

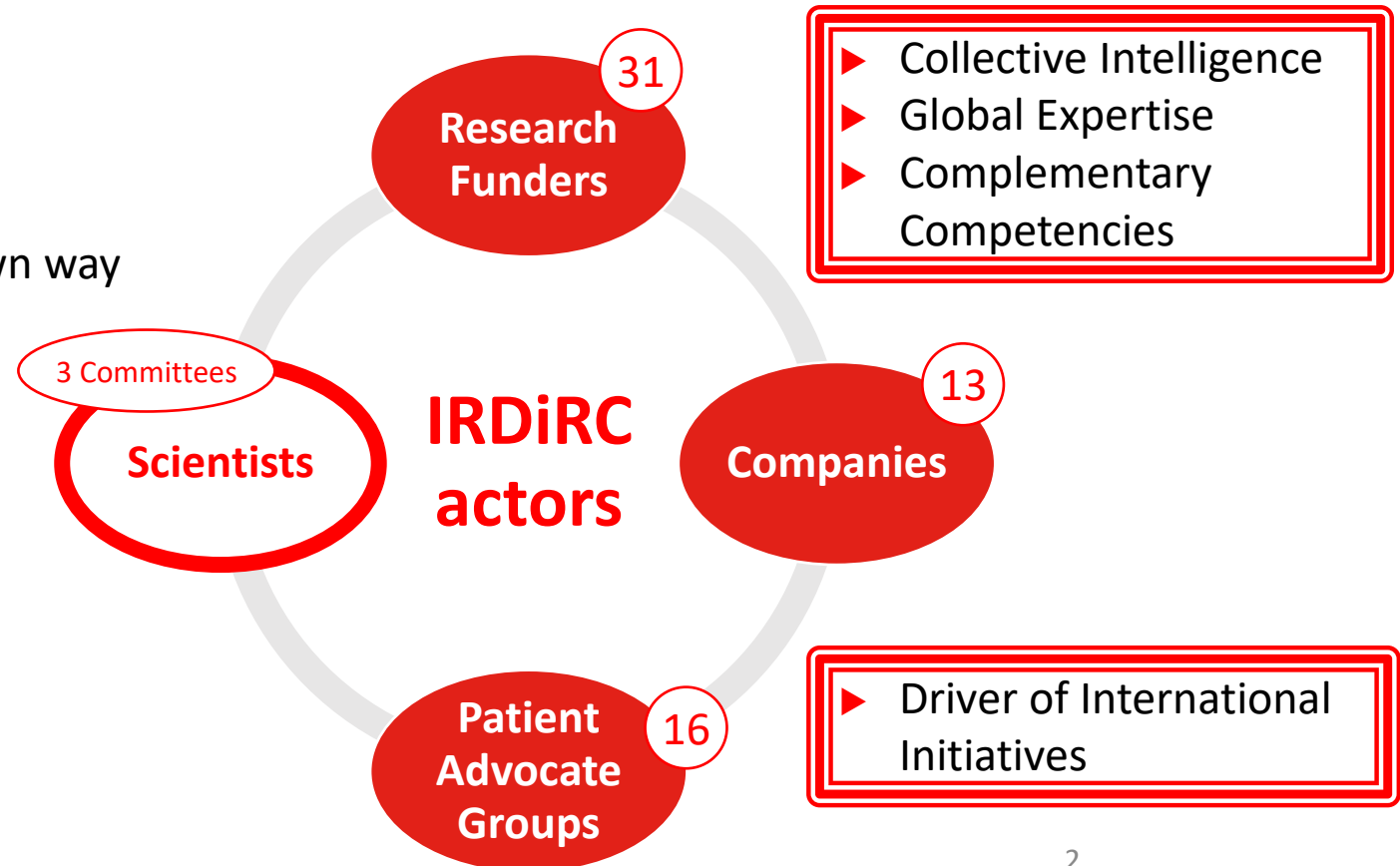
# International Rare Diseases Research Consortium (IRDIRC)

