Fostering patient involvement in international research

International Rare Disease Research Consortium
Sharon F. Terry, Genetic Alliance
Disclaimer

I do not speak on behalf of the International Rare Disease Research Consortium – these are my own thoughts and have not been shared with the Executive Committee
International Rare Disease Research Consortium

Cooperation at international level to stimulate, better coordinate and maximize output of rare disease research efforts around the world
IRDiRC – basic principles

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

- Teams up public and private organizations investing in rare diseases research
  - Research funders with relevant programs >$10 million US over a 5-year period can join & work together
  - Each organization funds research its own way
  - Funded projects adhere to a common framework
Goals of IRDiRC by 2020

200 new therapies for rare diseases

Means to diagnose most rare diseases
Governance structure

Executive Committee

Scientific Secretariat

Scientific Committees

Diagnostics
- Genome/Phenome
- Model Systems
- Ontologies and Rare Diseases Prioritization
- Population Controls Variant Datasets
- Sequencing

Interdisciplinary
- Biobanks
- Bioinformatics and Data Sharing
- Ethics and Governance
- Registries and Natural History

Therapies
- Biomarkers for Disease Progression & Therapies Responses
- Biotechnology-Derived Products Including Cell & Gene-Based Therapies
- Chemically-Derived Products Including Repurposing
- Orphan Drug-Development and Regulatory Processes

Members with balanced representation of scientists, patients, industry, etc.

Representatives of funded projects or patients organizations, experts, etc.
# Executive Committee: FUNDING AGENCIES

<table>
<thead>
<tr>
<th>Name</th>
<th>Agency</th>
<th>Country</th>
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</table>
| Paul Lasko (chair of IRDiRC) | • Canadian Institutes for Health Research  
• Canada                                      |          |
| Christopher Austin        | • National Center for Advancing Translational Sciences (NIH)  
• USA                                           |          |
| Pedro Cortegoso Fernández | • National Institute of Health Carlos III  
• Spain                                         |          |
| Hugh Dawkins              | • Western Australian Department of Health  
• Australia                                       |          |
| Ruxandra Draghia-Akli     | • European Commission  
• Europe                                          |          |
| Enrico Garaci             | • Istituto Superiore de Sanita  
• Italy                                           |          |
| Stephen Katz              | • National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIH)  
• USA                                           |          |
| Nicolas Lévy              | • French Foundation for Rare Diseases  
• France                                         |          |
| Pamela McInnes            | • Office of Rare Diseases (NIH)  
• USA                                           |          |
| Pierre Meulien            | • Genome Canada  
• Canada                                         |          |
| Lucia Monaco              | • Telethon Foundation  
• Italy                                          |          |
| Katherine Needleman       | • Office of Orphan Products Development (FDA)  
• USA                                           |          |
## Executive Committee: FUNDING AGENCIES

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<tbody>
<tr>
<td>Marie-Christine Ouillade</td>
<td>French Muscular Dystrophy Association • France</td>
<td>France</td>
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<tr>
<td>Willem Ouwehand</td>
<td>National Institute for Health Research • UK</td>
<td>UK</td>
</tr>
<tr>
<td>Melissa Parisi</td>
<td>National Institute of Child Health and Human Development (NIH) • USA</td>
<td>USA</td>
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<tr>
<td>Hyun-Young Park</td>
<td>Korea National Institute of Health • South Korea</td>
<td>South Korea</td>
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<tr>
<td>Jeffery Schloss</td>
<td>National Human Genome Research Institute (NIH) • USA</td>
<td>USA</td>
</tr>
<tr>
<td>Ralph Schuster</td>
<td>Federal Ministry of Education and Research • Germany</td>
<td>Germany</td>
</tr>
<tr>
<td>Bertrand Schwartz</td>
<td>Agence Nationale de la Recherche • France</td>
<td>France</td>
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<tr>
<td>Danilo Tagle</td>
<td>National Institute of Neurological Disorders and Stroke (NIH) • USA</td>
<td>USA</td>
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<td>Edward Trimble</td>
<td>National Cancer Institute (NIH) • USA</td>
<td>USA</td>
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<tr>
<td>Santa Tumminia</td>
<td>National Eye Institute (NIH) • USA</td>
<td>USA</td>
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<tr>
<td>Sonja van Weely</td>
<td>The Netherlands Organisation for Health Research and Development • Netherlands</td>
<td>Netherlands</td>
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<tr>
<td>Heikki Vilen</td>
<td>Academy of Finland • Finland</td>
<td>Finland</td>
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Executive Committee

