Paul Lasko, PhD
Chair, International Rare Disease Research Consortium (IRDiRC)
Scientific Director, Institute of Genetics, Canadian Institutes of Health Research
• Australia
  — Western Australian Department of Health
• Canada
  — CIHR
  — Genome Canada
• China
  — Beijing Genomics Institute
• EU
  — European Commission
• France
  — French Association against Myopathies
  — Agence National de la Recherche
  — Lysogene
• Germany
  — Federal Ministry of Education and Research
• Italy
  — Italian Higher Institute of Health
  — Telethon Foundation
• International Consortium
  — E-RARE 2 Consortium
• Netherlands
  — The Netherlands Organization for Health Research and Development
  — Prosensa
• Spain
  — National Institute of Health Carlos III
• UK
  — National Institute for Health Research
• USA
  — Food and Drug Administration Orphan Products Grants Program
  — 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)
  — National Eye Institute (NIH)
  — NKT Therapeutics
  — Office of Rare Diseases (NIH)
  — PTC Therapeutics
  — Sanford Research Institute
  — Shire
More members wanted!

• Many countries remain unrepresented.
• Some public-sector funders who are active in the rare disease area are absent.
• *Many* companies are missing.

To join:
• Demonstrated commitment of $10M USD for research in rare diseases in the next 5 years.
• Agreement with IRDiRC policies and guidelines, now ratified.
IRDiRC Scientific Secretariat

• In place in Paris, at the Rare Disease Platform
• Management of the network / assistance to scientific committees and working groups
• Launch of the website:

WWW.IRDiRC.ORG
Analysis of the research landscape

- Collection of research projects funded since 2010 in all participating countries to be released on Orphanet (www.orpha.net)
- Analysis of overlaps and of areas for possible collaboration
- Identification of trends / emerging fields to be supported in the future
Indicators of output

• Research activity
  – No. of IRDiRC members / allocated budget

• For diagnostics
  – No. of new causative genes discovered / No. of clinical tests of diagnostics / No. of diseases with available test / by country and by region

• For therapeutics
  – No. of designations / No. of clinical trials / Attrition rate / No. of diseases covered / Type of products
  – No. of disease registries / sources of clinical data
Goals of IRDiRC

200 new therapies for rare diseases by 2020

Means to diagnose most rare diseases by 2020
200 new therapies, diagnostics for nearly all rare diseases

• Are these goals sufficient?
• How will you help? What is your personal goal?

—Tell us at contact@irdirc.org!