International Rare Diseases Research Consortium (IRDiRC)
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 Goals, by 2027

- All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

- 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

- Methodologies will be developed to assess the impact of diagnoses and therapies on rare diseases patients.
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

- 58 IRDiRC members
  - 31 funders
  - 14 companies
  - 13 patient advocates organizations
Number of New Orphan Drugs

Source: EMA and FDA

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

Source: Orphanet Data
Number of Diagnostic Tests

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

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
Task Forces in Action

Actionable projects to ensure IRDiRC meets its objectives for the rare diseases community are carried out by Task Forces.
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

IRDiRC

INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM
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.

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<td>TREAT-NMD Advisory Committee for Therapeutics (TACT)</td>
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<td>50 Years MIM Human Disease Knowledge for the World</td>
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<td>- Framework for Responsible Sharing of Genomic and Health-Related data</td>
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<td>- Gene/Disease Specific Variant Database Quality Parameter Guidelines</td>
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<td>- Guidelines for Diagnostic by Next-Generation Sequencing</td>
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<td>- Guidelines for the Informed Consent Process in International Rare Disease Research</td>
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<td>- International Charter of Principles for Sharing Bio-Specimens and Data</td>
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<td>- Standard Operating Procedures for Preclinical Studies</td>
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Annual State-of-Play Reports

Freely available on www.irdirc.org