**David A. Pearce, PhD**  
**Chair, IRDiRC**


# International Rare Diseases Research Consortium (IRDiRC)


**Collaborate, stimulate, coordinate, and leverage output of rare diseases research efforts around the world**

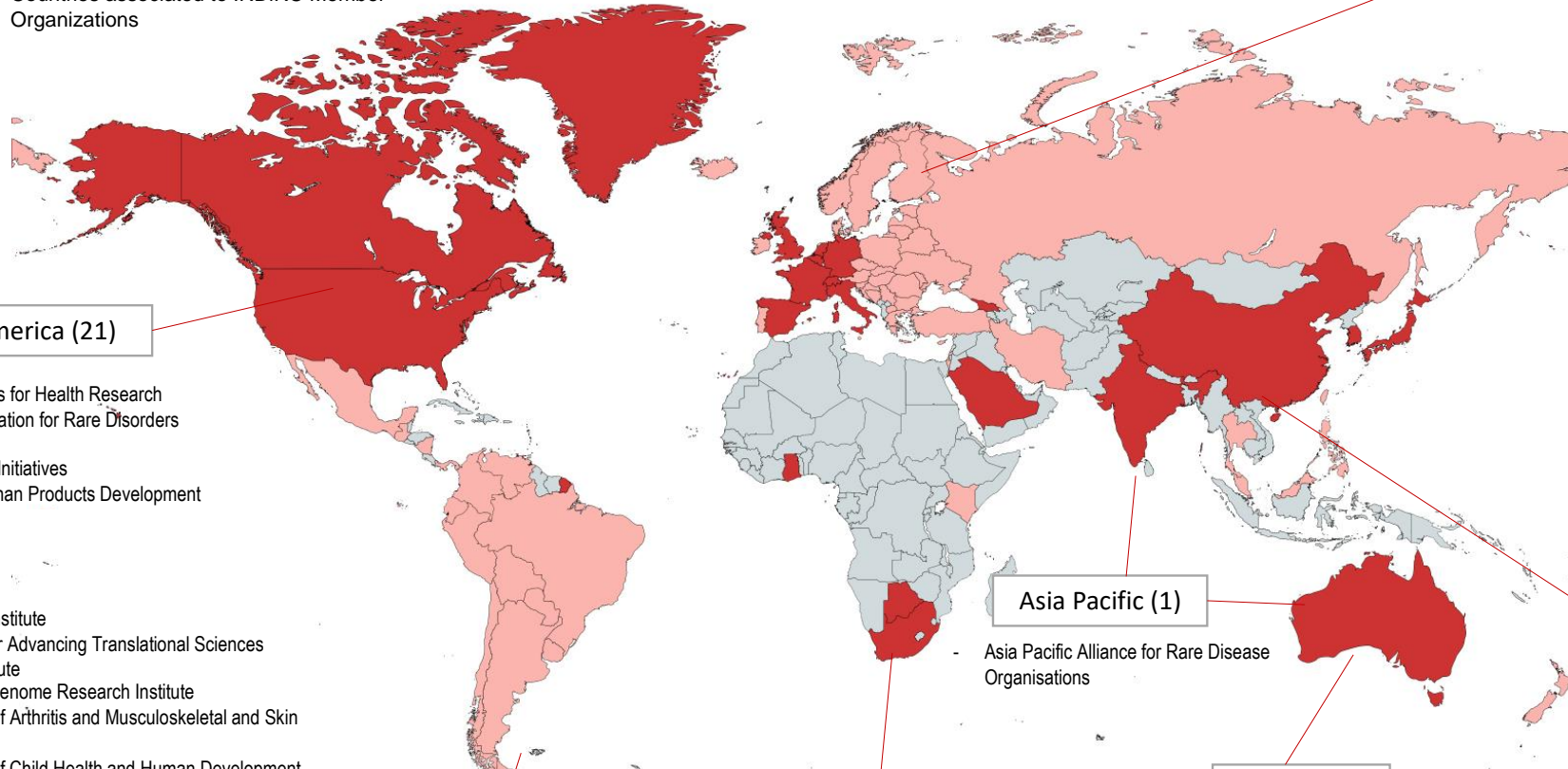
- Launched in 2011
- Unite **research funders** and **companies** investing in RD research
  - Each organization funds research its own way
  - Funded projects adhere to a common framework
- Umbrella **patient advocacy groups**
- **3 Scientific Committees**
  - Diagnostics
  - Therapies
  - Interdisciplinary



