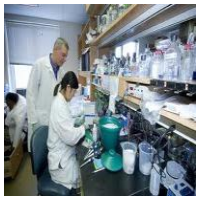
Welcome address: 2nd IRDiRC Conference

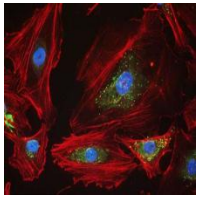
IRDiRC

INTERNATIONAL
RARE
DISEASES
RESEARCH
CONSORTIUM

Paul Lasko, PhD
Chair, International Rare Disease Research Consortium (IRDiRC)
Scientific Director, Institute of Genetics, Canadian Institutes of Health Research

IRDiRC—ambitious goals (diagnostic tools for all rare diseases, 200 new therapies by 2020)





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News

Rare-disease project has global ambitions

Consortium aims for hundreds of new therapies by 2020.

Alison Abbott

Prader–Willi syndrome. Fabry renal disease. Spinocerebellar ataxia. Few people have heard of these and the other 'rare diseases', some of which affect only hundreds of patients worldwide. Drug companies searching for the next blockbuster pay them little attention. But the diseases are usually incurable — and there are thousands of them.

This week, the US National Institutes of Health (NIH) and the European Commission launch a joint assault on these conditions, whose small numbers of patients make it difficult to test new treatments and develop diagnostic methods. The International Rare Disease Research Consortium being formed under the auspices of the two bodies has the ambitious goal of developing a diagnostic tool for every known rare disease by 2020, along with new therapies to treat 200 of them. "The number of individuals with a particular rare disease is so small that we need to be able to pool information from patients in as many countries as possible," says Ruxandra Draghia-Akli, the commission's director of health research.

Canada

IRDiRC – basic principles

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

- Teams up public and private organisations investing in rare diseases research
- Research funders can join & work together
- Each organisation funds research its own way
- Funded projects adhere to a common framework

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Growth of IRDiRC

- Third coordination meeting was held in Montreal (9-10 Oct 2011), sponsored by IG and Genome Canada.
- Initially involved EU, NIH, Canada, Italy, Spain.
- Funder members commit a minimum of USD 10M to rare disease research over a 5-yr period.
- Present commitment exceeds $1B worldwide.
- Formally launched in 2012, first public symposium held in Dublin in April 2013.
- Second public symposium happening now in Shenzhen, China, November 7-9, 2014.

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Australia
- Western Australian Department of Health

Canada
- Canadian Institutes for Health Research
- Genome Canada

China
- Beijing Genomics Institute
- Chinese Rare Disease Research Consortium
- WuXi Apptec

EU
- European Commission

Finland
- Academy of Finland

France
- French Association against Myopathies
- Agence National de la Recherche
- Lysogene
- EORTC

Georgia
- Children's New Hospital Management Group

Germany
- Federal Ministry of Education and Research

Italy
- Italian Higher Institute of Health
- Telethon Foundation
- Chiesi Farmaceutica

International Consortium
- E-RARE 3 Consortium

Republic of Korea
- Korean National Institute of Health

Netherlands
- The Netherlands Organization for Health Research and Development
- Prosensa

Kingdom of Saudi Arabia
- Saudi Human Genome Project

Spain
- National Institute of Health Carlos III

UK
- National Institute for Health Research

USA
- Food and Drug Administration Orphan Products Grants Program
- Isis Pharmaceuticals
- National Human Genome Research Institute (NIH)
- National Center for Advancing Translational Sciences(NIH)
- National Cancer Institute (NIH)
- National Eye Institute (NIH)
- National Institute of Neurological Disorders and Stroke (NIH)
- National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIH)
- National Institute of Child Health and Human Development (NIH)
- NKT Therapeutics
- Office of Rare Diseases (NIH)
- PTC Therapeutics
- Sanford Research Institute

- International Pharma Companies
  - Genzyme (Sanofi)
  - Shire

Executive Committee

PATIENT ORGANIZATIONS

Béatrice de Montleau

- European Organisation for Rare Diseases (EURORDIS)
- Europe

Peter Saltonstall

- National Organization for Rare Disorders (NORD)
- USA

Sharon Terry

- Genetic Alliance
- USA

Principles applying to research activities

Sharing and collaborative work in RD research

- Sharing of data and resources
- Rapid release of data
- Interoperability and harmonization of data
- Data in open access databases

Scientific standards, requirements and regulations in RD research

- Projects should adhere to IRDiRC standards
- Develop ontologies and biomarkers
- Cite use of databases and biobanks in publications

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Principles applying to research activities

