The International Rare Diseases Research Consortium (IRDiRC)

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U.S. National Institutes of Health

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August 2, 2019

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International Rare Diseases Research Consortium (IRDiRC)

- About IRDiRC
  - "Unites… international governmental and non-profit funding bodies, companies, umbrella patient advocacy organizations, and scientific researchers to promote international collaboration and advance rare diseases research worldwide."
- Established 2011
  - Members from Europe, North America, Asia, Australia, Middle East

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About IRDiRC: http://www.irdirc.org/about-us/

IRDiRC: About (2)

- Initial focus on developing common scientific and policy frameworks
- Initial objectives 2011-2020:
  - 200 new therapies for rare diseases (RD) by 2020
  - Means to diagnose most RD by 2020
  - ***Achieved in 2017***
  - New goals established for 2017-2027

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Vision and Goals 2017-2027

Released 9 August 2017

- Vision: "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."
  - Goal 1: Receive a diagnosis within 1 year if disorder is known; undiagnosed individual enter a globally coordinated diagnostic pipeline
  - Goal 2: 1000 new therapies for RD approved
  - Goal 3: Develop methodologies to assess the impact of diagnoses and therapies on RD patients

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IRDiRC Vision and Goals: http://www.irdirc.org/about-us/vision-goals/

nature International weekly journal of science

NATURE | CORRESPONDENCE

Medical research: Next de diseases

Christopher P. Austin & Hugh J. S. Dawkins

Affiliations | Corresponding author

Nature 548, 158 (10 August 2017) | doi:10.1038
Published online 09 August 2017

Subject terms: Genetics · Medical research

The International Rare Diseases Research Consor
achieved its ambitious goals for 2020 — three years ahead of schedule (see *Nature* **472**, 17; 2011). The consortium has now forged a further se
people who have debilitating and lethal rare diseas

Rare diseases were once considered medical curi
negligible public-health impact. The molecular bas
However, diagnosis of most of these conditions re
approved treatments.

The new IRDiRC goals aim to achieve diagnosis w
a disorder being known, this will be accomplished
cases. Other goals are to develop 1,000 new thera
treatment, and to create methods for assessing th
patients' well-being. (For details, see H. J. S. Dawl
P. Austin *et al. Clin. Transl. Sci.*, in the press.)

The IRDiRC has nearly 50 organizations in 18 nati
than US$2 billion (see go.nature.com/2htbauh). Ne
coordinated effort.

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Hugh J. S. Dawkins

CTS Clinical and Translational Science
Open Access

Citation: Clin Transl Sci (2017) 00, 1–11; doi:10.1111/cts.12501

REVIEW

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

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

Citation: Clin Transl Sci (2017) 00, 1–7; doi:10.1111/cts.12500

REVIEW

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

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

C) was founded in 2011 with the
uncture. Proof of principle
ts successfully developed and
approved, and improvements in quality and quantity of life achieved. Government research
had all demonstrated their
rare diseases research.
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licative solutions. The scale of
vast preponderance of them
ysseys for many patients—led
tion and collaboration among
pitalize on these proofs of
efforts around the world.
erarching objectives: to
means to diagnose most rare
the history, governance, and
panying piece on the first 6

Process for Advancing the IRDiRC Goals
Roadmap

- Planning Process
  - Based on IRDiRC Goals, embarked on Roadmap planning process for identifying top priority Activities to advance the Goals
  - Each Committee defined 3-5 Activities, with timelines and metrics
  - Consolidated and refined proposed Activities
- Roadmap Generation
  - Defined capacity and scope given status of existing Task Forces/Activities
  - Prioritized Activities
  - Synthesized Activities into unified Roadmap
  - Discussion and approval with Committees and Consortium Assembly
- Implementation
  - Implemented plans for approved Activities within the Roadmap (completed for both 2018 and 2019)
  - Development of metrics to measure success toward each goal

