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The International Rare Diseases Research Consortium (IRDiRC)

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Global Genomic Medicine Collaborative

International Rare Disease Research Consortium (IRDiRC)

- Co-operation at international level to stimulate, better coordinate & maximize output of rare disease research efforts around the world
- A 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

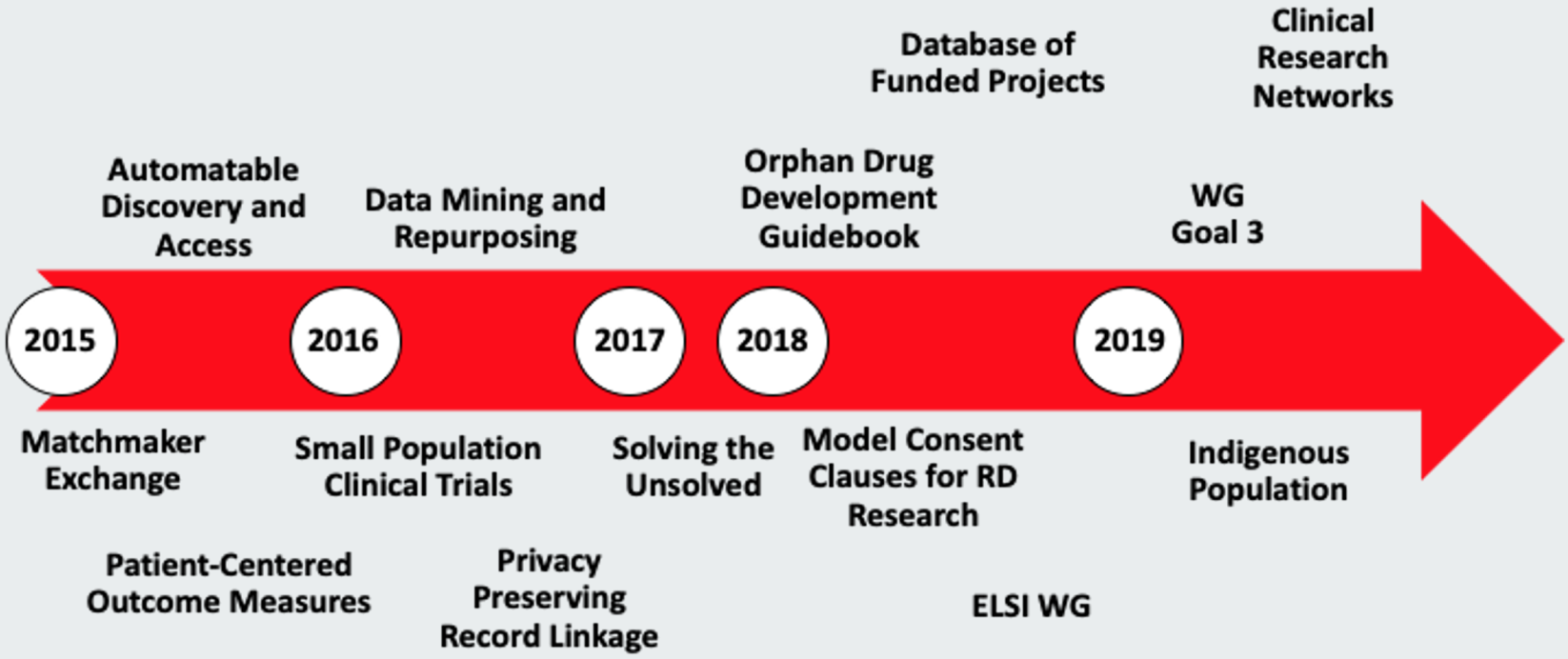
IRDiRC Goals, by 2027

- Goal 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
- Goal 2
  - 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options
- Goal 3
  - Methodologies will be developed to assess the impact of diagnoses and therapies on rare diseases patients

Task Forces – *Overarching themes*

- Address specific RD research needs or bottleneck
- Projects are identified and developed by IRDiRC committees
- Projects can sometimes be developed in collaboration with other organizations
- Involve IRDiRC members and external experts
- Provide solution through
  - Policy Recommendations
  - Technical Applications

Task Forces & WG



IRDiRC Diagnostics Scientific Committee (DSC)

- Committed to support IRDiRC's diagnostic Goal 1 for 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".
- Identifies current and future bottlenecks to rare disease gene discovery, addresses challenges and roadblocks in rare disease diagnosis, and collaborates with international partners to develop tools and resources to facilitate genomic data discovery, analyses and sharing.

Current Composition

- 15 members
  - Australia, China, India, Israel, Japan, Netherlands, Saudi Arabia, South Africa, Spain, Sweden, United Kingdom, United States

Ongoing Task Force

Task Force on RD diagnostics for Indigenous Population and underrepresented populations

- To address barriers to RD diagnostics for Indigenous peoples so as to improve access to diagnostics in underserved populations
- Composed of 18 experts with broad geographic spread (Africa, AUS, CA, EU, NZ, US) and diverse expertise (patient advocates; aboriginal clinical geneticists, aboriginals researches)
- Workshop March 10, 2020, Berlin

New Directions (JR & Co)?

Yachay's promise

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Human Genomics

MEETING REPORT | Open Access

Meeting report: the Human Genome Meeting (HGM) 2019 in Seoul, Korea

Angela Solano[1], Giuseppe Novelli[2*], Shruti Baghat[3], Piero Carninci[3], Gert-Jan van Ommen[4] and Juergen K. V. Reichardt[5*]

nature

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Ecuador's research dream crushed by politics

Juergen Reichardt

nature genetics

Comment | Published: 23 December 2019

A call for global action for rare diseases in Africa

Gareth S. Baynam, Stephen Groft, Francois H. van der Westhuizen, Safiyya D. Gassman, Kelly du Plessis, Emily P. Coles, Eda Selebatso, Moses Selebatso, Boikobo Gaobinelwe, Tebogo Selebatso, Dipesalema Joel, Virginia A. Llera, Barend C. Vorster, Barbara Wuebbels, Benjamin Djoudalbaye, Christopher P. Austin, Judit Kumuthini, John Forman, Petra Kaufmann, James Chipeta, Désirée Gavhed, Annika Larsson, Maja Stojiljkovic, Ann Nordgren, Emilio J. A. Roldan, Domenica Taruscio, Durhane Wong-Rieger, Kristen Nowak, Gemma A. Bilkey, Simon Easteal, Sarah Bowdin, Juergen K. V. Reichardt, Sergi Beltran, Kenjiro Kosaki, Clara D. M. van Karnebeek, Mengchun Gong, Zhang Shuyang, Ruty Mehrian-Shai, David R. Adams, Ratna D. Puri, Feng Zhang, Nicholas Pachter, Maximilian Muenke, Christoffer Nellaker, William A. Gahl, Helene Cederroth, Stephanie Broley, Maryke Schoonen, Kym M. Boycott & Manuel Posada - Show fewer authors

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