Participation by patients and / or their representatives in research

- Act in the best interest of patients
- Involve patients in all aspects of research
- Involve patients in governance of registries
- Involve patients in the design, conduct and analysis of clinical trials
- Acknowledge patients' contribution in articles

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Principles applying to funding bodies

- Promote the discovery of genes
- Promote the development of therapies
- Fund pre-clinical studies for proof of concept
- Promote harmonization, interoperability, sharing, open access to data
- Promote coordination between human and animal models
- Promote active exchanges between stakeholders through dissemination of ongoing projects and events

Progress toward IRDiRC's goals
(source: www.irdirc.org)

- Diagnostics (goal: most rare diseases by 2020)
  - >3,200 genes linked to rare diseases at end of 2013, as compared with 2,400 in 2010.
  - >3,100 rare diseases for which there is a genetic test available, as compared with 2,200 in 2010.
- Therapies (goal: 200 new therapies by 2020)
  - 131 as of October 2014.

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Challenges to overcome

- Cross-geographical area funding
- International data sharing
- Patient privacy/concerns
- Regulatory issues
- Ambitious timeline
- Translation from bench to bedside
- Lengthy drug development

IRDiRC Governance

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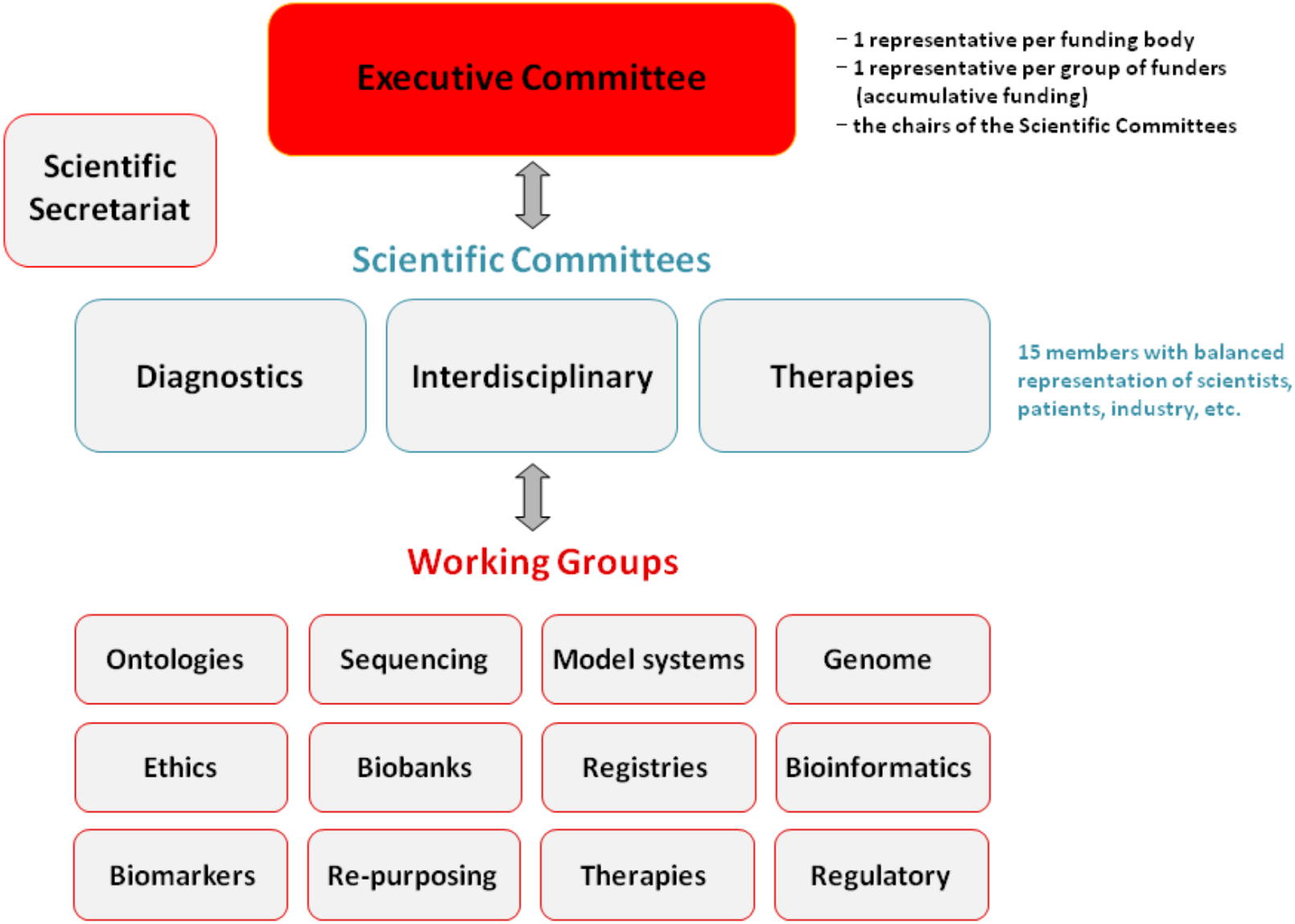
IRDiRC Organizational Structure