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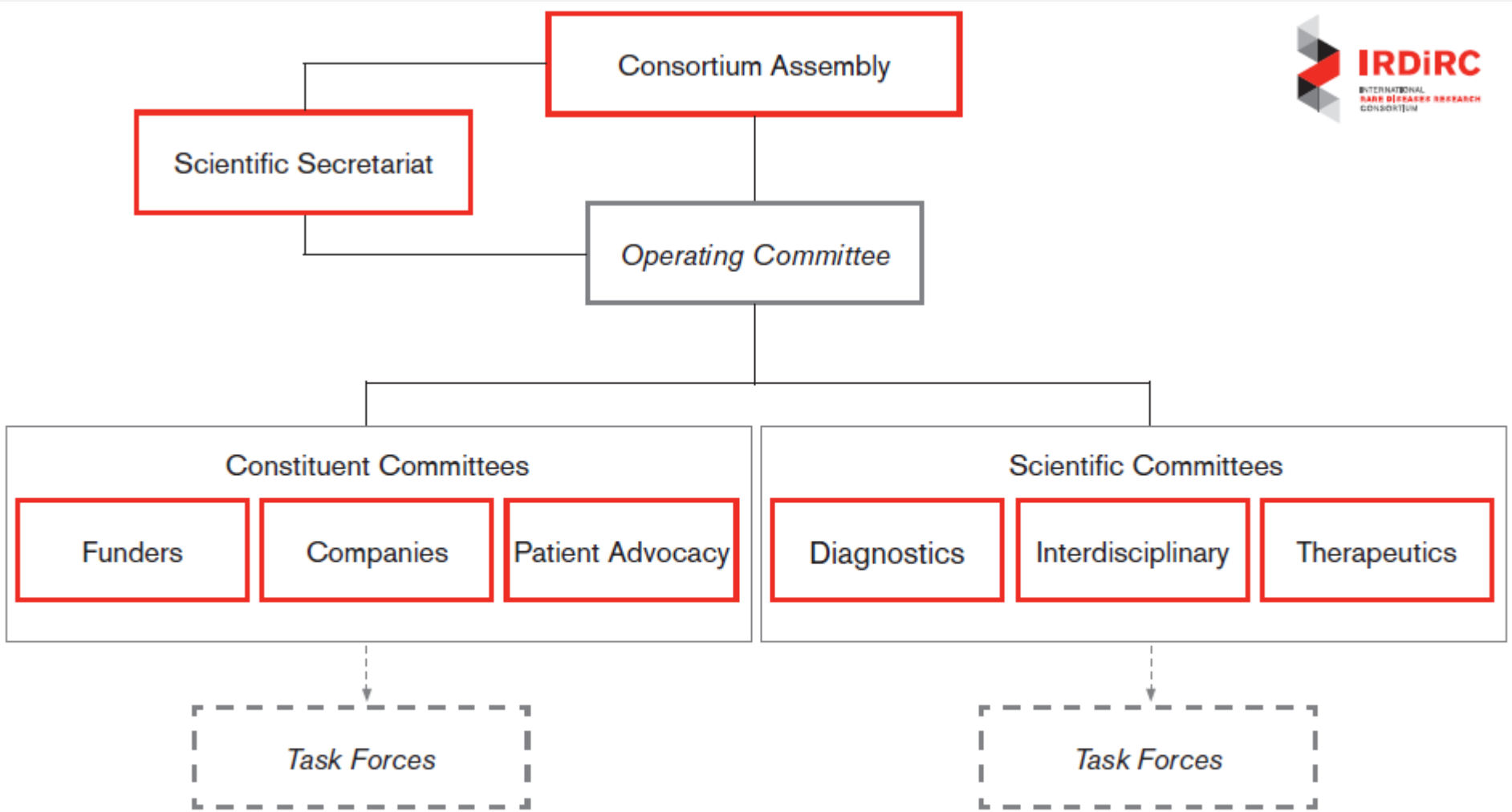
IRDiRC Structure



IRDiRC Consortium Assembly

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, National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS
- NIH, National Institute of Child Health and Human Development, NICHD
- NIH, National Institute of Dental and Craniofacial Research, NIDCR
- NIH, National Institute of Neurological Disorders and Stroke, NINDS
- NIH, National Human Genome Research Institute, NHGRI
- National Institute of Health Carlos III, ISCIII
- National Institute for Health Research
- 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 Next Code

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

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IRDiRC Consortium Assembly

NIH & FDA Representation

- NCATS
  - Anne Pariser
- NIAMS
  - Faye Chen
- NINDS
  - Adam Hartman
- NICHD
  - Melissa Parisi
- FDA, OOPD
  - Katherine Needleman

