



# The International Rare Diseases Research Consortium (IRDiRC) *Progress – Goals – Activities*

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**IRDiRC**

INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM

# International Rare Diseases Research Consortium (IRDiRC)

- ▶ Established 2011 to maximize global coordination and cooperation in rare disease research
  - ↳ Members from Europe, North America, Asia, Australia, Middle East
  - ↳ Each funder supports its own research
- ▶ Initial focus on developing common scientific and policy frameworks
- ▶ Initial objectives 2011-2020:
  - ↳ 200 new therapies for rare diseases by 2020
  - ↳ Means to diagnose most rare diseases by 2020
  - ↳ ***Achieved in 2017 → new objectives formulated***



## Medical research: Next decade's goals for rare diseases

Christopher P. Austin & Hugh J. S. Dawkins

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The International Rare Diseases Research Consortium (IRDIRC) achieved its ambitious goals for 2020 — three years ahead of schedule (2011). The consortium has now forged a further set of goals for 2030 for people who have debilitating and lethal rare diseases.

Rare diseases were once considered medical curiosities of intractable and negligible public-health impact. The molecular basis of almost 6,000 such conditions is now known, and many have approved treatments. However, diagnosis of most of these conditions remains arduous and costly.

The new IRDiRC goals aim to achieve diagnosis within one year of a disorder being known, this will be accomplished through international coordination of diagnostic and research pipelines for diseases with no approved treatments. Other goals are to develop 1,000 new therapies, particularly for diseases with no approved treatment, and to create methods for assessing the impact of diagnostic treatment, and to create methods for assessing the impact of diagnostic treatment on patients' well-being. (For details, see H. J. S. Dawkins *et al.* *Clin. Transl. Sci.*, in the press.)

The IRDiRC has nearly 50 organizations in 18 nations, with a combined budget of more than US\$2 billion (see [go.nature.com/2htbauh](http://go.nature.com/2htbauh)). New members are being added to this coordinated effort.

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# IRDIRC Goals 2017–2027

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## REVIEW

# Progress in Rare Diseases Research 2010–2016: An IRDiRC Perspective

Hugh J.S. Dawkins<sup>1</sup>, Ruxandra Draghia-Akli<sup>2,3</sup>, Paul Lasko<sup>4</sup>, Lilian P.L. Lau<sup>5</sup>, Anneliene H. Jonker<sup>5</sup>, Christine M. Cutillo<sup>6</sup>, Ana Rath<sup>5,7</sup>, Kym M. Boycott<sup>8</sup>, Gareth Baynam<sup>9,10</sup>, Hanns Lochmüller<sup>11</sup>, Petra Kaufmann<sup>6</sup>, Yann Le Cam<sup>12</sup>, Virginie Hivert<sup>12</sup> and Christopher P. Austin<sup>6</sup> on behalf of the International Rare Diseases Research Consortium (IRDIRC)

coordinated diagnostic and research pipeline

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## REVIEW

# Future of Rare Diseases Research 2017–2027: An IRDiRC Perspective

Christopher P. Austin<sup>1,\*</sup>, Christine M. Cutillo<sup>1</sup>, Lilian P.L. Lau<sup>2</sup>, Anneliene H. Jonker<sup>2</sup>, Ana Rath<sup>2,3</sup>, Daria Julkowska<sup>4</sup>, David Thomson<sup>5</sup>, Sharon F. Terry<sup>6</sup>, Béatrice de Montleau<sup>7</sup>, Diego Ardigo<sup>8</sup>, Virginie Hivert<sup>7</sup>, Kym M. Boycott<sup>9</sup>, Gareth Baynam<sup>10,11</sup>, Petra Kaufmann<sup>1</sup>, Domenica Taruscio<sup>12</sup>, Hanns Lochmüller<sup>13</sup>, Makoto Suematsu<sup>14</sup>, Carlo Incerti<sup>15</sup>, Ruxandra Draghia-Akli<sup>16,17</sup>, Irene Norstedt<sup>16</sup>, Lu Wang<sup>18</sup> and Hugh J.S. Dawkins<sup>19</sup> on behalf of the International Rare Diseases Research Consortium (IRDIRC)

