

# DRUG REPURPOSING FOR RARE DISEASES

## THE CURE FOR THE 21ST CENTURY

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# DRUG REPURPOSING FOR RARE DISEASES THE CURE FOR THE 21ST CENTURY

November 17<sup>th</sup> and 18<sup>th</sup>, 2016

## WELCOME

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Dear Invited Speakers and Participants,

It is our pleasure to welcome you to the B·Debate “Drug Repurposing for Rare Diseases. The Cure for the 21st Century” . B·Debate, an initiative of Biocat and “La Caixa” Foundation, is organized by the Parc Científic de Barcelona (PCB), Universitat de Barcelona (UB), and Sant Joan de Déu Children’s Hospital (HSJD), with the collaboration of the Universitat Autònoma de Barcelona (UAB), Centro Nacional de Análisis Genómico (CNAG-CRG), Université de Montréal, International Rare Diseases Research Consortium (IRDiRC), and Centro de Investigación Biomédica en Red – Enfermedades Raras (CIBERER).

Rare diseases affect a reduced number of patients each, but there are around 7000 rare diseases, and therefore the percentage of the global population affected is nearly 6-8%. Most rare diseases have a genetic origin, and only 5% of them have an available treatment. The drug discovery and development process is long and expensive, and a possible strategy to make it more effective to find new therapies for patients with rare diseases is the repurposing of known drugs for other diseases.

This event has the main goal of generating a forum for an interdisciplinary and international debate, where scientists from the areas of chemistry, computation, biology and genomics, exchange ideas with clinical experts on rare diseases, and with representatives of agencies and entities involved in the development of therapies, on how repurposing of drugs may be optimized for the benefit of patients.

The four roundtables of this event address complementary aspects of the event’s focus theme: International approaches for coordinating and funding rare diseases research; case studies of drug repurposing and other approaches leading to clinical studies on new rare diseases therapies; data Integration, computational and experimental approaches for drug discovery on rare diseases; and challenges in the clinical development and the view of patients with rare diseases.

We want to generate an interdisciplinary discussion on rare diseases and drug repurposing research and on how to improve the number of approved therapies for patients, with the participation of leading investigators and international experts in the field. At the end of this B·Debate we would like to contribute to the scientific community and in the broader sense, to society, and the main conclusions of the B·Debate will be shared with representatives of rare diseases patients’ associations.

We encourage you to participate actively in all the discussions and thank you for joining us.

Yours sincerely,

Jordi Quintana and Francesc Palau (Scientific Leaders), Scientific Committee and B·Debate

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# PROGRAM

Thursday, November 17th, 2016

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**9:00 Welcome**

**Ignasi López**, Director, Science and Grants Department, la Caixa Foundation

**Albert Barberà**, CEO, Biocat

**Jordi Quintana**, Business Development, PCB

**Francesc Palau**, Director at Dept. of Genetic Medicine, Pediatric Institute for Rare Diseases, Sant Joan de Déu Children's Hospital

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**9:30 SESSION 1: INTERNATIONAL APPROACHES FOR NEW THERAPIES**

Chairs: **Jordi Quintana**, PCB, Barcelona, Spain

**International Rare Diseases Research Consortium (IRDiRC): Global Coordination to Stimulate Rare Disease Research Efforts**

**Noel Southall**, National Institutes of Health, Rockville, USA

**E-Rare: International funding of Clinical research for new therapeutic uses of already existing molecules (repurposing) in rare diseases**

**Julliane Halftermeyer**, French National Research Agency (ANR), Paris, France

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**10:30 Coffee Break**

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**11:00 Re-Engineering Repurposing and Re-Thinking Disease**

**Christine Colvis**, NIH/NCATS, Bethesda, MD, USA

**Innovative Medicines Initiative: A new model for public-private partnerships**

**Pierre Meullen**, Innovative Medicines Initiative (IMI), Brussels, Belgium

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**12:00 Open debate**

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**13:00 Lunch**

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**14:30 SESSION 2: CHALLENGES AND CASE STUDIES**

Chairs: **Jordi Surrallés**, UAB, Barcelona, Spain

**Systematic DRP<sub>x</sub> in rare diseases; an integrated approach**

**Raül Insa**, SOM Biotech, Barcelona, Spain

**Rare Purposing-Past Successes and Future Economics**

**Bruce Bloom**, Cures Within Reach, Skokie, USA

**Towards a Gene Therapy for Neurological and Somatic Mucopolysaccharidosis**

**Fàtima Bosch**, UAB, Barcelona, Spain

**Building a patient centric model for rare disease drug discovery**

**Tim Guilliams**, Healx, Cambridge, UK

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**16:30 Open debate**

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**17:30 End of sessions 1 and 2**