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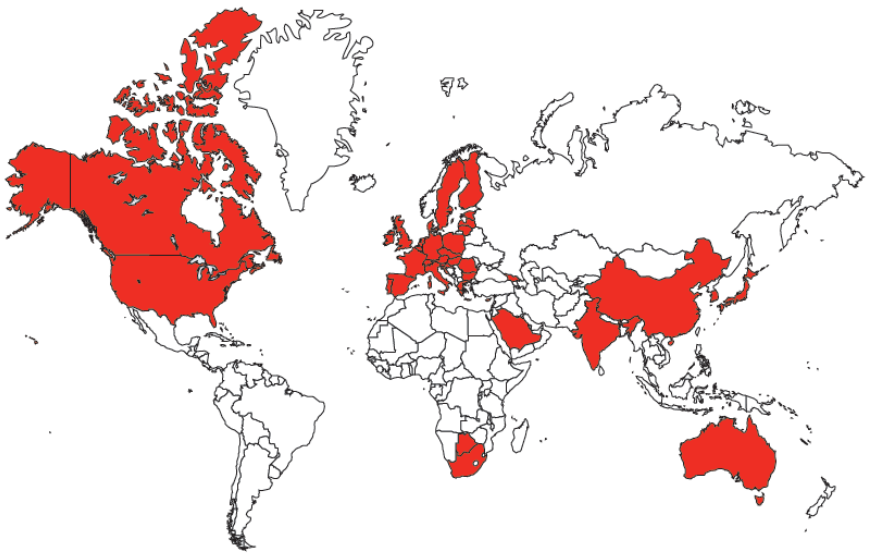
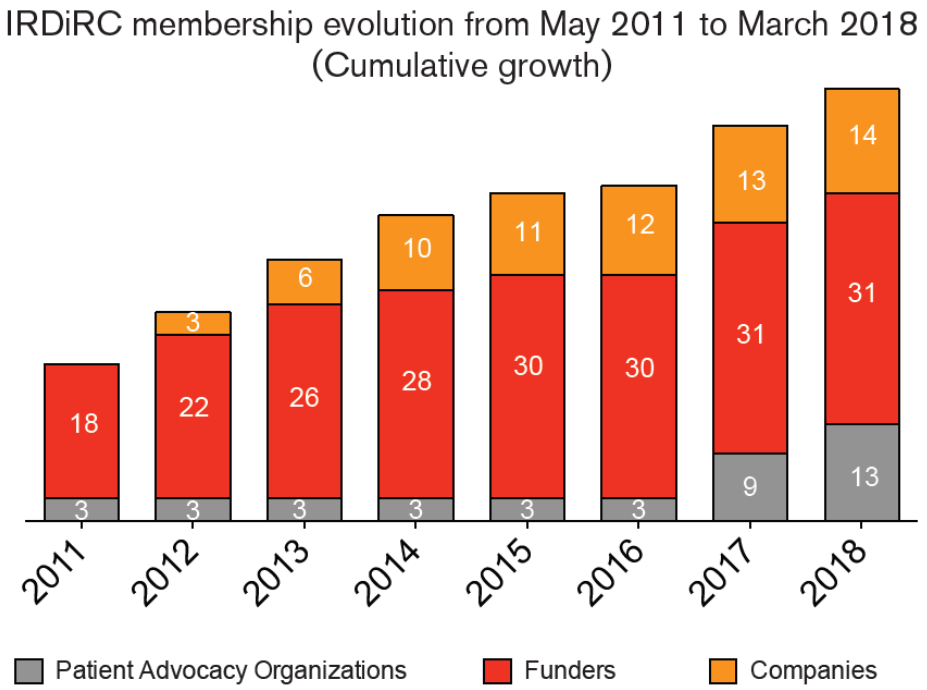
Representation





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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

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IRDiRC Committees
Mission

- Identify roadblocks/priorities
- Implement Task Forces and activities to address priorities/gaps
- Establish and promulgate best practices, operating procedures, quality standards, roadmap to address priorities
- Inform other Committees of scientific and programmatic states, needs, opportunities, emerging issues

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IRDiRC Constituent Committees
Chairs

- Funders
  - Chair: Daria Julkowska, Agence Nationale de la Recherche/E-Rare, France
  - Vice Chair: Adam Hartman, NINDS NIH
- Patient Advocates
  - Chair: Durhane Wong-Rieger, Canadian Organization for Rare Disorders
  - Vice Chair: Yukiko Nishimura, Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan
- Companies
  - Chair: Mathew Pletcher, Roche, Switzerland
  - Vice Chair: Madhu Natarajan, Shire, USA