# IRDiRC Consortium Assembly

## *Representation*

### ○ Funders

- ▶ Academy of Finland
- ▶ Agence Nationale de la Recherche, ANR
- ▶ Canadian Institutes for Health Research
- ▶ Children's New Hospitals Management Group
- ▶ Chinese RD Research Consortium
- ▶ E-Rare Consortium
- ▶ European Commission DG RTD
- ▶ European Organisation for Treatment & Research on Cancer, EORTC
- ▶ Federal Ministry of Education and Research
- ▶ Food and Drug Administration, FDA
- ▶ Fondation Maladies Rares
- ▶ French Muscular Dystrophy Association, AFM
- ▶ Genome Canada
- ▶ Istituto Superiore de Sanità
- ▶ Japan Agency for Medical Research and Development, AMED
- ▶ Korea National Institute of Health
- ▶ Loulou Foundation

- ▶ NIH, National Cancer Institute, NCI
- ▶ NIH, National Center for Advancing Translational Sciences, NCATS
- ▶ NIH, National Eye Institute, NEI, NIH
- ▶ NIH, National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS, NIH
- ▶ NIH, National Institute of Child Health and Human Development, NICHD, NIH
- ▶ NIH, National Human Genome Research Institute, NHGRI
- ▶ National Institute of Health Carlos III, ISCIII
- ▶ National Institute for Health Research
- ▶ National Institute of Neurological Disorders and Stroke, NINDS, NIH
- ▶ National Institutes of Biomedical Innovation, Health and Nutrition, NIBIOHN
- ▶ Netherlands Organisation for Health Research and Development
- ▶ Sanford Research
- ▶ Saudi Human Genome Project
- ▶ Telethon Foundation
- ▶ Western Australia Department of Health

### ○ Companies

- ▶ BGI
- ▶ Chiesi Pharmaceuticals
- ▶ Cydan II

- ▶ Genzyme
- ▶ Ionis Pharmaceuticals
- ▶ Lysogene
- ▶ NKT Therapeutics
- ▶ Pfizer
- ▶ PTC Therapeutics
- ▶ Recursion Pharmaceuticals
- ▶ Roche
- ▶ Shire
- ▶ WuXi AppTec

### ○ Patient Advocacy

- ▶ Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan
- ▶ Botswana Organization for Rare Diseases
- ▶ Canadian Organization for Rare Disorders
- ▶ Chinese Organization for Rare Disorders
- ▶ EURORDIS
- ▶ Genetic Alliance
- ▶ Global Genes
- ▶ Indian Organization for Rare Diseases
- ▶ National Organization for Rare Diseases
- ▶ Organization for Rare Diseases India
- ▶ Rare Diseases International
- ▶ Rare Diseases South Africa
- ▶ Rare Voices Australia

