

# The International Rare Diseases Research Consortium (IRDIRC)

Christopher P. Austin, M.D.

ICORD RareX 2016

Cape Town, SA

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# International Rare Diseases Research Consortium (IRDiRC)

- ▶ Global coordination and cooperation to stimulate and maximize output of rare disease research efforts
  - ↳ Members from Europe, North America, Asia, Australia, Middle East
  - ↳ *(Need members from Africa and Latin America!)*
  - ↳ Each funder supports its own research
- ▶ Initial focus on developing common scientific and policy frameworks
- ▶ 2011-2016 objectives:
  - ↳ 200 new therapies for rare diseases by 2020
  - ↳ Means to diagnose most rare diseases by 2020
  - ↳ *Will be largely achieved by 2017 → new objectives being formulated*

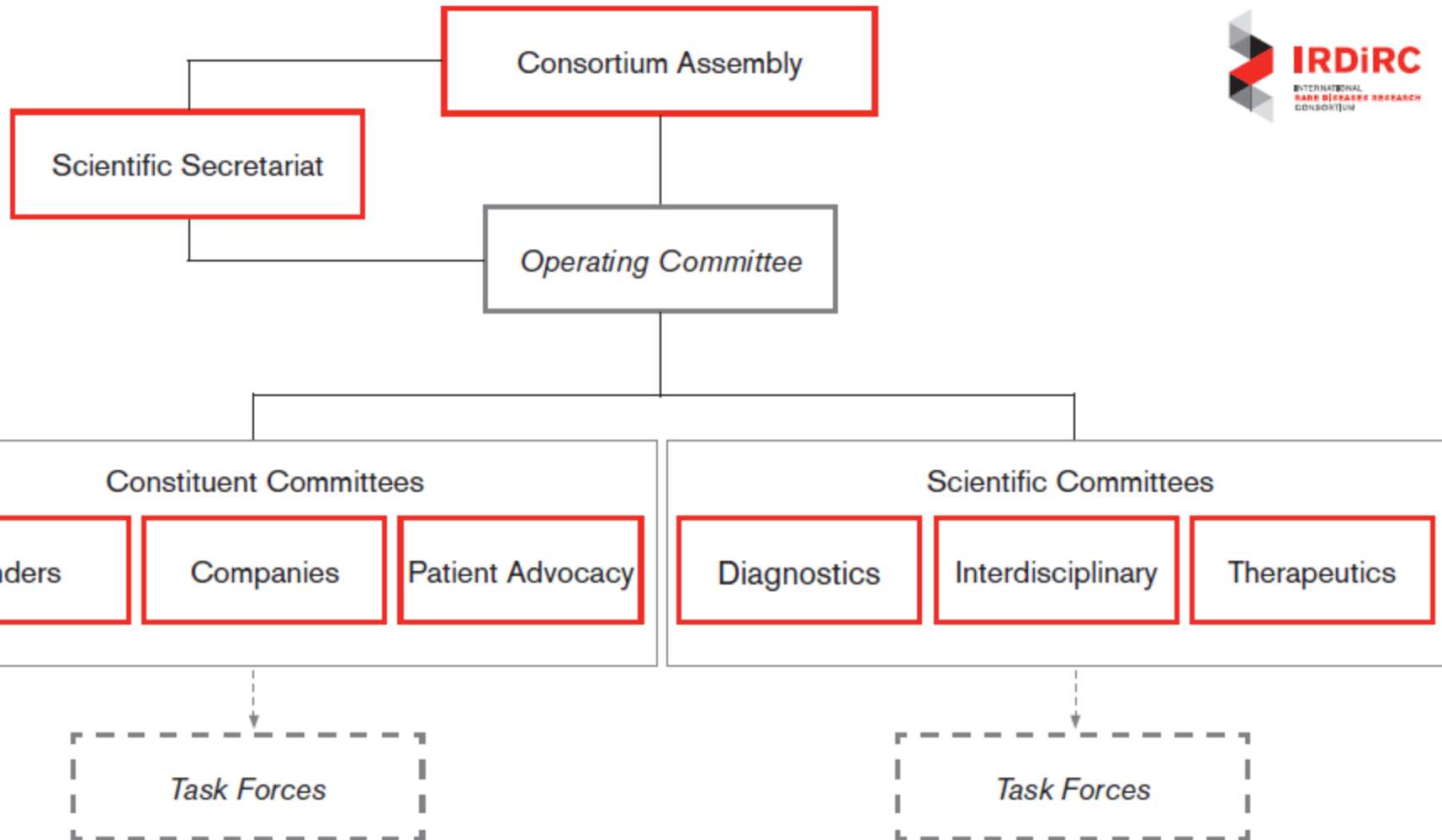
# IRDiRC History

- ▶ **2009** – Idea (Draghia-Akli, Collins)
- ▶ **October 2010** – IRDiRC announced
- ▶ **April 2011** – IRDiRC established
- ▶ **April 2013** – First IRDiRC Conference, Dublin
- ▶ **November 2014** – Second IRDiRC Conference, Shenzhen
- ▶ **February 2017** – *Third IRDiRC Conference, Paris*
- ▶ **Chairs**
  - ↳ **April 2011** – Dr. Ruxandra Draghia-Akli (European Commission)
  - ↳ **January 2013** – Dr. Paul Lasko (Canadian Inst. of Health Research)
  - ↳ **February 2016** – Dr. Chris Austin (NIH/NCATS)