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# Friday, November 18th, 2016

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## 9:30 SESSION 3: DATA INTEGRATION, COMPUTATIONAL AND EXPERIMENTAL APPROACHES

Chair: **Ivo Gut**, CNAG, Barcelona, Spain

### **Polypharmacology and Drug Repurposing**

**Jordi Mestres**, IMIM, Barcelona, Spain

### **Repositioning FDA-approved drugs alone and in combination with epigenetic drugs to reprogram cancer epigenome**

**Noël Raynal**, Université de Montréal, Montreal, Canada

### **Repurposing analysis based on systems biology approach: Network-Centric Therapy for neurodegenerative diseases**

**Mireia Coma**, Anaxomics, Barcelona, Spain

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11:00 **Coffee break**

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### **11:30 RD-Connect: data sharing and analysis for rare disease research within the platform and through the Matchmaker Exchange and Beacon projects**

**Sergi Beltran**, CNAG, Barcelona, Spain

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12:00 **Open discussion**

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13:00 **Lunch**

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## 14:00 SESSION 4: FROM RESEARCH TO PATIENTS

Chair: **Francesc Palau**, HSJD, Barcelona, Spain

### **Drug Repurposing in Pediatric Patients**

**Ainhoa Andueza**, HSJD, Barcelona, Spain

### **Repurposed drugs for rare diseases - an attractive development option**

**Hermann A. M. Mucke**, Hermann Mucke Pharma Consultancy, Vienna, Austria

### **Repurposing the mechanism of action. Case study on X-ALD**

**Marc Martinell**, Minoryx, Barcelona, Spain

### **Patients' Perspective on Drug Repurposing in the Rare Disease Field**

**Virginie Hivert**, EURORDIS, Paris, France

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16:00 **Open discussion and closing of the meeting**

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16:30 **Coffee break**

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## 17:00 OPEN SESSION

### **Round table: Drug repurposing for rare diseases. From laboratory to patients**

Chair: **Jordi Cruz**, FEDER

**Marta Príncep**, RICHI Foundation

**Jordi Surrallés**, UAB

**Josep Torrent**, UAB / Eurordis

**Francesc Palau**, Hospital Sant Joan de Déu

**Raül Insa**, SOM Biotech

**Marc Martinell**, Minoryx

**Jaume Reventós**, Department of Health, Generalitat de Catalunya

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19:00 **End of the open session**

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# SCIENTIFIC COMMITTEE



**Francesc Palau**, Director of the Department of Genetic Medicine and the Pediatric Institute for Rare Diseases (IPER) at **Sant Joan de Déu Children's Hospital (HSJD)**, Barcelona, Spain.

Francesc Palau directs the Department of Genetic Medicine and the Pediatric Institute for Rare Diseases (IPER) of the Sant Joan de Déu Children's Hospital (HSJD) of Barcelona. He is visiting professor of Pediatrics at the University of Barcelona. In 1982 he earned MBChB graduate and MD degrees from the University of Valencia. He is a specialist in pediatrics formed at La Fe University Hospital in Valencia, where he was trained in human genetics. He obtained a PhD degree in 1989 at the University of Valencia and performed postdoctoral studies at St. Mary's Hospital Medical School in London. In 1991 he returned to the La Fe Hospital where he served until 2000 as a clinical geneticist. He has been research professor at the Institute of Biomedicine of Valencia, CSIC, and scientific director of the Center for Biomedical Network Research on Rare Diseases (CIBERER). Its scientific and medical interest is oriented towards the study of genetics, pathophysiological and therapeutics of neuromuscular and neurodevelopmental diseases. Among his scientific contributions it highlights the participation in the discovery of Friedreich ataxia and Charcot-Marie-Tooth disease genes by either positional cloning or whole exome studies. At HSJD he is currently implementing a comprehensive program addressed to ameliorate health care of children with rare and undiagnosed diseases based on patient-centered multidisciplinary clinics, clinical and functional genomics, and translational research.

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**Jordi Quintana**, Business Development at **Barcelona Science Park (PCB)**, Barcelona, Spain.

