



NGO COMMITTEE FOR  
**RARE DISEASES**

# The International Rare Diseases Research Consortium (IRDIRC)

Christopher P. Austin, M.D.

United Nations NGO Committee for Rare Diseases

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A Committee of **CoNGO**



CoNGO  
The Conference of NGOs  
in Consultative Relationship  
with the United Nations



# 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
  - \* Members from Africa and South America in progress
  - \* 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 achieved by 2017 → new objectives being formulated*

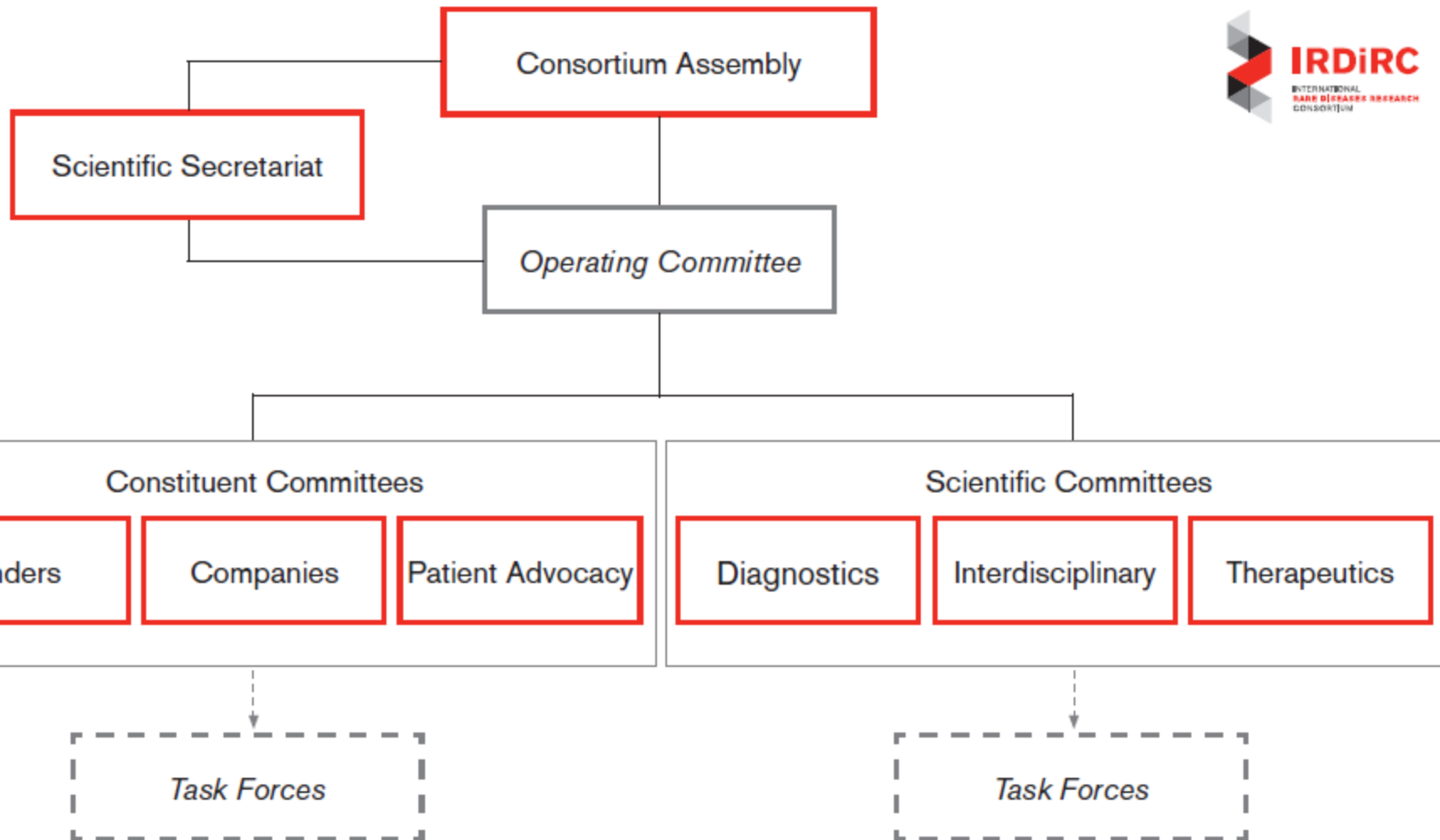
# IRDiRC History

- \* **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



# 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



# 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



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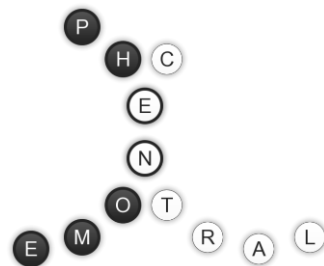


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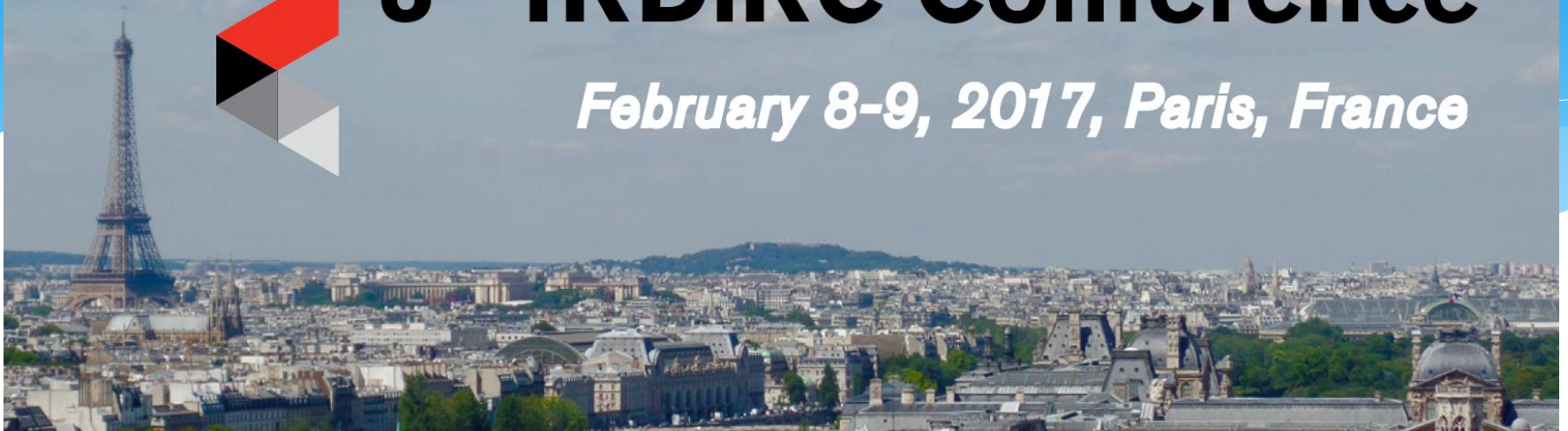
**DECIPHER**  
GRCh37





# 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)