**Reykjavik Meeting, Oct 2010 – IRDiRC announced**

# IRDiRC Structure



# IRDiRC Constituency Committees

▶ (1) Funders; (2) Industry; (3) Patient Advocacy

▶ Goals:

↳ To identify,

- Overlap and gaps of priorities within constituency space
- Common roadblocks across constituency space worldwide
- Other people within the constituency space who would benefit the committee

↳ Use this information to

- Determine next goals for IRDiRC
- Identify how the constituency will contribute to the new set of goals

# IRDiRC Scientific Committees

- ▶ (1) Diagnostic; (2) Interdisciplinary; (3) Therapeutic
- ▶ Goals:
  - ↪ Advising the Consortium Assembly on research priorities, progress, and emerging issues
  - ↪ Encouraging exchange of protocols and best practices
  - ↪ Agreeing on standard operating procedures, quality standards, roadmap to reach IRDiRC goals in their scientific area
  - ↪ Identifying projects and contribute to their implementation
- ▶ Balanced representation of constituencies

# Diagnostics Scientific Committee (DSC)



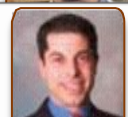
**Kym Boycott (chair)**

•Children's Hospital Eastern Ontario (Canada)



**Fowzan Sami Alkuraya**

•King Faisal Specialist Hospital (Saudi Arabia)



**Michael Bamshad**

•Seattle Children's Hospital (USA)



**Gareth Baynam (co-chair)**

•Western Australia Department of Health (Australia)



**Anthony Brookes**

•Leicester University (UK)



**Han Brunner**

•Nijmegen University Hospital (The Netherlands)



**Johan Den Dunnen**

•Center for Human and Clinical Genetics (The Netherlands)



**Xavier Estivill**

•Genomic Regulation Centre (Spain)



**Milan Macek**

•Charles University Prague (Czech Republic)



**Gert Matthijs**

•University Hospital Leuven (Belgium)



**Woong-Yang Park**

•Samsung Genome Institute (Korea)



**Pak-Chung Sham**

•Chinese Rare Disease Research Consortium (China)



**Jun Wang**

•BGI (China)



**Hendrik Stunnenberg**

•Radboud University (The Netherlands)



**Feng Zhang**

•WuXi AppTec (China)

# Interdisciplinary Scientific Committee (ISC)



**Hanns Lochmüller (chair)**

•University Newcastle upon Tyne (UK)



**Bartha Maria Knoppers**

•McGill University (Canada)



**Angel Carracedo**

•University of Santiago de Compostela (Spain)



**Jeffrey Krischer**

•University of South Florida (USA)



**Gema Chicano**

•EURORDIS, AADE (Spain)



**Samantha Parker**

•Lysogene (France)



**Jack Goldblatt**

•Genetic Services and the Familial Cancer Program of Western Australia (Australia)



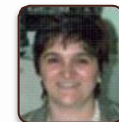
**Rumen Stefanov**

•Medical University of Plovdiv (Bulgaria)



**Steven Groft**

•NCATS/ORDR, NIH (USA)



**Domenica Taruscio**

•Italian National Centre for Rare Diseases (Italy)



**Petra Kaufmann (co-chair)**

•NCATS/ORDR, NIH (USA)

# Therapies Scientific Committee (TSC)



**Yann Le Cam (chair)**

• EURORDIS (France)



**Sandrine Marreaud**

• EORTC (Belgium)



**Diego Ardigo (co-chair)**

• Chiesi Farmaceutici S.p.A. (Italy)



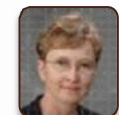
**Akifumi Matsuyama**

• NIBIOHN (Japan)



**Seng H. Cheng**

• Genzyme (USA)



**Asla Pitkänen**

• University of Eastern Finland (Finland)



**Robin Conwit**

• NIH (USA)



**Karin Rademaker**

• University Medical Centre (Netherlands)



**Shuling Guo**

• Ionis Pharmaceuticals (USA)



**Josep Torrent i Farnell**

• Spanish Medicines Agency (Spain)



**Adam Heathfield**

• Pfizer (UK)