With a scientific training in organic chemistry (undergraduate) and biochemistry / structural biology (Ph.D.), and two postdoctoral stays in structural biology / drug design, Dr. Jordi Quintana developed a career in R+D in the pharmaceutical company Esteve for 14 years, contributing from drug design and discovery to the clinical development of two new chemical entities. Dr. Quintana also led the implementation of the R+D chemico-biological database of the company, as well as the Electronic Laboratory Notebook. After moving to Barcelona Science Park in Spain, Dr. Quintana built a new Drug Discovery Platform that has participated in 10 competitive research projects (leading 4 of them), and led the implementation of the Spanish Chemical Biology infrastructure, Chembiobank, representing Spain in the Steering Committee of the European Chemical Biology infrastructure, EU-OPENSREEN. Dr. Quintana was the coordinator of the research project Drugs4Rare, funded by IRDiRC through the ISCIII in Spain, to analyze Drug Repurposing for Rare Diseases.

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**Jordi Surrallés**, Professor of Genetics, **Universitat Autònoma de Barcelona (UAB)**, Barcelona, Spain.

With a PhD in Genetics and postdoctoral experience in The Netherlands (Leiden University Medical Center) and Finland (Finnish Institute of Occupational Health), he set up his research team at the UAB where he is currently Full Professor of Genetics. He is Director of the Biobank of DNA Repair Syndromes, Head of the Chromosome Fragility Laboratory Service and team leader at the Center for Biomedical Network Research on Rare Diseases, where he participates in the Area of Familial Cancer, Hematology and Dermatology. He has supervised over 25 research grants awarded from public and private institutions world-wide and participated in clinical trials, several patents and contracts with private foundations and biotech-pharma companies. He has given tens of invited lectures in international meetings. He has published over 100 articles and book chapters, supervised 25 PhD and Master degree students. He is reviewer of tens of scientific journals including Science, Nature, or Blood and Referee of national and international funding agencies including European Research Council, Agence Nationale de la Recherche, Cancer Research UK, Commission of the European Union, Dutch Cancer Society (The Netherlands), Fanconi Anemia Research Fund Inc. (USA), German-Israeli Foundation for Scientific Research and Development (Germany), INSERM (France) and the French National Cancer Institute (France). He was awarded with the Fanconi Anemia Discovery Award (Denver 2013, USA) and two ICREA-Academia Awards (2008 and 2014).

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**Ivo Gut**, Director of **Centro Nacional de Análisis Genómico (CNAG-CRG)**, Barcelona, Spain.

Dr. Gut is the Director of the Centro Nacional de Análisis Genómico (CNAG-CRG), one of the largest genome sequencing center in Europe. From 1999 to 2009 he established, at the Centre National de Génotypage (CNG), the highest throughput genotyping platform in Europe and executed many genome-wide association studies; he was the coordinator of the EU-funded Project READNA in which 2nd, 3rd and 4th generation nucleic acid analysis technologies were developed. He received his PhD in Physical Chemistry from the University of Basel in 1990. He is author of more than 300 research papers, 11 reviews and 12 book chapters, cited over 21 000 times, inventor of 25 patents or patent applications, founder of 4 biotech start-ups, and serves on numerous international advisory boards.

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**Sergi Beltran**, Bioinformatics Analysis Group Leader at **Centre Nacional d'Anàlisi Genòmica, CNAG-CRG**, Barcelona, Spain.

Dr. Sergi Beltran is the Bioinformatics Analysis Group Leader at the National Center of Genomic Analysis in Barcelona (CNAG-CRG) since 2012. He received his PhD in Biology from the Universitat de Barcelona (UB) in 2006. After working at the Transcriptomics Platform of the Barcelona Science Park and being a postdoctoral fellow at the Centre for Genomic Regulation (CRG), he became the Bioinformatics manager from UB's Scientific and Technological Centers. Dr. Beltran's group is devoted to the development and operation of sequencing data analysis and management tools and pipelines. The group collaborates with several national and international projects, mostly related to human health. Regarding rare disorders, he leads the RD-Connect platform development and is part of the ELIXIR rare disease use case, MatchMaker Exchange and the GA4GH's Beacon project.

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# DETAILED PROGRAM AND INVITED SPEAKERS

Thursday, November 17th, 2016

## Session 1: International approaches for new therapies

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**Jordi Quintana**, Business Development at **Parc Científic de Barcelona (PCB)**, Barcelona, Spain.

(See his CV at the Scientific Committee section)

Chair of the **SESSION 1**

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**Noel Southall**, Informatics, **National Institutes of Health**, Rockville, Maryland, USA.

