Accelerate R&D in genetic diseases and therapy: Initiatives from the International Rare Diseases Research Consortium (IRDiRC)

Aymé S¹, Lau LPL¹, Jonker AH¹, Peixoto S¹, Lasko P²

1) Inserm-US14, Paris, France
2) CIHR, McGill University, Montreal, Canada
Contact: contact@irdirc.org - www.irdirc.org

IRDiRC teams up researchers and organizations investing in rare diseases research to achieve two main objectives:

► 200 new therapies for rare diseases by 2020
► Means to diagnose most rare diseases by 2020

Members are funding organizations spending more than 10 million USD over 5 years in research projects contributing to IRDiRC objectives. It includes governmental department, academia, industry and patient organizations.

IRDiRC’s Task Forces have prioritized research areas specific to rare diseases to advance policy change.

**International Consortium of Human Phenotype Terminologies (ICHPT)**

► Provide standards for interoperability between databases
► Linking phenotype and genotype databases in rare diseases

Available at www.irdirc.org/ICHPT

**Task Force: Machine Readable Consent**

► Associate clinical data with the scope of consent given by a patient
► Develop standardized and computer-readable data use types in consent forms
► Aligning a user’s permission against permitted data use types

**Task Force: Matchmaker Exchange**

► Tool to match unsolved genome/exome sequence cases

*Matchmaker Community Engagement Event at ASHG, 7 Oct, 7-9 PM, Room 339-342.*

**Task Force: Patient-Centered Outcome Measures**

► Development and adoption of patient-centered outcome measures
► Improve feasibility and quality of trials
► Provide data of relevance to patients and decision makers

**Task Force: Small Population Clinical Trials (SPCTs)**

► Adaptive design, statistical methods and acceptability of new methods in SPCTs
► Challenges in identifying and recruiting patients
► More sensitive outcome to quantify disease evolution

**Task Force: Data Mining/Repurposing**

► Opportunities for collaborators to exploit data mining tools
► Identify new therapeutic targets and repurpose drugs
► Increase speed of new drugs available for rare disease patients

---

**Timeline**

- Matchmaker Exchange
- Machine Readable Consent
- Patient-Centered Outcome Measures
- Small Population Clinical Trials
- Data Mining/Repurposing

- Q4 2015
- Q1 2016

---

*Funded by the European Union*  
*Project funded by the European Community’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)”*