Rare Disease Research Overview

Lucia Monaco, Chair, International Rare Diseases Research Consortium Assembly

NATIONAL PRESS FOUNDATION
Online Workshop
Covering Rare Diseases
13 September 2021
Rare disease scenario

> 300 million people worldwide have a rare disease

- 70-80% affect children
- ~80% have a genetic origin
- 6,000 – 8,000 diseases are classified as rare
- ~80% are considered ultra-rare

- ~50% are life threatening diseases
- ~60% are serious and disabling diseases
- ~500 drugs have reached the market
- 700-800 treatments in development
- 5% of rare diseases have an approved treatment

~80% are considered ultra-rare
Rare disease disparities

- **Diverse**
  - legal recognition / definition
  - disease nomenclature

- **Scattered**
  - scientific knowledge
  - financial resources
  - clinical competence

- **Different**
  - geographic and socio-economic settings

- **Uneven**
  - diagnostic tests available
  - treatments available
  - access to diagnosis and care
Rare disease research challenges

- **Access to patients**
  - Clinical data
  - Biological material
  - Patient consent

- **Basic / preclinical research**
  - Data harmonization and sharing
  - Funding
  - Coordination / cooperation

- **Translational research/ drug development**
  - Patient recruitment in clinical trials
  - Clinical study design
  - Regulatory requirements
  - Profitability / sustainability

- **Patient access to treatments**
  - Marketing authorization
  - Pricing
  - Health system approval
Rare disease stakeholders – and stakes

**Players**
- People living with a rare disease
- Researchers
- Companies
- Regulators
- Health Systems

**Drivers**
- Solutions (diagnosis & treatments)
- Discovery
- Profit
- Safety
- Sustainability
International Rare Diseases Research Consortium (IRDiRC)

- Launched in 2011
- Teams up 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

International level co-operation to stimulate, better coordinate & maximize output of rare disease research efforts around the world

- Collective intelligence
- Global expertise
- Complementary competences
- Driver of international initiatives
62 IRDiRC Member Organizations from 22 Countries

- Europe 23
- North America 21
- Asia 11
- Africa 3
- Oceania 3
- South America 1

Stay updated on https://irdirc.org/
Key policies for research funders

- Promote **discovery of genes** underlying RDs
- Encourage **development of therapies**
- Encourage and facilitate rapid **data release**
- Promote **data** harmonization, interoperability and access to **registries and biobanks**
- Promote **coordination** between human and model systems research
- **Involve patients** in all relevant aspects of RD research

Key policies for researchers

- Perform **collaborative research**, share data and results, avoid duplication
- Make **registries and biobanks interoperable** and harmonized, with a global scope
- **Share and distribute** RD biomaterial through biobanks
- **Exchange scientific and regulatory information** on clinical research
- **Balance IP issues with the need to share information** to the benefit of the research and patient community
- Adhere to and develop **research standards**
- **Publish results in a timely manner** on peer-reviewed and possibly open access journals

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
Progress towards IRDiRC Goal 1

Cumulative number of genes linked to rare diseases since 2010

Number of genes


Number of genes linked to rare diseases:
- 2543 in 2011
- 2876 in 2012
- 3184 in 2013
- 3372 in 2014
- 3506 in 2015
- 3674 in 2016
- 3843 in 2017
- 4013 in 2018
- 4138 in 2019
- 4211 in 2020

https://irdirc.org/research/rd-metrics/
Progress towards IRDiRC Goal 2

Cumulative number of new orphan drugs since 2010

https://irdirc.org/research/rd-metrics/
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 activities - completed

IRDiRC GOALS

GOAL 1 DIAGNOSIS

- Diagnosis beyond genetic analysis

GOAL 2 THERAPIES

- Drug repurposing for RD
- Patient data, privacy and consent
- Small-population clinical trials
- Patient-centered outcome measures

GOAL 3 IMPACT

- Orphan drug development

Completed

IRDiRC

https://irdirc.org/activities/task-forces/
Proposal of a core set of terms to describe human phenotypes by the International Consortium of Human Phenotype Terminologies

Introduction
Collectively rare diseases constitute a substantial healthcare burden, although difficult to quantify as most coding nomenclatures do not include specific codes for these diseases. Although each...
Tools for the community
Research and innovation

INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM
Small Population Clinical Trials Task Force Workshop Report and Recommendations
July 2016

A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers

Position Statement

The use or generation of biomedical data and existing medicines to discover and establish new treatments for patients with rare diseases – recommendations of the IRDirc Data Mining and Repurposing Task Force

Measuring what matters to rare disease patients – reflections on the work by the IRDirc taskforce on patient-centered outcome measures

https://irdirc.org/activities/task-forces/
IRDiRC ongoing/new activities
Research and development for RD

GOAL 1
DIAGNOSIS

- Disregarded rare diseases

GOAL 2
THERAPIES

- Making RD research attractive to industry

GOAL 3
IMPACT

- Drug repurposing for RD

Ongoing

https://irdirc.org/activities/task-forces/
IRDiRC ongoing /new activities
Innovation in clinical RD research

- Leveraging the existing clinical ecosystem
- Widening patient access to clinical trials
- Medical devices for RD

GOAL 1
DIAGNOSIS

GOAL 2
THERAPIES

GOAL 3
IMPACT

IRDiRC GOALS

BASIC RESEARCH ➔ PRECLINICAL RESEARCH ➔ CLINICAL RESEARCH ➔ DEVELOPMENT ➔ AUTHORIZATION ➔ ACCESS

Ongoing
New

https://irdirc.org/activities/task-forces/
IRDiRC ongoing /new activities
Access and impact

IRDiRC GOALS

GOAL 1 DIAGNOSIS

GOAL 2 THERAPIES

GOAL 3 IMPACT

IRDiRC BASIC RESEARCH
PRECLINICAL RESEARCH
CLINICAL RESEARCH
DEVELOPMENT
AUTHORIZATION
ACCESS

- Diagnosis for Indigenous populations
- Tele-medicine
- Primary care
- Access barriers to therapies
- Impact of diagnosis and therapies on peoples lives

https://irdirc.org/activities/task-forces/
IRDiRC’s widening horizons

GOAL 1 DIAGNOSIS
- Diagnosis beyond genetic analysis
- Disregarded rare diseases
- Drug repurposing for RD
- Patient data,
- Implementing new diagnostic tools
- Leveraging the existing clinical ecosystem
- Making RD research attractive to industry
- Widening patient access to clinical trials
- Drug repurposing for RD
- Orphan drug development
- Medical devices for RD
- Impact of diagnosis and therapies on peoples lives

GOAL 2 THERAPIES
- Diagnosis for Indigenous populations
- Tele-medicine
- Primary care
- Access barriers to therapies

GOAL 3 IMPACT

IRDiRC GOALS

BASIC RESEARCH
PRECLINICAL RESEARCH
CLINICAL RESEARCH
DEVELOPMENT
AUTHORIZATION
ACCESS

https://irdirc.org/activities/task-forces/
THANK YOU!

https://irdirc.org/

IRDiRC Consortium Assembly meeting
Paris, 21-22 November 2019