IRDiRC Achievements and Road Map

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Achievements so far
Launched in April, 2011, with ambitious goals by 2020

Rare-disease project has global ambitions


BY ALISON AGBut

Launched in April, 2011, with ambitious goals by 2020.

RARE-DISEASE RESEARCH

P rader-Willi syndrome, Fabry renal disease, Spinal muscular atrophy. Few people have heard of these or other rare diseases, some of which affect only hundreds of patients worldwide. Drug companies searching for the next blockbuster pharma hit frown at these conditions. 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 launched a joint initiative 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 (IRDiRC) was formed under the auspices of the two bodies to address the ambitious goal of developing a diagnostic tool for every rare disease by 2020, along with new therapies to treat 700 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 Renata Douakis-Agladze, the consortium’s director of health research.

At the launch meeting in Bethesda, Maryland, on 4–5 April, prospective partners will map out research strategies to identify diagnostic biomarkers, design clinical trials and coordinate genome sequencing in those diseases. Nearly all the rare diseases, of which there are an estimated 6,000–8,000, are the result of small genetic changes.

The meeting will also discuss the governance of the project, which is most likely to be modelled on the successful Human Genome Project. As such, the consortium is open to research agencies and organizations from all over the world. Representatives from countries including Canada, Japan and some individual European nations will all attend the meeting, and may join the consortium. Those wishing to participate will have to pledge a minimum financial contribution, which has not yet been agreed, and share all relevant data. Indeed, this project will have to overcome numerous obstacles to information sharing, such as the fact that physicians in different countries often use entirely different words to describe the same disease.

Douakis-Agladze points out that the project could yield not only benefits for the emerging field of personalized medicine — another political priority for the NIH and the commission — but also faces the challenge of small populations of patients.

Regulatory agencies such as the US Food and Drug Administration and the European Medicines Agency refer on large, randomized and controlled clinical trials when deciding whether to approve new medications, and one of the aims of the consortium will be to develop alternative clinical trial methods for diseases that affect few people.

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Number of identified Genes causing Rare Diseases

Cumulative number of genes linked to rare diseases by year since 2010
New Orphan Drugs marketed since 2010 in the USA or Europe

- Monthly updated data are captured into a cumulative and cross-linked table

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- The total is reported into the IRDiRC counter
Expansion of the Consortium

Formally launched in 2012

Funding organizations from:
- Asia & Middle East
- Australia
- Europe
- North America

Present commitment exceeds $1B worldwide
Adoption of Principles applying to Research activities

- Sharing and collaborative work in RD research
- Scientific standards, requirements and regulations in RD research
- Participation by patients and / or their representatives in research

- Launch of ICHPT: International consortium of Human Phenotype Terminologies
  - Set of 2,300 terms which should be included in any terminology used to describe phenotypic features
  - Recommendation to use HPO and ORDO
  - (Human Phenome Ontology and Orphanet Rare Disease Ontology)

- Launch of IRDiRC recommended
  - to promote platforms, tools, guidelines
  - Contributing to IRDiRC goals
2015 Action Plan to boost Therapy Development

- **Patient-Centered Outcome Measures (PCOM)**
  - To improve quality of trials and allow assessment of the medical added-value of new therapies
    - Report on initiatives worldwide already available and items for action
    - Workshop in Paris on 30 November 2015
    - Review the documents / post recommendations

- **Small population clinical trials (SPCT)**
  - To agree with Regulators on acceptable alternative methods
    - Report on state of play of science and regulatory recommendations
    - Workshop in London at EMA, first trimester 2016
Selected Key Topics for 2015/2016

- **Matchmaker Exchange (MME)**
  - Facilitate matching of unsolved genome/exome sequence cases, based on similar phenotypics/genotypic profiles
  - Workshop in Baltimore on 6 October 2015

- **Machine readable consent (MRC)**
  - To access electronically patient consent to share data and improve research participation
  - Workshop in Paris on 9-10 November 2015

- **Data mining and repurposing (DMR)**
  - to identify new therapeutic targets and to repurpose drugs
  - Members identified / express your interest
  - Process not yet launched/ Workshop planned for T2 or T3 2016
All documents are accessible at www.irdirc.org
Thank you for your attention