INDUSTRY

Karen Aiach
• Lysogene
• France

Andrea Chiesi
• Chiesi Farmaceutici S.p.A.
• Italy

Luc Dochez
• Prosensa
• Netherlands

Diane Goetz
• PTC Therapeutics
• USA

Robert Mashal
• NKT Therapeutics
• USA

Brett Monia
• Isis Pharmaceuticals
• USA

Albert Seymour
• Shire
• USA

RESEARCH & DEVELOPMENT SERVICE

Carlo Incerti
• Genzyme
• USA

Mao Mao
• WuXi AppTec Co., Ltd.
• China
### Executive Committee

#### RESEARCH ORGANIZATIONS

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<tr>
<th>Name</th>
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<tr>
<td>Denis Lacombe</td>
<td>European Organisation for Treatment &amp; Research on Cancer (EORTC)</td>
<td>Belgium</td>
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<tr>
<td>Ning Li</td>
<td>Beijing Genomics Institute (BGI)</td>
<td>China</td>
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<tr>
<td>David Pearce</td>
<td>Sanford Research</td>
<td>USA</td>
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<tr>
<td>Qing K Wang</td>
<td>Chinese Rare Disease Consortium</td>
<td>China</td>
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#### HOSPITALS

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<tr>
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<tbody>
<tr>
<td>Oleg Kvolidze</td>
<td>Children’s New Hospitals Management Group</td>
<td>Georgia</td>
</tr>
<tr>
<td>Sultan Turki AlSedairy</td>
<td>Saudi Human Genome Project</td>
<td>Kingdom of Saudi Arabia</td>
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#### RARE DISEASE PROJECT

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<tr>
<td>Daria Julkowska</td>
<td>E-RARE 2 Consortium</td>
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#### CONSORTIUM OF RESEARCH INSTITUTES

#### CONSORTIUM OF FUNDING BODIES
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
How to Involve More Patients in the Executive Committee

- Executive Committee
  - Umbrella organizations – usually large national, regional, and international, well established bodies
  - The Executive Committee invites representatives of umbrella organisations of patient advocacy groups to serve on the Committee. These umbrella organisations should represent broad patients' interests for a wide variety of diseases and geographical area. The representatives are considered as full members of the Executive Committee.
How to Involve More Patients Broadly

- Serve on the various scientific committees
  - Diagnostic
  - Interdisciplinary
  - Therapeutic
- Serve on task forces
Task Forces will be constituted according to the following objectives:

- Topics specific to rare diseases
- High leverage projects with strong translational potential and international scope
- Actions for international scope and relevance
- Projects that have not been covered by international initiatives
- Well targeted action, with potential to produce results before 2020
- Actionable projects with secured human and financial resources
- Clear objectives and timelines to improve participation and member motivation
- Coordination with other organisations to identify gaps and needs
- Alignment and harmonisation of projects with other international initiatives
Overall

- Consider the policies within your country
- Advocate for more collaboration between academia, industry and government
- Encourage sharing of precious resources
- Collaborate internationally
- Present evidence for the need whenever possible
- Celebrate diversity and commonality
To Contact IRD IRC

- [http://www.irdirc.org](http://www.irdirc.org)
- Speak with any member: Yann Le Cam, Béatrice de Montleau, Peter Saltonstall, Sharon Terry
- Email the Secretariat: Ségolène Aymé