IRDiRC Scientific Secretariat

- In place in Paris, at the Rare Disease Platform, supported by an EU grant
- Management of the network / assistance to scientific committees and working groups
- Launch of the website:

WWW.IRDIRC.ORG

Diagnostics Scientific Committee (DSC)

Kym Boycott (chair)

- Children's Hospital Eastern Ontario
- Canada

Han Brunner

- Nijmegen University Hospital
- Netherlands

Michael Bamshad

- Seattle Children's Hospital
- USA

Johan Den Dunnen

- Center for Human and Clinical Genetics
- Netherlands

Xavier Estivill

- Genomic Regulation Centre
- Spain

Milan Macek

- Charles University Prague
- Czech Republic

Gert Matthijs

- University Hospital Leuven
- Belgium

Woong-Yang Park

- Samsung Genome Institute
- South Korea

Pak-Chung Sham

- Chinese Rare Disease Research Consortium
- China

Hendrik Stunnenberg

- Radboud University
- Netherlands

Jun Wang

- BGI
- China

Feng Zhang

- WuXi AppTec Co., Ltd.
- China

Interdisciplinary Scientific Committee (ISC)

Hanns Lochmüller (chair)

- Newcastle University
- UK

Alastair Kent

- Genetic Alliance
- UK

Jamel Chelly

- National Institute of Health and Medical Research
- France

Jeffrey Krischer

- University of South Florida
- USA

Angel Carracedo

- University of Santiago de Compostela
- Spain

Bartha Maria Knoppers

- McGill University
- Canada

Jack Goldblatt

- Genetic Services & Familial Cancer Program of Western Australia
- Australia

Samantha Parker

- Lysogene
- France

Stephen Groft

- Ex-NCATS
- USA

Rumen Stefanov

- Medical University of Plovdiv
- Bulgaria

Petra Kaufmann

- NCATS
- USA

Domenica Taruscio

- Italian National Centre for Rare Diseases
- Italy

Therapies Scientific Committee (TSC)

Yann Le Cam (chair)

- EURORDIS (France)

Gert-Jan Van Ommen

- Leiden University Medical Centre (Netherlands)

Giles Campion

- Prosensa Therapeutics (Netherlands)

Seng Cheng

- Rare Diseases Science Genzyme Corporation (USA)

Shuling Guo

- Isis Pharmaceuticals (USA)

Adam Heathfield

- Pfizer (UK)

Virginie Hivert

- EURORDIS (France)

Fulvio Mavilio

- Genethon (France)

John McKew

- NIH (USA)

Elizabeth McNeil

- NIH (USA)

Luigi Naldini

- Telethon Institute (Italy)

Glen Nuckolls

- NIH (USA)

Asla Pitkänen

- University of Eastern Finland (Finland)

Karin Rademaker

- University Medical Centre (Netherlands)

Robert Schaub

- NKT Therapeutics (USA)

Josep Torrent i Farnell

- Spanish Medicines Agency (Spain)

Ellen Welsh

- PTC Therapeutics (USA)

Anne Zajicek

- NICHD (USA)

IRDiRC Activities

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Organization of IRDiRC Conferences

- First IRDiRC Conference
  - In Dublin, Ireland
  - April 16-17, 2013
  - Organized by the European Commission

- Second IRDiRC Conference
  - In Shenzhen, China
  - November 7-9, 2014
  - Organized by BGI

IRDiRC Recommended

- Label to be used in highlighting tools, standards and guidelines, which contributes directly to IRDiRC objectives
- Application for 'IRDiRC Recommended' label is open to all, including non-IRDiRC members
- 'IRDiRC Recommended' may be awarded to similar tools, standards and guidelines
- Submission of 1-2 page application
- Evaluation of the application by a review panel
- Approval/rejection of the application by the Executive Committee

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RECOMMENDED

IRDiRC ongoing initiatives

Release of a core set of >2,300 terms common to all terminologies

- Agreement to define a core set of terms common to all terminologies and a methodology
- Selection of 2,370 terms
- Establishment of the International Consortium for Human Phenotype Terminologies – ICHPT


S. Aymé


A. Hamosh


P. Robinson

Establishment of a Data Standards Clearinghouse for Rare Diseases

- To promote data standards and contribute to their adoption, accessible from the IRDiRC website