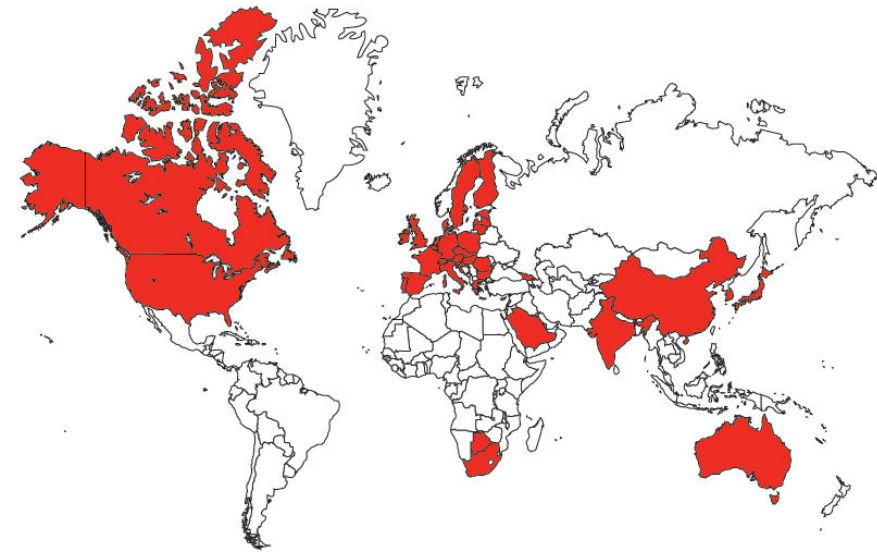


**IRDiRC**

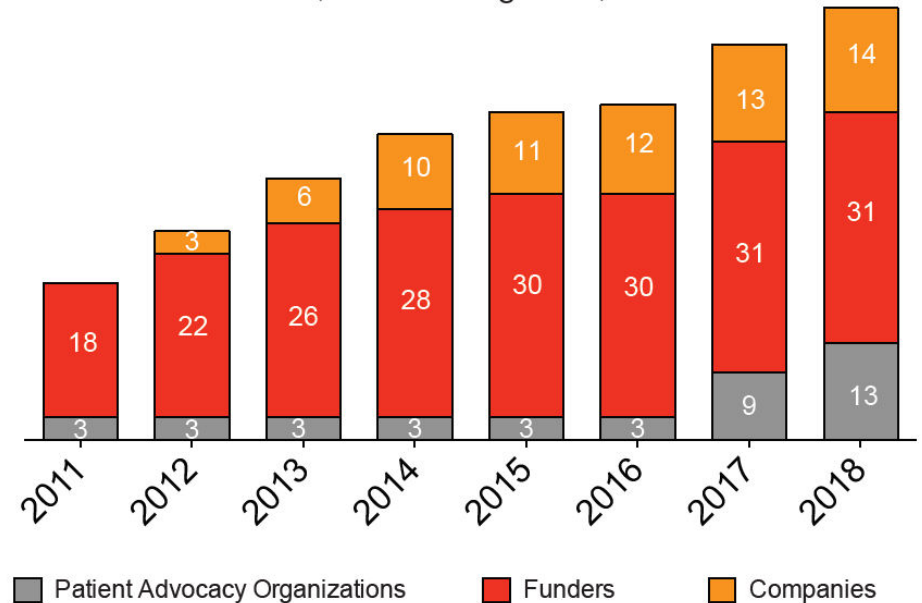