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IRDiRC Scientific Committees
Chairs

- Diagnostics
  - Chair: Gareth Baynam, Health Department of Western Australia
  - Vice Chair: Sarah Bowdin, Addenbrooke's Hospital, Cambridge, UK
- Foundational (aka, Interdisciplinary)
  - Chair: Steve Groft, National Center for Advancing Translational Sciences, NCATS, NIH
  - Vice Chair: Dixie Baker, Martin, Blanck and Associates, USA
- Therapies
  - Chair: Diego Ardigò, Chiesi Pharmaceuticals, Italy
  - Vice Chair: Virginie Hivert, EURORDIS, France

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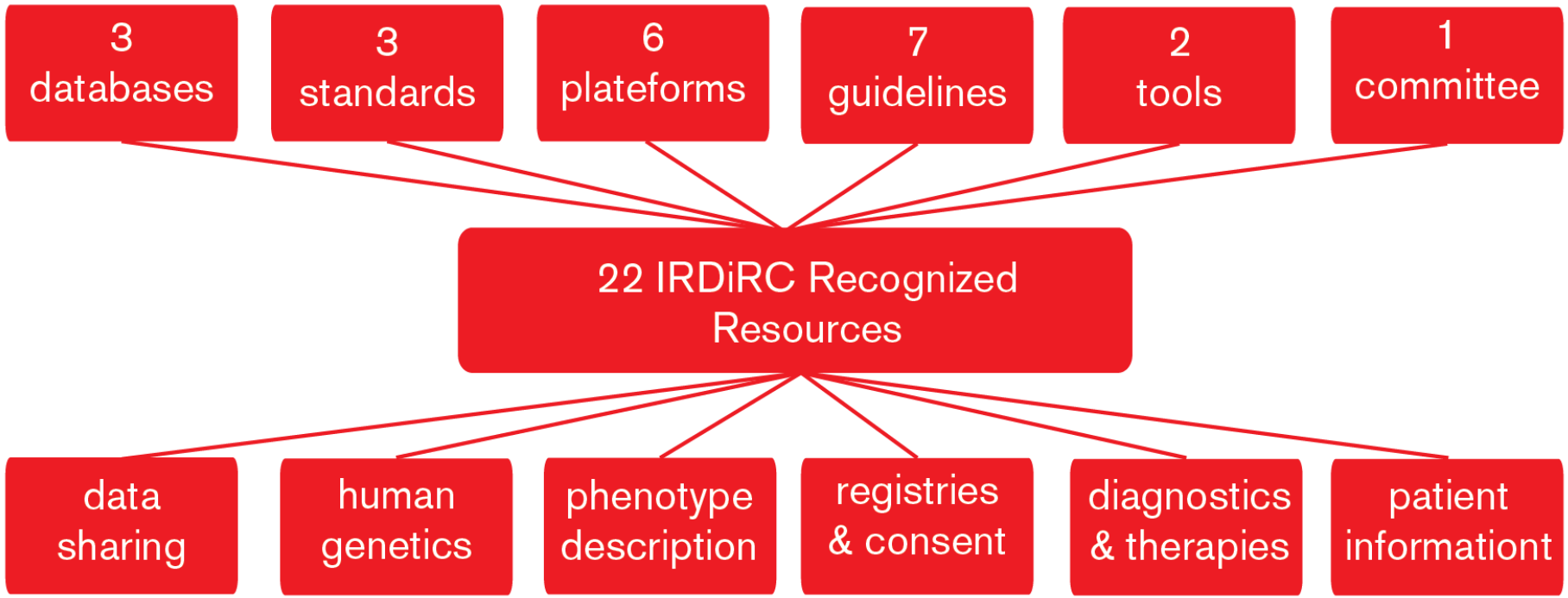
Current IRDiRC Task Forces

- Task forces established in 2015
  - Tackle specific topics of importance to RD research
  - ***Diagnostics Scientific Committee (DSC)***
    - Solving the Unsolved (STU)
    - Clinical Data Sharing (CDS)
    - *Carrier Screening*
    - *Underrepresented Populations*
  - ***Interdisciplinary Scientific Committee (ISC)***
    - Model Consent Clauses (MCC; joint effort with GA4GH)
    - *Clinical Research Network for Rare Diseases (CRNRD)*
    - *Facilitating the Conduct of Natural History Studies related to Rare Diseases*
  - ***Therapies Scientific Committee (TSC)***
    - Data Mining and Repurposing (DMR)
    - *Orphan Drug Development Guidebook (ODDG)*
    - *Support the Reframing of the Current International Research Agenda for RDs to Enable Achievement of Goal 2*