Dr. Noel Southall co-chairs the task force on Data Mining and Repurposing of the International Rare Diseases Research Consortium (IRDiRC). For many rare diseases, drug repurposing is the only pragmatic strategy for having clinical impact. In his research, he has labored to develop the infrastructure, tools and libraries to facilitate repurposing and has gained experience prosecuting leads, transitioning to clinical proof-of-concept studies, and generating new molecular entities starting from existing drugs. After earning a PhD in Biophysics from the University of California, San Francisco, Dr. Southall began his career in industry as a computer-aided drug discovery scientist and later joined the National Institutes of Health in 2006. In the Informatics group at NIH/NCATS, he develops software, standards, and provides informatics analysis for therapeutic project teams, from interpreting high-throughput screening data to bioinformatics and structure-based modeling to biomarker discovery and development. These activities help move promising projects from screening toward the clinic. Therapeutic discovery and clinical development project teams are highly interdisciplinary, and informatics is often at the nexus of activity, helping to build software infrastructure to support analysis, modeling, and interpretation – working closely with each of the disciplines to provide support and help communicate those results to the rest of the team.

### **International Rare Diseases Research Consortium (IRDiRC): Global Coordination to Stimulate Rare Disease Research Efforts**

The International Rare Diseases Research Consortium (IRDiRC) provides global coordination and cooperation to stimulate and maximize output of rare disease research efforts. The consortium includes members from Europe, North America, Asia, Australia, Middle East and each funder supports its own research. Its initial focus is on developing common scientific and policy frameworks to facilitate the development of 200 new therapies for rare diseases by 2020 and the means to diagnose most rare diseases by 2020. IRDiRC has formed a task force on data mining and repurposing to understand how advances in data mining may lead to new repurposing opportunities. From funders' perspective, here is a valuable opportunity to align and prioritise research agenda on data mining. However, one of IRDiRC's goals is to deliver more therapies, therefore repurposing is a logical area to be studied, analysed and understood. At the same time, translation from data mining to repurposing is also a crucial aspect of development that deserves further attention.

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**Juliane Halftermeyer**, Scientific Officer at **French National Research Agency (ANR)**, Paris, France.

Dr Juliane Halftermeyer, scientific officer for transnational collaborations in the Biology & Health department, is the project manager of the ERA-Net E-Rare-3. She has been involved in E-rare since 2015. She earned her PhD in oncology-hematology from the University Paris Diderot (France) and she worked at the French Consulate General in Atlanta, as the deputy scientific attaché (USA). She then



joined the ANR in November 2013. She has been in charge of national and bilateral programmes and representing the ANR in several transnational collaborations.

### **E-Rare: International funding of Clinical research for new therapeutic uses of already existing molecules (repurposing) in rare diseases**

E-Rare, the ERA-Net for Research Programmes on Rare Diseases, was established in 2006. Only a few European countries fund research on rare diseases through specific dedicated programs. Therefore, the funding of transnational collaborative research is the most effective joint activity to enhance the cooperation between scientists working on the unmet medical needs of patients with rare diseases in Europe and beyond, both reducing fragmentation of research in this field and increasing research access to rare disease patients. The E-Rare consortium was built to link responsible funding organizations and ministries that combine the scarce resources for rare diseases research and thus enable the participation of many researchers to transnational projects via Joint Transnational Calls (JTCs).

In addition, E-Rare also identifies rare diseases research needs to implement specific funding opportunities based upon IRDiRC recommendations since 2012. In particular, the JTC2016 spotlights clinical trials for new therapeutic uses of already existing molecules (repurposing) in rare diseases. This is a precise example of how E-Rare can influence and strengthen the collaboration between national Ministries of Research and of Health to foster use of repurposed drugs as new therapies with clear benefit to rare disease patients.

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**Christine Colvis**, Director, Drug Development Partnership Programs, NCATS/NIH, Bethesda, MD, USA

Christine Colvis joined NCATS in June 2012 to lead the “NIH-Industry Pilot Program: Discovering New Therapeutic Uses for Existing Molecules” that tests a new model for public-private partnership collaborations including template agreements to shorten the time it takes to establish collaborations between an academic institution and a pharmaceutical company and move more rapidly into the actual research. Collaborations established between academic institutions and the company will test ideas for new therapeutic uses, with the ultimate goal of identifying promising new treatments for patients. A high level of interest in the program led to the availability of additional pharmaceutical company assets and NCATS funding opportunities including expansion of the program to support development for pediatric indications. Christine also oversees the NCATS Biomedical Data Translator program, which launched in 2016 with an initiative to assess feasibility and architectural design of a system that would enable new insights into the biology of disease in order to improve translation and deliver more treatments to more patients more quickly.