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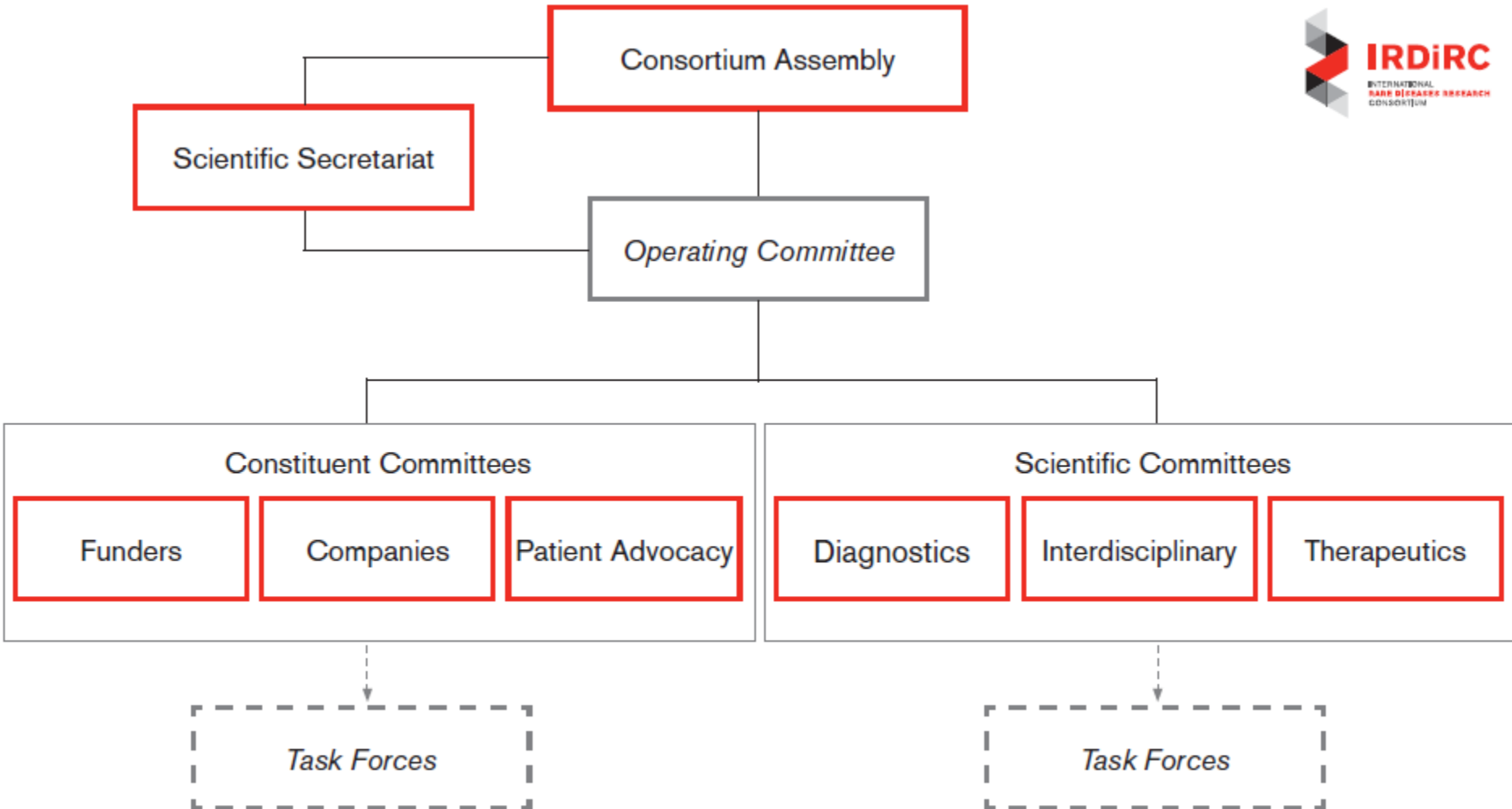
## *Representation*