# 60 Member Organizations

 Countries hosting IRDiRC Member Organizations

 Countries associated to IRDiRC Member Organizations



**International (1)**

- Rare Diseases International

**Europe (21)**

- AFM-Telethon
- Agence nationale de la recherche
- Chiesi Farmaceutici
- Congenica
- European Commission
- EURORDIS-Rare Diseases Europe
- Federal Ministry of Education and Research (BMBF)
- Fondazione Telethon
- French Foundation for Rare Diseases
- French Muscular Dystrophy Association (AFM-Téléthon)
- French National Institute of Health and Medical Research (INSERM)
- Innoskel
- Georgian Foundation for Genetic and Rare Diseases
- Institute of Health Carlos III
- Istituto Superiore di Sanità
- LouLou Foundation
- Lysogene SA
- National Institute for Health Research
- Roche Biotechnology
- The Netherlands Organisation for Health Research and Development (ZonMw)
- Ultragenyx Pharmaceutical Inc.

**North America (21)**

- Canadian Institutes for Health Research
- Canadian Organization for Rare Disorders
- Cydan II, Inc.
- Chan Zuckerberg Initiatives
- FDA Office of Orphan Products Development
- Genetic Alliance
- Genome Canada
- Global Genes
- Illumina, Inc
- National Cancer Institute
- National Center for Advancing Translational Sciences
- National Eye Institute
- National Human Genome Research Institute
- National Institute of Arthritis and Musculoskeletal and Skin Diseases
- National Institute of Child Health and Human Development
- National Institute of Dental and Craniofacial Research
- National Institute of Neurological Disorders and Stroke
- National Organization for Rare Disorders
- Pfizer Inc.
- Recursion Pharmaceuticals
- Sanford Research

**Asia Pacific (1)**

- Asia Pacific Alliance for Rare Disease Organisations

**Africa (3)**

- Botswana Organization for Rare Diseases
- Rare Disease Ghana Initiative
- Rare Diseases South Africa

**Australia (2)**

- Rare Voices Australia
- Western Australian Department of Health

**Asia (10)**

- Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan
- BGI Genomics, China
- Chinese Organization for Rare Disorders
- Indian Organization for Rare Diseases
- Japan Agency for Medical Research and Development
- Korea Disease Control and Prevention Agency
- National Rare Diseases Registry System of China
- Organization for Rare Diseases India
- Saudi Human Genome Project
- Takeda Pharmaceuticals