### **Re-Engineering Repurposing and Re-Thinking Disease**

Launched in May 2012 as part of the New Therapeutic Uses program, NCATS' NIH-Industry Partnerships initiative fosters collaboration between pharmaceutical companies and the biomedical research community to advance therapeutics development. The focus is on matching researchers with a selection of pharmaceutical assets to help the scientists test ideas for new therapeutic uses. Through this initiative, NCATS is re-engineering the research pipeline, using innovative strategies to identify new uses for existing therapies that have undergone significant research and development by industry, including safety testing in humans. Coordinating the access that scientists nationwide have to existing therapies that have already cleared several key steps in the development process can accelerate the pace of therapeutics development and get more treatments to more patients more quickly. Another program that will be discussed is the Biomedical Data Translator, the “Translator”. As a result of recent scientific advances there is a tremendous amount of biomedical research data and data from disease classifications, health records, clinical trials and adverse event reports that are available and could be useful for understanding health and disease and for developing and identifying treatments for diseases. Unfortunately, these very rich, but different, data sources exist in different locations, and often in different forms that are not compatible or interoperable with each other. Furthermore, clinicians and biologists think of disease in different ways, and speak different languages. Physicians diagnose and treat disease based on signs and symptoms affecting specific target organs. In contrast, biomedical researchers think of disease in terms of molecular changes in specific proteins, pathways or cell types. The Translator will integrate multiple types of existing data sources, including objective signs and symptoms of disease, drug effects, and intervening types of biological data relevant to understanding pathophysiology. The intent of developing a Biomedical Data Translator is to accelerate biomedical translation and to facilitate the generation of new hypotheses for understanding and treating disease.

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**Pierre Meulien**, Executive Director, **Innovative Medicines Initiative (IMI)**, Brussels, Belgium.

Pierre Meulien is Executive Director of the Innovative Medicines Initiative (IMI), a €5 billion public-private partnership between the European Union and the European pharmaceutical industry. At IMI, he is responsible for the overall management of the programme, which is working to improve and accelerate the entire medicines development process by facilitating collaboration between the key players involved in health research, including universities, pharmaceutical and other companies, patient organisations, and medicines regulators. Dr Meulien joined IMI in September 2015. From 2010 to 2015,

Dr Meulien was President and CEO of Genome Canada, where he raised significant funds for the organisation and oversaw the launch of novel projects and networks in the field of genomics-based technologies. Prior to that from 2007 to 2010, he was Chief Scientific Officer for Genome British Columbia. From 2002 to 2007, Dr Meulien served as founding CEO of the Dublin Molecular Medicine Centre (now Molecular Medicine Ireland), which linked medical schools and teaching hospitals in Dublin to build a critical mass in molecular medicine and translational research. Dr Meulien also worked in the private sector with the French biotechnology company Transgene, and with Aventis Pasteur (now Sanofi Pasteur). He has a PhD in molecular biology from the University of Edinburgh and carried out a post-doctoral fellowship at the Institut Pasteur in Paris. Read more at: <http://www.imi.europa.eu>

### **Innovative Medicines Initiative: A new model for public-private partnerships**

The Innovative Medicines Initiative (IMI) was launched in 2008 with the goal of significantly improving the efficiency and effectiveness of the drug development process in Europe, to make drug R&D processes in Europe more innovative and efficient, to enhance Europe's competitiveness and to address key societal challenges of health, demographic change and wellbeing. IMI's €2 billion budget for the period 2008-2013 made it the largest life sciences public private partnership in the world. IMI is now in its second phase of development and IMI2 expands the scope of IMI1 to cover the entire medical research and innovation value chain going from bottlenecks in industry to bottlenecks in industry and society, involving all relevant partners: pharmaceutical; other health-related industries; patients; academia; SMEs; regulators; HTA bodies; in open innovation networks. IMI 2 will run from 2014 to the end of 2024 and it will have a total budget of up to €3.276 billion, split with € 1.638 billion from the EU's Horizon 2020 programme, to match the contribution of the industrial partners, be it €1.425 billion from EFPIA and its member companies, or €213 million from other partners from the public and private space that decide to join IMI2 as associated partners at project or program level. The IMI platform could be used for public-private collaborative efforts to repurpose existing drugs for rare diseases.