IRDiRC membership evolution from May 2011 to March 2018  
(Cumulative growth)



# IRDiRC Structure



# IRDiRC Policies and Guidelines

- ▶ Provide guidance and recommendations on key topics in rare diseases research, including:

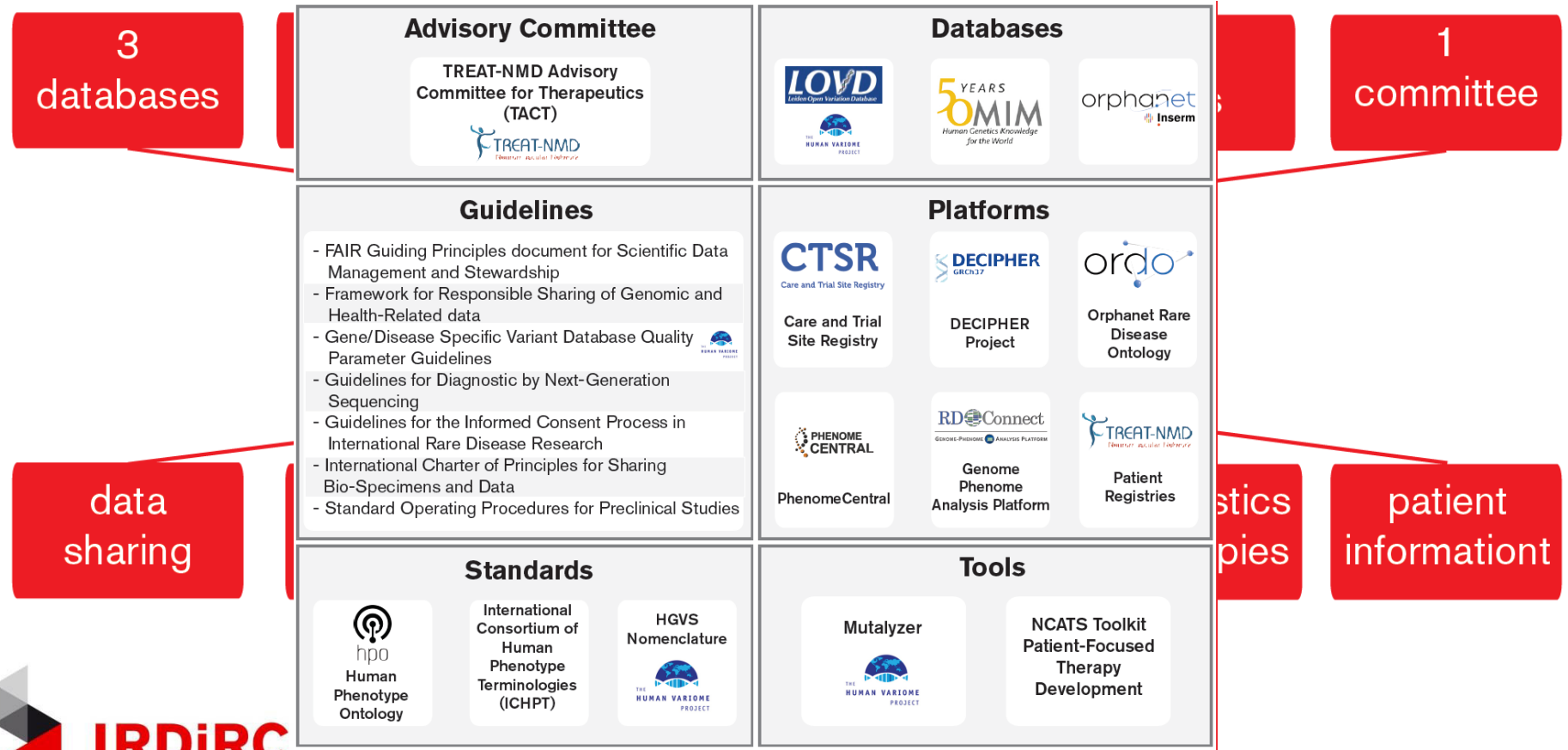
- ↪ Ontologies,
- ↪ Diagnostics,
- ↪ Biomarkers,
- ↪ Patient registries,
- ↪ Natural history studies, etc.

- ▶ They are adopted by IRDiRC member organizations through their own research programs and the programs they fund



# IRDIRC Recognized Resources

► Designation highlighting resources which contribute





# Process for Advancing the New IRDiRC Goals

- ▶ Based on new Goals announced Q3 2017, embarked on planning process for identifying top priority Activities to advance the Goals
  - ↳ Each Committee defined 3-5 Activities, with timelines and metrics
  - ↳ Fall 2017
    - ↳ Committee brainstorm
    - ↳ Consolidated and refined proposed Activities
  - ↳ Winter 2017-2018
    - ↳ Defined capacity and scope given status of current Task Forces and other activities
    - ↳ Prioritized Activities
    - ↳ Synthesized Activities into unified Roadmap for 2018
    - ↳ Committee and Consortium Assembly approved 2018 Roadmap
- ▶ *Current status:* Committees beginning to implement plans for each approved Activity within the Roadmap for 2018
  - ↳ Including development of metrics to measure success toward each goal

# The IRDiRC Mission

## *Transformation, not Incrementalism*

- ▶ Catalyze radically more efficient and effective paradigms
  - ↻ Many have been developed and demonstrated
- ▶ The common factor in all of these radical improvements:  
SHARING
  - ↻ of knowledge
  - ↻ of data
  - ↻ of infrastructure
  - ↻ of expertise
  - ↻ of viewpoints

# More information on IRDiRC

▶ Website:

↪ <http://www.irdirc.org/>

*Get involved! <http://www.irdirc.org/get-involved/>*

▶ Chair:

↪ Christopher Austin, [austinc@mail.nih.gov](mailto:austinc@mail.nih.gov)

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