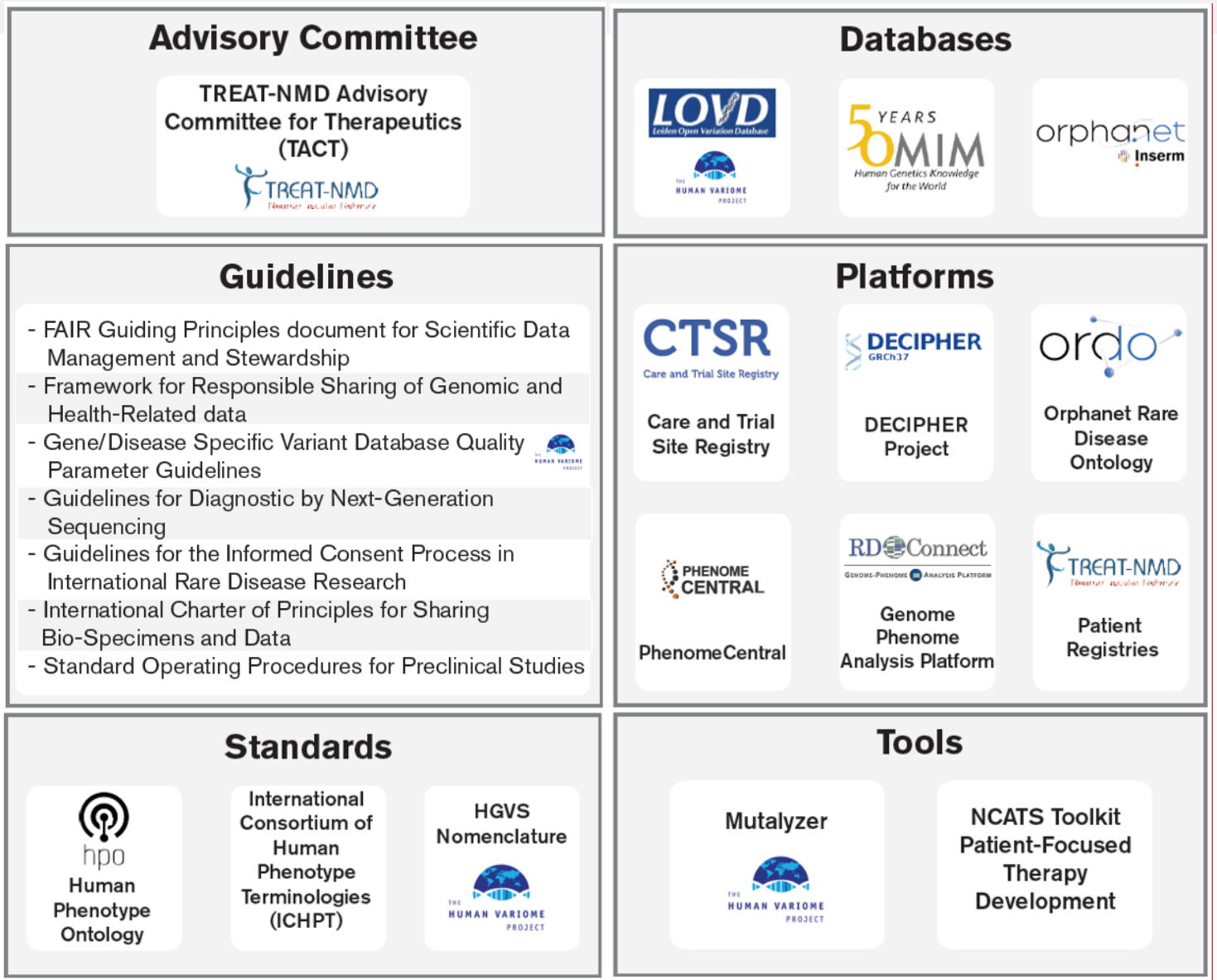
IRDiRC “Recognized Resources” 1

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

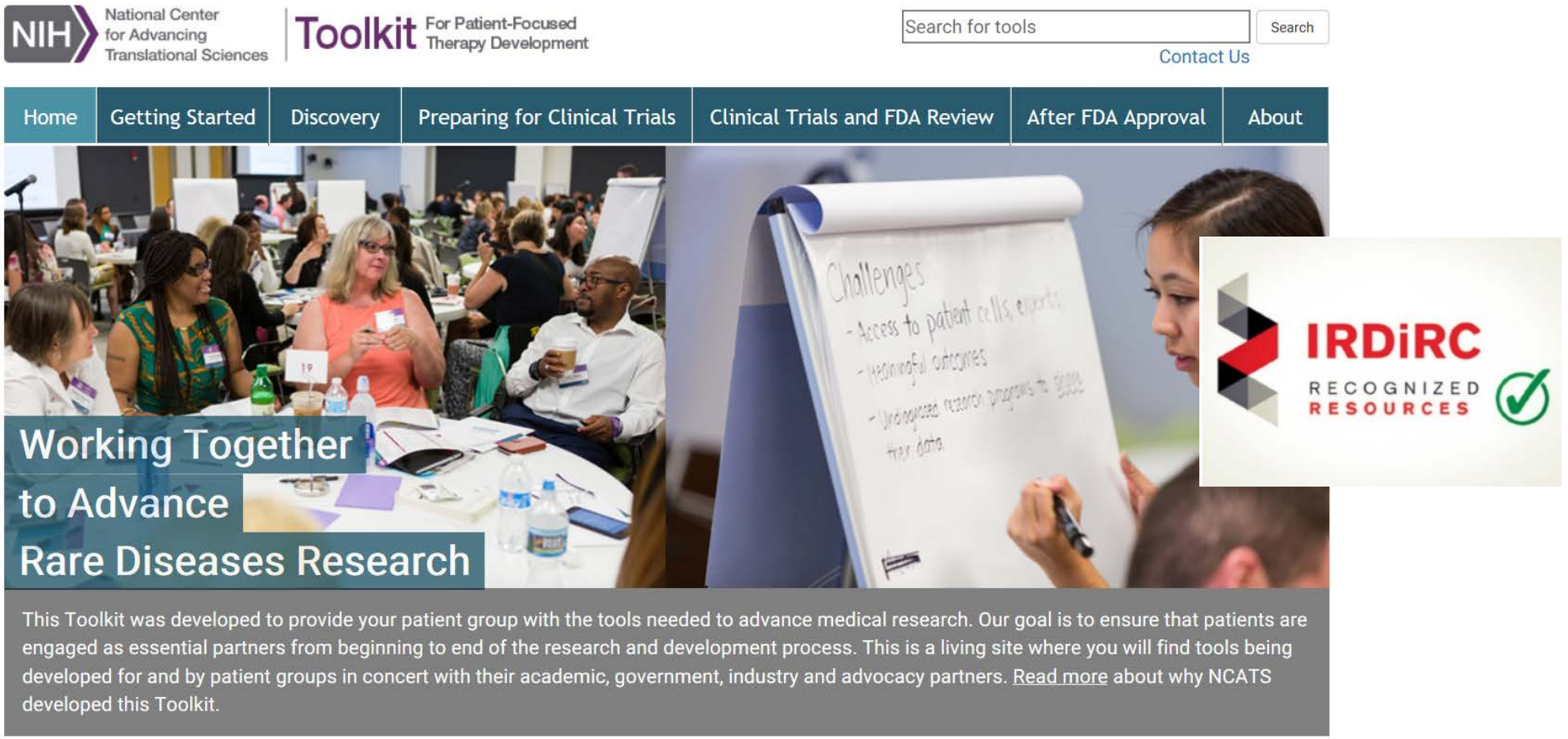
IRDiRC "Recognized Resources" 2



IRDiRC "Recognized Resources" 3



e.g., NCATS “Toolkit: for Patient-Focused Therapy Development IRDiRC Recognized Resource



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Toolkit: for Patient-Focused Therapy Development:
https://rarediseases.info.nih.gov/toolkit/home

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

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More information on IRDiRC

- Website:
  - http://www.irdirc.org/
- Chair:
  - Lucia Monaco, LMonaco@Telethon.it
- Vice Chair:
  - David Pearce, David.Pearce@SanfordHealth.org
- Secretariat:
  - contact@irdirc.org
    - NIAMS, Faye Chen — chenfl@mail.nih.gov
    - NINDS, Adam Hartman — adam.hartman@nih.gov
    - NICHD, Melissa Parisi — parisima@mail.nih.gov
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