IRDiRC INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

International Rare Diseases Research Consortium (IRDiRC)

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InTBIR

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IRDiRC: International Rare Disease Research Consortium

- Heterogeneous pathophysiology & dispersed nature of rare diseases: research & development efforts, patient populations are scattered around the world
- Work largely atomized: each organization, each country, and champions of each disease pursuing independent, often duplicative solutions

IRDiRC: International Rare Disease Research Consortium

- Founded in 2011, IRDiRC is a consortium that unites public and private sector funders of research, patient advocacy groups, and scientific researchers
- Dr Ruxandra Draghia-Akli of the Directorate-General for Research and Innovation (DG RTD) of the European Commission (EC) and Dr Francis Collins of the US National Institutes of Health (NIH) had initial discussions that led to the formation of IRDiRC
- Membership based on track record of investing > $10 mil/ year in rare diseases research funding over 5 years

IRDiRC: International Rare Disease Research Consortium

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 disease patients.*

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Examples of deliverables

- eRare-RDCRN collaboration
- Matchmaker Exchange
- Provide guidance on key topics: ontologies, diagnostics, biomarkers, patient registries, biobanks, natural history studies, therapeutics, models, publications, intellectual property, and communication
- IRDiRC Task Forces
  (1) International Consortium of Human Phenotype Terminologies
  (2) Patient-Centered Outcome Measures Task Force
  (3) Small Population Clinical Trials Task Force
  (4) Data Mining and Repurposing Task Force

Source:IRDiRC

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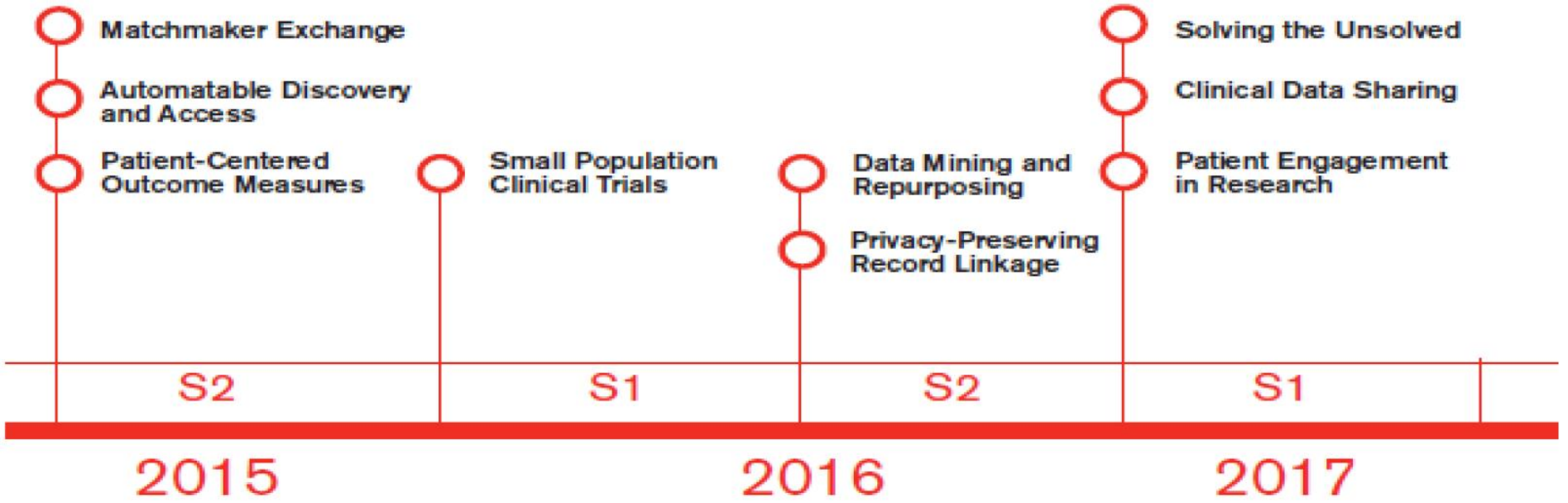
Task Forces



National Institute of Neurological Disorders and Stroke

Source:IRDiRC

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Problem: data sharing

- Linking a genetic variant with a disease requires finding ≥2 unrelated individuals who share genetic variant & phenotype
- Thus, need data sharing
- Obstacles
  - Competitive academic framework
  - Limitations in consent obtained from research participants
  - Lack of standardized vocabulary to describe phenotypes, further complicating the identification of patients with similar symptoms

Source:IRDiRC

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Partial solutions: data sharing

1. Matchmaker Exchange: collaboration between IRDiRC, Global Alliance for Genomics and Health (GA4GH) and other organizations
2. *Automatable Discovery and Access* Task Force: developing standardized way to represent consent & other conditions of clinical data use, making information computer-readable & available for automated search/sharing
3. *International Consortium of Human Phenotype Terminologies:* provided community with standards of interoperability between databases by enabling linkage of phenotype and genotype databases for rare diseases

Source:IRDiRC

Partial solutions: data sharing

4. *Privacy-Preserving Record Linkage* Task Force: develop guidelines on ethical, legal, and technical requirements of participant identifiers in rare diseases research, and investigate technical solution for de-duplication of research participants in datasets without knowledge of their identities

References for data sharing

Rath, A., et al., *Representation of rare diseases in health information systems: the Orphanet approach to serve a wide range of end users.* Hum Mutat, 2012. **33**(5): p. 803-8.

Ayme, S., B. Bellet, and A. Rath, *Rare diseases in ICD11: making rare diseases visible in health information systems through appropriate coding.* Orphanet J Rare Dis, 2015. **10**: p. 35.

Mascalzoni, D., et al., *International Charter of principles for sharing bio-specimens and data.* Eur J Hum Genet, 2015. **23**(6): p. 721-8.

McCormack, P., et al., *'You should at least ask'. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research.* Eur J Hum Genet, 2016. **24**(10): p. 1403-8.