**Latin America (1)**

- Iberoamerican Alliance for Rare Diseases

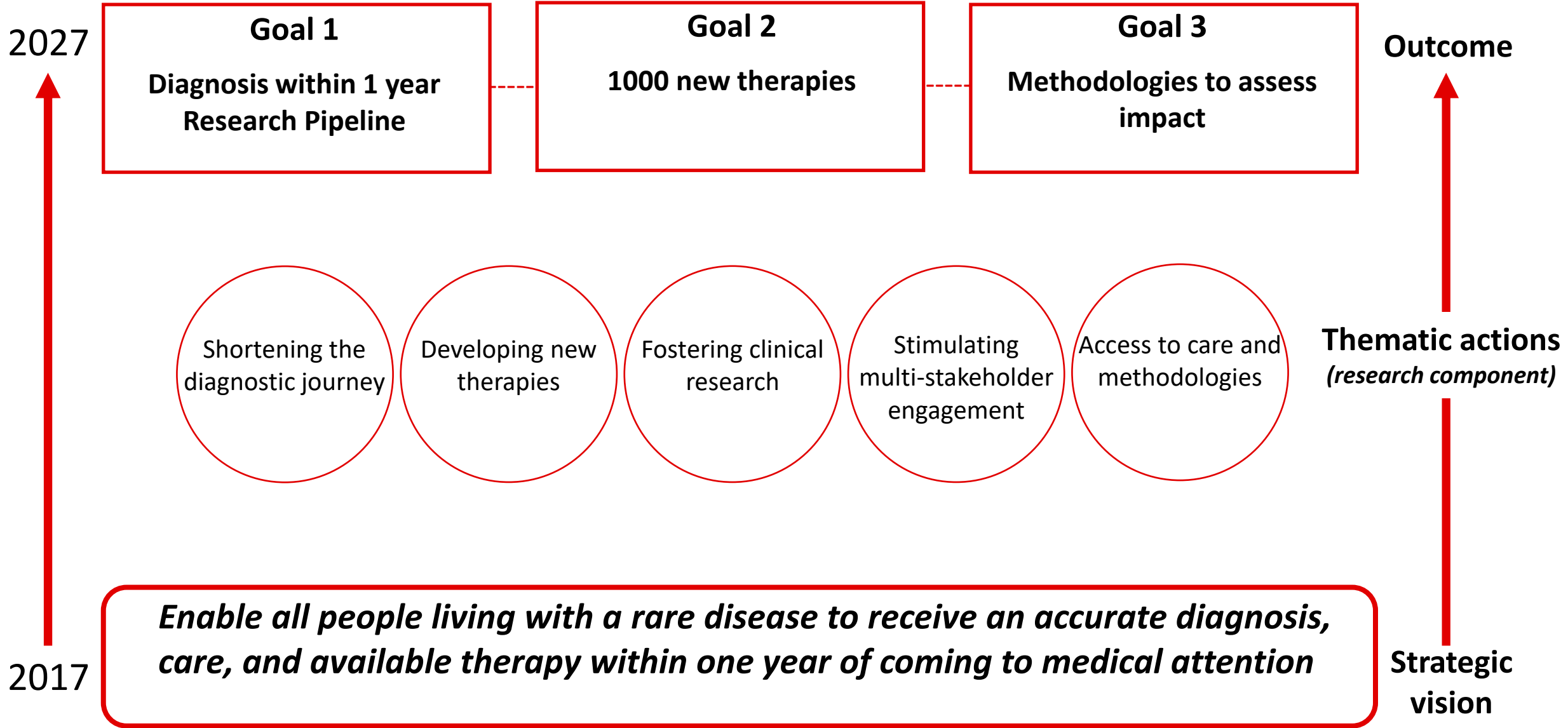
# IRDiRC Vision and Goals, by 2027

## Vision

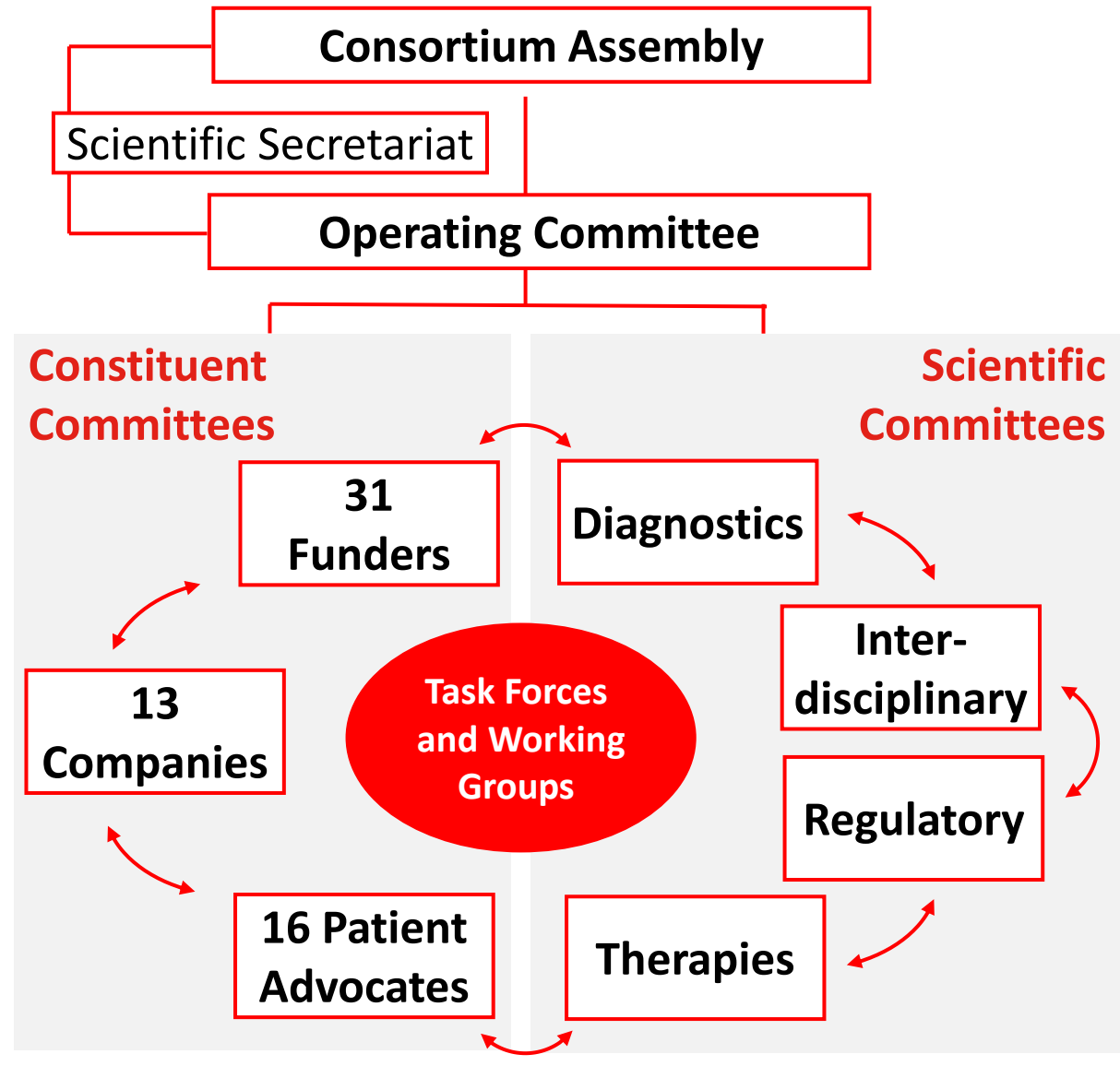
Enable all people living with a rare disease to **receive** an accurate **diagnosis, care,** and available therapy **within one year** of coming to medical attention

## Goals

1. All patients coming to medical attention with a suspected rare disease will be **diagnosed within one year** if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated **diagnostic and research pipeline**
2. **1000 new therapies** for rare diseases will be approved, the majority of which will focus on diseases without approved options
3. Methodologies will be developed to **assess the impact of diagnoses and therapies** on rare diseases patients