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## **Session 2: Challenges and case studies**

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**Jordi Surrallés**, Professor of Genetics, **Universitat Autònoma de Barcelona (UAB)**, Barcelona, Spain.

(See his CV at the Scientific Committee section)

Chair of the **SESSION 2**



**Raül Insa**, CEO of **SOM Biotech**, Barcelona, Spain.

Raül Insa, MD, PhD, MBA, is the CEO and Founder of SOM Biotech, a start-up biopharmaceutical company established in the Barcelona Scientific Park and specialized in drug repositioning through an in-silico solution. Previously, he has been working for more than 20 years in multinational pharmaceutical companies, such as Parke-Davis (now Pfizer), UCB Pharma, Uriach Group (now Palau Pharma) and ISDIN (from Esteve Group). Raül received his Medical training and his PhD in Clinical Neurology at the University of Alicante, Spain; an MBA from ESADE Business School in Barcelona and an

Executive Education Program from IESE, Barcelona. He has also attended a couple of biotech leadership programs from Harvard Business School, Boston. His background includes more than 45 publications and papers, 7 chapters in books, 87 congress communications and 9 pharmaceutical patents.

## Systematic DRPx in rare diseases; an integrated approach

SOM Biotech, founded in late 2009 has done more than 28 repurposed (DRPx) programs in different indications, with two compounds arriving to phase 2a proof of concept. We own a virtual screening in-silico technology able to identify new activities of already know drugs plus complementary software and open eyes on the academia opportunities. We focus in rare diseases because of the great medical need and because by DRPx it is possible to reach fast the clinical proof of concept and the market sooner than with new chemical entities. We will present the case study of SOM0226, a repositioned product for the prevention and treatment of Transthyretin Amyloidosis (ATTR) which has jumped to Phase 2 proof of concepts in just two years after its discovery and is four times more potent/active than the most advanced new chemical entity (Tafamidis). Also, the case of SOM3355 for the treatment of chorea in Huntington patients will be discussed; a repurposed product with a better safety and compliance than the gold standard (Tetrabenazine). Pros and cons of our systematic and innovative approach to DRPx will be discussed.

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**Bruce Bloom**, President, Cures Within Reach, Skokie, USA.

Dr. Bruce Bloom is President and Chief Science Officer of Cures Within Reach, a non-profit corporation saving lives since 2005 by repurposing human approved drugs and devices to quickly deliver safe and affordable treatments and cures for diseases that have no currently effective therapy. Cures Within Reach uses CureAccelerator™, the only global online repurposing research collaboration platform, to bring together clinicians, researchers, funders, and industry to create and conduct pilot clinical trials that drive more treatments to more patients more quickly. Dr. Bloom is an Ashoka Fellow, recognized as a social entrepreneur for his system-changing solutions to one of the world's most urgent social problems, finding new treatments for unsolved diseases. Dr. Bloom is the Patient Advisory Board Chair for the Institute for Translational Medicine, Director of Scientific Affairs for Vision for Tomorrow, on the Science Advisory Boards of Rediscovery Life Sciences, the GARROD AKU Consortium, the Dr. Ralph and Marian Falk Medical Research Trust Awards Programs, the Findacure Social Impact Bond Initiative, and Healx, LTD, the Chair of the Governance Committee of the Kendall College Charitable Trust, a member of the Board of Councilors of Midwestern University, and a member of the editorial board of ASSAY and Drug Development Technologies. Dr. Bloom hosts the Clinician's Roundtable on ReachMD.com, and is a facilitator for Pathways to Successful Living.

## Rare Purposing-Past Successes and Future Economics

There are 7000+ unsolved diseases, and the vast majority are rare diseases. Cures Within Reach and our partner funders and researchers have successfully repurposed generic drugs and nutraceuticals that have created effective "new" treatments for rare diseases, including rare pediatric autoimmune diseases and rare pediatric neurological diseases. This presentation will describe how the repurposing process from genetic discovery to initial ideation, from mouse modeling to clinical trials, from results to publishing, from dissemination to clinical use. The presentation will also describe an ongoing effort to create a social finance economic engine for the repurposing of generic drugs for rare diseases.

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**Fàtima Bosch**, Full Professor of Biochemistry and Molecular Biology, Center of Animal Biotechnology and GeneTherapy- Universitat Autònoma Barcelona, Barcelona, Spain.

