International Rare Diseases Research Consortium (IRDiRC)
Objectives of IRDiRC, by 2020

- 200 new therapies for rare diseases
- Means to diagnose most rare diseases
IRDiRC – Basic Principles

- International level co-operation to stimulate, better coordinate & maximize output of rare disease research efforts around the world
- Teams up public and private organizations investing in RD research
- Research funders with relevant programs >$10 million over a 5-year
- Each organization funds research its own way
- Funded projects adhere to a common framework
IRDiRC’s Members

IRDiRC members

- 25 funding agencies (including E-Rare Consortium)
- 10 companies
- 5 institutes
- 2 ministries
- 1 consortium
- 3 invited patient advocacy groups
Number of New Orphan Drugs

- Monthly update of number of new indications for orphan diseases

- Source: EMA and FDA

Updated on www.irdirc.org
Number of Rare Diseases in Europe

Cumulative number of new rare diseases by month since 2010

Source: Ophanet Data
Number of Diagnostic Tests

Source: Orphanet Data
IRDiRC Task Forces

- To work on selected, actionable topics/research areas and push forward policy change

- *Ad hoc* committees: nominated experts from different backgrounds, affiliations, geographical areas

- Collaborate through teleconferences and workshops
  - Production and dissemination of reports
  - Implementation of outcomes
  - Publication in peer-reviewed journals
  - Presentation at conferences
Actionable projects to ensure IRDiRC meets its objectives for the rare diseases community are carried out by Task Forces

Updated on www.irdirc.org
International Consortium of Human Phenotype Terminologies

- Agreement to define a core set of terms common to all terminologies
- Provide standards for interoperability between databases
- Core set identified by cross-referencing: HPO, PhenoDB, Orphanet, UML, LDBB
- Selection of 2370 terms

Available at www.irdirc.org/ICHPT
Matchmaker Exchange

- Provides data sharing tools between clinical geneticists to match unsolved genome/exome sequence cases

- Ensures optimal collaboration between all projects contributing to the interpretation of variants and of matching phenotypes and variants

- Joint IRDiRC-GA4GH collaboration
Automatable Discovery and Access

- Associate clinical data with the scope of consent given
- Develop standardized and computer-readable data use types in consent forms
- Aligning a user’s permission against permitted data use type
- Coordinate with the GA4GH and other initiatives (e.g. RD-Connect)
Patient-Centered Outcome Measures

- To boost the development and adoption of patient-centered outcome measures
- Explore to whether, how and to what extent these initiatives can be expanded to target rare disease research in order to improve feasibility and quality of trials
- Post-workshop report and recommendations available on IRDiRC website
Small Population Clinical Trials

- Contribute consensus about non-conventional statistical methods used for small population clinical trials
- Contribute to the acceptability of new statistical methods and coordinate with the different agencies; EMA, FDA, industry, IDEAL, INSPIRE, ASTERIX
- Post-workshop report and recommendations available on IRDiRC website
Data Mining and Repurposing

- Leverage on developments in Computational Linguistics and Graph Theory to build a representation of knowledge which is automatically analyzed to discover hidden relations between any drug and diseases.

- 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
Privacy-Preserving Record Linkage

- Development of participant unique identifiers for research data sharing across multiple projects and institutions

- Product: Guidelines on the technical and ethical-legal requirements of patient identifiers in Rare Disease Research; recommendations for the most practical, streamlined and minimalistic approach that maximises uptake whilst complying with relevant legal regulations.

- Joint IRDiRC-GA4GH collaboration
Solving the Unsolved

- Identification of the genetic basis of rare conditions presently intractable to existing approaches

- Based on exome sequencing requires development of innovative approaches for discovery

- The objective is to bring together the community addressing this challenge to share best practices regarding approaches
Patient Engagement in Research

- To promote patient engagement in all RD research activities and health product development

- Based on guiding principles for the engagement of patient groups or patient experts in research activities

- Joint ISC-TSC Task Force
Clinical Data Sharing

- Facilitate access to clinical genome-wide sequencing for secondary use of data focused on discovery of disease mechanism

- Focuses on:
  - Technical aspects of data sharing
  - Ownership and cost for access
  - Patient-driven sharing
  - Multi-stakeholder engagement
“IRDiRC Recognized Resources”

Label highlighting resources which contribute directly to IRDiRC objectives and to accelerate research-clinic translation

Application information via website; to date awarded to:

- Orphanet
- International Charter of Principles for sharing Bio-Specimens and Data
- PhenomeCentral
- ORDO
- HPO
- ICHPT
- GA4GH Framework for Responsible Sharing
- DECIPHER
- TREAT-NMD Patient registries
- TREAT-NMD Standard operating procedures
- TREAT-NMD Advisory Committee for Therapeutics
- Care and Trial Site Registry
- OMIM
Annual State-of-Play Reports

State of Play of Research in the field of Rare Diseases: Trends and Gaps in 2014

Annual report from the Scientific Secretariat to the IRDiRC Executive Committee

October 2014

State of Play of Research in the Field of Rare Diseases: 2014-2015

Freely available on www.irdirc.org
IRDiRC Policies and Guidelines

Freely available on www.irdirc.org