International Rare Diseases Research Consortium (IRDiRC)

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC Vision, by 2027

- 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

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IRDiRC Goals, by 2027

- 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

- 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options

- Methodologies will be developed to assess the impact of diagnoses and therapies on rare diseases patients

IRDiRC – Basic Principles

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

- Teams up public and private organizations investing in RD research
- Research funders with relevant programs >$10 million over a 5-year
- Each organization funds research its own way
- Funded projects adhere to a common framework

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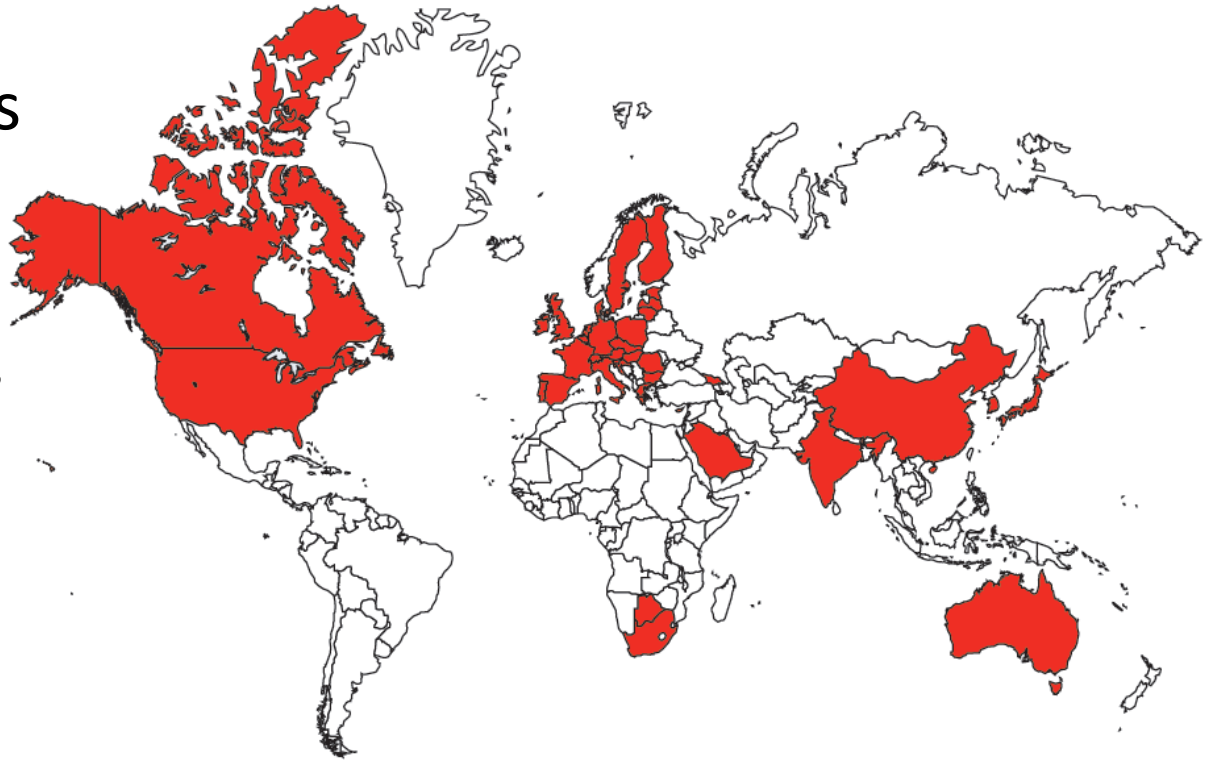
IRDiRC's Members

- 58 IRDiRC members
  - 31 funders
  - 14 companies
  - 13 patient advocates organizations



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Patient-Centered Outcome Measures

- To boost the development and adoption of patient-centered outcome measures
- Explore to whether, how and to what extent these initiatives can be expanded to target rare disease research in order to improve feasibility and quality of trials
- Post-workshop report and recommendations available on IRDiRC website

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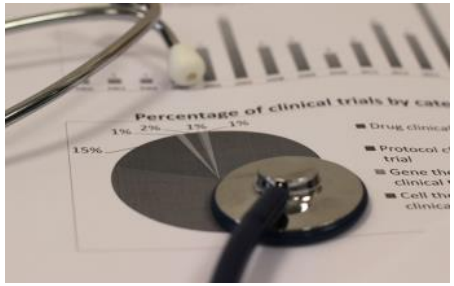
Small Population Clinical Trials

- Contribute consensus about non-conventional statistical methods used for small population clinical trials
- Contribute to the acceptability of new statistical methods and coordinate with the different agencies; EMA, FDA, industry, IDEAL, INSPIRE, ASTERIX
- Post-workshop report and recommendations available on IRDiRC website



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Solving the Unsolved

- Identification of the genetic basis of rare conditions presently intractable to existing approaches
- Based on exome sequencing requires development of innovative approaches for discovery
- The objective is to bring together the community addressing this challenge to share best practices regarding approaches

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Patients Advocacy Constituent Committee (PACC)

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PACC members


Sharon Terry (Chair)
Genetic Alliance, USA


Yukiko Nishimura (Vice-Chair)
AsRid, Japan


Eda Selebasto
BORDIS, Botswana


Durhane Wong-Rieger
CORD, Canada


Kevin Huang
CORD, China


Virginie Bros-Facer
EURORDIS-Rare Diseases Europe


Nicole Boice
Global Genes, USA


Ramaiah Muthyala
I-ORD, India


Peter Saltonstall
NORD, USA


Prasanna Kumar Shirol
ORD-I, India


Ritu Jain
Rare Diseases International, Singapore


Kelly du Plessis
RDSA, South Africa


Nicole Millis
Rare Voices Australia, Australia

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Mission

- Brings together representatives from the umbrella patient advocacy organization members of IRDiRC

- Highlights the importance of patient involvement in rare diseases research, with a strong emphasis on the inclusion of patients' needs and perspectives in all relevant aspects of research and the need for worldwide representation