**Gert-Jan Van Ommen**

• Leiden University Medical Centre (Netherlands)



**Virginie Hivert**

• EURORDIS (France)



**Anne Zajicek**

• NICHD (USA)

# IRDiRC Task Forces

- ▶ **Diagnostics Scientific Committee (DSC)**
  - ↳ Matchmaker Exchange (joint effort with GA4GH)
  
- ▶ **Interdisciplinary Scientific Committee (ISC)**
  - ↳ Automatable Access and Discovery (joint effort with GA4GH)
  - ↳ Participant Unique Identifiers (joint effort with GA4GH)
  
- ▶ **Therapies Scientific Committee (TSC)**
  - ↳ Patient Centred Outcome Measures
  - ↳ Small Population Clinical Trials
  - ↳ Data Mining/Repurposing

# DSC: Matchmaker Exchange TF

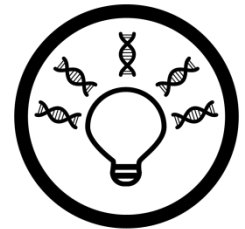
## ► Goals

- ✚ Provides data sharing tools for clinical geneticists to match unsolved genome/exome sequence cases
- ✚ Ensures optimal collaboration between projects contributing to the interpretation of variants and of matching phenotypes and variants

## ► Joint IRDiRC-GA4GH collaboration

## ► *Updates*

- ✚ *Publication in Human Mutation in Oct 2015*
- ✚ *Work ongoing*



Matchmaker  
Exchange

# www.matchmakerexchange.org



# ISC: Automatable Access and Discovery TF

## ► Goals

- ↪ 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

## ► Joint IRDiRC-GA4GH collaboration, led by GA4GH

## ► *Updates*

- ↪ *ADA-Matrix in beta-testing phase, paper in writing*



# ISC: Participant Unique Identifiers TF

## ► Goals

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

## ► Updates

- ✚ *Workshop will be held on 8-9 Dec 2016 in Paris*



# TSC: Patient Centered Outcome Measures TF

## ► Goals

- ✚ 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

## ► *Updates*

- ✚ *Report and recommendations on IRDiRC website*
- ✚ *Publication in process*

# TSC: Small Population Clinical Trials TF

## ► Goals

- ↪ 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 agencies and consortia; EMA, FDA, industry, IDEAL, INSPIRE, ASTERIX

## ► *Update:*

- ↪ *Report and recommendations available on IRDiRC website*
- ↪ *Publication in process*

# TSC: Data Mining/Repurposing TF

## ► Goals

- ↳ 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
- ↳ Gather the expertise and identify opportunities for collaborations to speed up the exploitation of these new tools

## ► *Update:*

- ↳ *Workshop will be held 16 Nov 2016 in Barcelona*

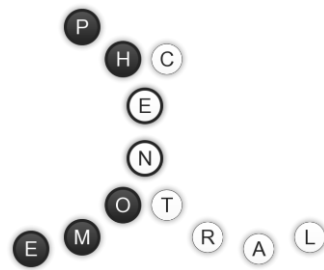
# IRDiRC Recognized Resources

- ▶ Label highlighting resources which contribute to IRDiRC objectives and accelerate research-clinic translation
- ↪ Generally useful resources for RD research that have received recognition by researchers in the RD community
- ↪ Peer-reviewed process
  - Including internal Sci Comm members and independent researchers
  - Criteria based on IRDiRC Policies and Guidelines

# IRDiRC Recognized Resources

- ▶ International Charter of Principles for sharing Bio-Specimens and Data
- ▶ Orphanet
- ▶ PhenomeCentral
- ▶ Orphanet Rare Disease Ontology (ORDO)
- ▶ DECIPHER
- ▶ Guidelines for the informed consent process in international collaborative RD research
- ▶ TREAT-NMD Advisory Committee for Therapeutics

- ▶ GA4GH Framework for Responsible Sharing
- ▶ HPO
- ▶ ICHPT
- ▶ TREAT-NMD Patient Registries
- ▶ TREAT-NMD Standard Operating Procedures
- ▶ Framework for Responsible Sharing of Genomic and Health-Related Data



orphanet



**IRDiRC**  
INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM

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**DECIPHER**  
GRCh37





# 3<sup>rd</sup> IRDiRC Conference

*February 8-9, 2017, Paris, France*



- ▶ Five year anniversary of IRDiRC celebration
- ▶ Celebrate achievements in the field, identify future milestones and goals, and work toward bringing diagnoses and therapies to all RD patients
- ▶ All RD stakeholders invited – investigators, policy makers, opinion leaders, critical thinkers, young investigators, patient advocates
- ▶ Registration open: [www.irdirc.org/conference-2017](http://www.irdirc.org/conference-2017)