Fatima Bosch is a Pharmacist (1980) and PhD in Biochemistry (1985) by the University of Barcelona. She conducted post-doctoral studies at Vanderbilt University (1985), Case Western Reserve University (1988-1990), and NCI-Frederick Cancer Research and Development Center (1991). She is currently Full Professor of Biochemistry and Molecular Biology (1999) and Director of the Center of Animal Biotechnology and Gene Therapy (2003) at the Universitat Autònoma Barcelona. She has been granted the Rey Juan Carlos I (1985), Francisco Grande Covián (1998), Narcís Monturiol (2002), Sant Jordi Cross (2005), Alberto Sols (2006) and ICREA Academia (2013) awards. She has been Founding member of the European Society of Gene and Cell Therapy (1992), President of the Spanish Society of Gene and Cell Therapy (2007-2009), Vice-President of the European Association for the Study of Diabetes (2009-2012) and member of the Gene Doping Expert Group of the World Anti-Doping Agency (2013-present). Her research focuses on studying the pathophysiological causes of diabetes mellitus using transgenic animal models and developing gene therapy approaches to this disease by in vivo genetic manipulation of tissues using viral vectors. In recent years, she has applied her know-how on gene transfer technologies to the development of gene therapies for inherited metabolic disorders such as Mucopolysaccharidoses.

## Towards a Gene Therapy for Neurological and Somatic Mucopolysaccharidosis

Mucopolysaccharidosis Type II (MPSII), Hunter Syndrome, and Type III (MPSIII), Sanfilippo Syndrome, comprises 5 autosomic recessive disorders caused by mutations in genes that encode for enzymes involved in the stepwise degradation of glycosaminoglycans (GAGs). Accumulation of GAGs in lysosomes leads to lysosomal pathology, and affected patients

undergo severe neurodegeneration with mild somatic disease, and usually die during adolescence. There is no cure and MPS diseases constitute an unmet medical need. This presentation will discuss the potentiality of intracerebrospinal fluid AAV vector-mediated gene therapy to counteract neurologic and somatic MPS. Using this approach to treat for MPSII and MPSIII, expression of the different therapeutic genes was detected in widespread brain regions and in the liver, leading to increased enzyme activity in CNS and serum and simultaneous correction of both central and somatic disease. The results of this study provide strong evidence supporting the clinical translation of the approach.

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**Tim Guilliams**, Chief Executive at **Healx**, Cambridge, United Kingdom.

Tim is a social entrepreneur from the Cambridge Cluster (UK). He is passionate about delivering the next generation of therapeutics to rare disease patients in need. He is the Founder and Chief Executive of Healx Ltd and the Co-Founder and Trustee of the Cambridge Rare Disease Network (CRDN), a charity aiming to foster dialogue and awareness around rare diseases in Cambridge, UK. Healx focuses on helping Disease Foundations identify existing drugs that could treat their rare disease. The company was awarded 'Life Science Business of the Year' 2015 and 'Cambridge Graduate Business of the Year' 2016. Prior to Healx and CRDN, Tim worked on University-Industry interactions in the area of Life Sciences for the UK Government Department for Business, Innovation and Skills (BIS). His PhD was obtained at the University of Cambridge in the field of Biophysics and Neuroscience. Before moving to Cambridge, Tim obtained an MEng in Bio-Engineering from the University of Brussels (VUB).

#### **Building a patient centric model for rare disease drug discovery**

In this talk it will be discussed: the role of patient groups in drug development; the application of novel drug-matching techniques for drug repurposing and the emerging non-for-profit drug discovery models.

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## Friday, November 18<sup>th</sup>, 2016

### **Session 3: Data integration, computational and experimental approaches**

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**Ivo Gut**, Director of **Centro Nacional de Análisis Genómico (CNAG-CRG)**, Barcelona, Spain.

(See his CV at the Scientific Committee section)

Chair of the **SESSION 3**

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**Jordi Mestres**, Head of Research Group at **IMIM Hospital del Mar Medical Research Institute**, Barcelona, Spain.

