IRDiRCs Roadmap
Criteria for Selection of Objectives

- Projects contributing to IRDiRC to objectives
- High leverage projects with strong translational potential and international scope
- Targeted action to produce results by 2020
- Projects not covered by national initiatives
- Actionable projects with secured human and financial resources
- Coordination with other organizations when appropriate
Organization put in place to drive action: Task Forces

- Temporary committees that work on selected actionables of IRDiRC’s road map in specific research areas to push forward policy change

- Nominated experts from different backgrounds, on ad hoc basis

- Uses a variety of tools, such as workshops, production and dissemination of reports, and implementation of outcomes
Currently Selected Topics for Action

- Minimum set of terms in terminologies
- Matchmaker Exchange
- Automatable Access and Discovery
- Patient Centred Outcome Measures
- Small Population Clinical Trials
- Data Mining and Repurposing
- Privacy-Preserving Record Linkage
- Solving the Unsolved
- Patient Engagement
- Clinical Data Sharing
- “IRDiRC Recognized Resources”
- State-of-Play of research activities
Timeline for Task Forces

- Matchmaker Exchange
- Automatable Discovery and Access
- Patient-Centered Outcome Measures
- Small Population Clinical Trials
- Data Mining and Repurposing
- Privacy-Preserving Record Linkage
- Solving the Unresolved
- Clinical Data Sharing
- Patient Engagement in Research

2015 | 2016 | 2017
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
International Consortium of Human Phenotype Terminologies: key members

- Peter Robinson, Institute for Medical Genetics Universitätsklinikum Charité / HPO, Germany
- Ada Hamosh, John Hopkins University School of Medicine/OMIM, USA
- Ana Rath, Orphanet, Inserm US14, France
- Ségolène Aymé, IRDiRC, Inserm US14, France

- Past workshop Q2, 2013
- Recommandations released in September 2015 on www.ichpt.org
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-Global Alliance Project
Matchmaker Exchange: Key Members

- Kym Boycott, Children’s Hospital Eastern Ontario, Canada
- Tony Brookes, University of Leicester, UK
- Han Brunner, Radboud University Medical Centre, the Netherlands
- Ada Hamosh, John Hopkins University School of Medicine, USA
- Bartha Knoppers, McGill University, Canada
- Anthony Philippakis, Broad Institute of MIT & Harvard, USA
- Heidi Rehm, Partners & Harvard Medical School, USA

- Workshop October 6, 2015, at ASHG2015
Automatable Access and Discovery

- 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 type
- Coordinate with the Global Alliance subcommittee and other initiatives (e.g. RD-Connect)

- Workshop November 9-10, 2015, in Paris
- ADA-Matrix in beta-testing phase
Patient Centered Outcome Measures

- Boost the development and adoption of patient-centered outcome measures with PCORI, ISPOR, COMET, MAPI, ICHOM, FDA, EMA, IMI

- Explore to whether, how and to what extent these initiatives can be expanded to target RD research in order to improve feasibility and quality of trials
Patient Centered Outcome Measures: Key Members

- Thomas Kelley, UK Foundation Program Office, member of ICHOM, UK
- Marshall Summar, Children’s National Medical Center, member of PCORI, USA
- Sharon Terry, Genetic Alliance, USA
- Margaret Vernon, ISPOR, UK
- Paula Williamson, Liverpool University/ member of COMET, UK

- Workshop November 30, 2015, in Paris
- Post-workshop report 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
Small Population Clinical Trials: Key Members

- Ralf-Dieter Hilgers, RWTH Aachen, group leader IDEAL, Germany
- Ilan Irony, FDA/CBER/OCTGT, USA
- Nigel Stallard, University of Warwick, group leader INSPIRE, UK
- Kit Roes, UMC Utrecht, group leader ASTERIX, The Netherlands
- Kristina Larsson, EMA, UK
- Simon Day, CTCT, UK

- Workshop March 3, 2016, in London
- Post-workshop report 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 analysed 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
- Gather the expertise and identify opportunities for collaborations to speed up the exploitation of these new tools
Data Mining and Repurposing: Key Members

- Benoît Deprez, Institut Pasteur Lille, APTEEUS, France
- Peter-Bram ’t Hoen, Leiden University Medical Center, The Netherlands
- Caroline Kant, EspeRare Foundation, Switzerland
- Frédéric Marin, GMP-Orphan, France
- Madhu Natarajan, Shire, USA
- Jordi Quintana, Plataforma Drug Discovery, Spain
- Noel Southall, NIH/NCATS, USA

- Workshop November 24, 2016, in Barcelona
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
Privacy-Preserving Record Linkage: Key members

- The Participant Unique Identifiers Task Force is co-chaired by:
  - Dr. Petra Kaufmann, ORDR, NCATS, NIH, US
  - Prof. Bartha Maria Knoppers, McGill University, Canada
  - Dr. Dixie Baker, Senior Partner at Martin, Blanck and Associates

- The coordinators of this Task Force are:
  - Academic coordinator: Mark Phillips, GA4GH, Canada
  - Logistical coordinator: Lilian Lau, IRDiRC, France

- Workshop December 8-9, 2016, in Paris
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

- Aimed 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