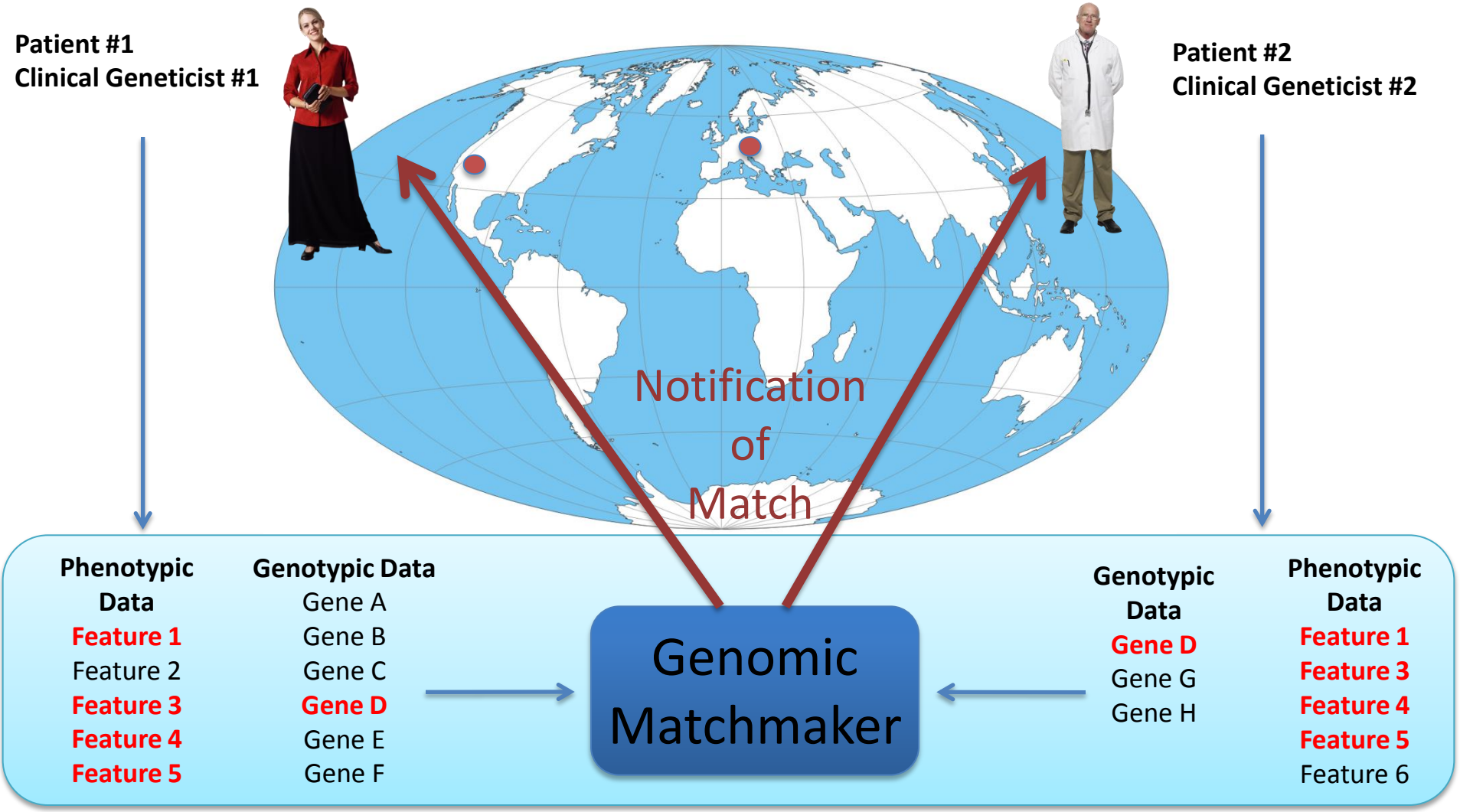

J. Krischer

Development of Model Consent Clauses for Rare Diseases

- Based on robust bioethical and legal approaches to allow rare disease researchers to select relevant consent clauses based on their research contexts and participant populations


Bartha Knoppers

Genomic Matchmaker



Courtesy of Joel Krier

The Challenge

Global Alliance for Genomics & Health

- ~50-75% of exome sequencing cases are unsolved
- No unified database exists for patients with unsolved rare genetic diseases
- Phenotypic data is under-represented in most genomic datasets

Matchmaker Exchange Project

Global Alliance for Genomics & Health

- Collaborative effort
- Federated platform (exchange)
- Facilitate the matching of unsolved genome/exome sequence cases
- Based on similar phenotypic and genotypic profiles (matchmaking)

Thanks for your attention!

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