PRESS EVENT INVITATION

Camp Nou, Barcelona, 24 January 2013 at 15:30

International experts will explain the latest cutting-edge approaches to medical research in the rare disease field at a press conference to celebrate the launch of 38 million EUR of funding for rare disease research and therapy development.

This is an opportunity to meet the scientists, clinicians, patient representatives and policymakers involved in driving the advances in rare disease research and find out about the key collaborations being established through a new global initiative. Leaders in the field will explain how research and next-generation technologies aim to improve diagnosis and develop new therapies for patients with rare diseases.

The press conference and Q&A will be followed by an exclusive tour of Camp Nou.

The International Rare Diseases Research Consortium (IRDiRC) was launched in April 2011 to foster international collaboration in rare disease research, a highly challenging area of medical research that has the potential to benefit tremendously from the recent advances in genomics, proteomics and other omics technologies. 80% of rare diseases have a genetic component, and despite their individual rarity, they are collectively so numerous that they affect as many as 1 person in every 17. IRDiRC has set itself the bold aims of delivering 200 new rare disease therapies and diagnosis for all rare diseases by the year 2020. Spearheaded by the European Union and the United States National Institutes of Health, the IRDiRC now numbers 29 members across the world.

Four of the European Union's flagship IRDiRC projects are being launched in Barcelona this January, providing 38 million EUR of funding for cutting-edge research and collaboration over the next six years. By increasing diagnosis rates, enabling global cooperation and putting new potential therapies into the drug development pipeline, these projects will help ensure tangible benefits for patients with these historically neglected conditions are seen by 2020.

We look forward to seeing you in Barcelona.

Paul Lasko
Scientific Director, CIHR Institute of Genetics and Chair-Elect, IRDiRC Executive Committee
Ruxandra Draghia-Akli
Director, DG Research Health Directorate, European Commission and Chair, IRDiRC Executive Committee
Ségolène Aymé
Coordinator of SUPPORT-IRDiRC
Emeritus Research Director, INSERM
Ivo Gut (event host)
RD-Connect data platform lead
Director, Centre Nacional d'Anàlisi Genòmica, Barcelona
Franz Schaefer
Coordinator of EURenOmics
Head, Division of Pediatric Nephrology, Heidelberg University Medical Center, Germany
Olaf Riess
Coordinator of Neuromics
Head, Institute of Human Genetics, University of Tübingen, Germany
Hanns Lochmüller
Coordinator of RD-Connect
Professor of Experimental Myology, Newcastle University, UK

This is a ticketed event and places are limited. Please indicate your interest in attending by email to Paramita Chakraborty (paramita.chakraborty@inserm.fr) by 21 January 2013, providing your full name, press affiliation and contact information. Further details will be provided to all registrants.

About the projects

EURenOmics (www.eurenomics.eu) focuses on **rare kidney disorders** and aims to identify novel genetic and epigenetic causes and modifiers of disease and their molecular pathways, develop innovative technologies allowing rapid diagnostic testing, discover and validate biomarkers of disease activity, prognosis and treatment responses, and develop in vitro and in vivo disease models to apply high-throughput drug candidate screening.

Neuromics (www.rd-neuromics.eu) addresses **rare neurodegenerative and neuromuscular disorders** and will use next generation whole-exome sequencing (WES) to increase the number of known gene loci, increase patient cohorts through large scale genotyping by enriched gene variant panels and next generation sequencing, develop biomarkers for clinical application with a strong emphasis on presymptomatic utility and cohort stratification, identify disease modifiers and develop targeted therapies using latest generation genetic approaches.

RD-Connect (www.rd-connect.eu) will develop a **global infrastructure** for sharing the research outputs of these and other rare disease projects, enabling scientists and clinicians worldwide to access a single centralized repository for omics data, phenotypic and biomaterial information. Every IRDiRC research project will be entitled to share its own data and access related data from other projects under policies agreed at a global level.

Support-IRDiRC (www.irdirc.org) provides the organisational support for the implementation of the International Rare Disease Research Consortium in close collaboration with the European Commission, the NIH and research funding agencies from participating countries, as well as with relevant research projects supporting IRDiRC objectives, especially EURenOmics, Neuromics and RD-Connect.

For further information contact:

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