Disease Research
- International Charter of Principles for Sharing Bio-Specimens and Data
- Standard Operating Procedures for Preclinical Studies

Platforms

CTSR Care and Trial Site Registry; DECIPHER GRCh37 DECIPHER Project; ordo Orphanet Rare Disease Ontology; PHENOME CENTRAL PhenomeCentral; RD Connect Genome Phenome Analysis Platform; TREAT-NMD Neuromuscular Network Patient Registries

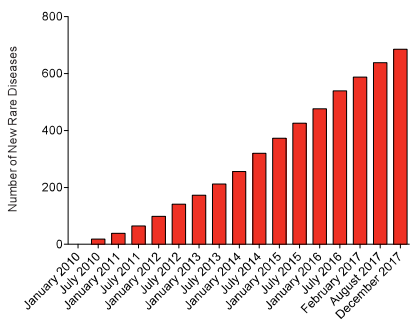
Standards

hpo Human Phenotype Ontology; International Consortium of Human Phenotype Terminologies (ICHPT); HGVS Nomenclature

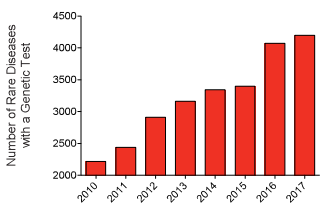
Tools

Mutalyzer; NCATS Toolkit Patient-Focused Therapy Development

Number of New Rare Diseases



Number of Rare Diseases with a Genetic Test



Based on Orphanet data from the following countries: Australia, Austria, Belgium, Bulgaria, Canada, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lebanon, Lithuania, Macedonia, Morocco, Netherlands, Norway, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Spain, Switzerland, Sweden, Tunisia, Turkey, Ukraine, United Kingdom

Number of Approved Orphan Drugs

Based on information extracted from the European Medicines Agency (EMA) and the Food and Drugs Administration (FDA) websites.

Interested in contributing to IRDiRC activities? Please consult the 'Get Involved' section of our website!

The IRDiRC Scientific Secretariat is funded by the European Commission's Seventh Framework Programme (FP7/2007-2013) under grant agreement n°305207 'Support for international rare disease research to serve the IRDiRC objectives (SUPPORT-IRDiRC).'

www.irdirc.org