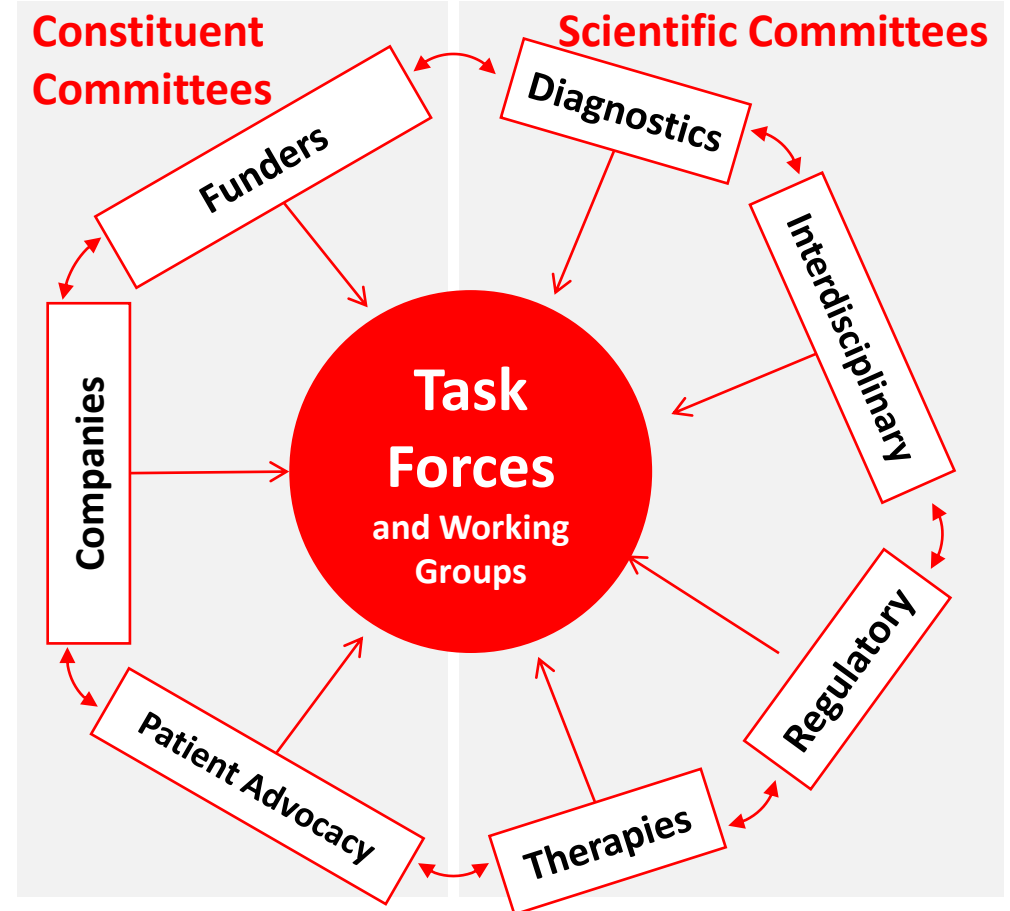


# Governance and Operating Model



# Working together towards IRDiRC Goals

- All Committees propose **priority actions** tackling the three IRDiRC goals
- **Task Forces** or Working Groups approved by the Consortium Assembly
- An **annual Road Map** is established by the Assembly
- Activities are managed by the **Scientific Secretariat**, integrated into the **EJP RD Coordination Office**



Nomination calls for **Task Forces/Working Groups** and **Scientific Committees** published on [www.irdirc.org](http://www.irdirc.org)

IRDiRC  
TFs/WGs  
December  
2021

Shortening the diagnostic journey

Developing new therapies

Fostering clinical research

Stimulating multi-stakeholder engagement

Access to care and impact methodologies

VISION

Goal 1  
Diagnosis within 1 year & Research Pipeline

Goal 2  
1000 new therapies

Goal 3  
Methodologies to assess impact

Matchmaker Exchange

Human Phenotype Terminologies

Solving the Unsolved

New Technologies for Diagnosis

Orphan Drug Development Guidebook

Data Mining and Repurposing

Sustainable Economic Models in Drug Repurposing

Drug Repurposing Guidebook

Pluto project - Disregarded RDs

Privacy-Preserving Record Linkage

Automatable Discovery and Access

Model Consent Clauses for RD Research

Machine Readable Consent Forms

Small Populations Clinical Trials

Patient - Centered Outcome Measures

Shared Molecular Etiologies

ELSI WG

Clinical Research Networks for RDs

Primary Care

Med Techs | Devices for RDs

Chrysalis Project

Indigenous Populations

RD Treatment Access WG

Indigenous Populations

Impact Methodologies

Tele-health

Completed

Ongoing

Planned/Starting