Jordi Mestres holds a PhD in Computational Chemistry from the University of Girona. After a post-doctoral stay at Pharmacia&Upjohn in Kalamazoo (Michigan, USA), in 1997 he joined the Molecular Design & Informatics department at N.V. Organon in Oss (The Netherlands) and in 2000 he was appointed Head of Computational Medicinal Chemistry at Organon Laboratories in Newhouse (Scotland, UK). In 2003, he took on his current position as Head of the Research Group on Systems Pharmacology, within the GRIB Research Program at the IMIM Hospital del Mar Research Institute in Barcelona. He is also Associate Professor at the University Pompeu Fabra (UPF). In 2006, he founded Chemotargets as a spin-off company of his group. He is also the recipient of the 2006 Corwin Hansch Award from the QSAR and Modelling Society and the 2007 Technology Transfer Award from the UPF. His current research interests focus on the use and development of computational approaches to integrate chemical, biological, and phenotypic spaces and their application to systems chemical biology and drug discovery. He is the author of over 140 publications, 9 patents among them.

## Polypharmacology and Drug Repurposing

Repurposing drugs requires finding novel therapeutic indications compared to the ones for which they are/were already approved. This is an increasingly utilized approach to finding novel medicines, one that capitalizes on previous investments while derisking clinical activities. A strategy to identify those new therapeutical opportunities for drugs is to unveil new interactions with proteins. In this respect, the ability of small molecules to interact with multiple proteins is commonly referred to as polypharmacology. The now widely accepted polypharmacology of drugs opens an avenue to drug repurposing. Examples of the use of state-of-the-art computational methods to predict polypharmacology and how can this exploited for drug repurposing will be presented.



**Noël Raynal**, Assistant Professor at **Université de Montréal**, Montreal, Canada.

Bachelor in Biochemistry, Université de Montpellier (2000)  
MSc in Biology, Université du Québec à Montréal (2001-2003)  
PhD in Bbiology, INRS-Institut Armand-Frappier, Université du Québec (2003-2008)  
Postdoctorat (2009-2013) MD Anderson Cancer Center, Department of Leukemia, University of Texas & Fels Institute for Cancer Research and Molecular Biology, Temple University

### **Repositioning FDA-approved drugs alone and in combination with epigenetic drugs to reprogram cancer epigenome**

Epigenetic drugs, such as DNA methylation inhibitors (DNMTi) or histone deacetylase inhibitors (HDACi), are approved in monotherapy for cancer treatment. These drugs reprogram gene expression profiles, reactivate tumor suppressor genes (TSG) producing cancer cell differentiation and apoptosis. Epigenetic drugs have been shown to synergize with other epigenetic drugs or various anticancer drugs. To discover new molecular entities that enhance epigenetic therapy, we performed a high-throughput screening using FDA-approved libraries in combination with DNMTi or HDACi. We discovered that 45 FDA-approved drugs enhanced DNMTi and HDACi activity, mainly belonging to anticancer and antiarrhythmic drug classes. Transcriptome analysis revealed that combination of decitabine (DNMTi) with the antiarrhythmic proscillaridin A, produced profound gene expression reprogramming which was associated with down-regulation of 153 epigenetic regulators, including 2 known oncogenes in colon cancer (SYMD3 and KDM8). Also, we identified about 85 FDA-approved drugs that antagonized DNMTi and HDACi activity through cytotoxic mechanisms, suggesting detrimental drug interactions for patients undergoing epigenetic therapy. Overall, our drug screening identified new combinations of epigenetic and FDA-approved drugs, which can be rapidly implemented into clinical trials.



**Mireia Coma**, Business Unit Manager & Principal Investigator at **Anaxomics Biotech**, Barcelona, Spain.

Mireia Coma, MSc, PhD. Business Unit Manager & Principal Investigator at Anaxomics. Degree in Biochemistry, MSc in Experimental Biochemistry and Ph.D. in Health and Life Sciences. Previously she was a research Fellow at the Memory Unit of Hospital de la Santa Creu i Sant Pau, Barcelona, and at MassGeneral Institute for Neurodegenerative Disease, Massachusetts General Hospital - Harvard Medical School, Boston, MA, USA. In 2010 joined Anaxomics where she is currently on the front of the business unit. She has a wide experience in preclinical testing for neurodegenerative diseases and quantifiable success in achieving revenue, profit, and business growth objectives. As a part of her task in Anaxomics, she is in charge of drug repurposing projects.

### **Repurposing analysis based on systems biology approach: Network-Centric Therapy for neurodegenerative diseases**

Due to the multifactorial nature of diseases, multi-target polypharmacological research is needed to interact with different targets and modify different molecular pathways. Given the number and the intricacy of the factors involved in the initiation and the progression of complex diseases such as neurodegenerative diseases, tools that are capable of dealing with such complex scenario are required. Systems biology has recently emerged as new discipline that addresses this pressing need by considering living organisms as networks of interacting genes, proteins and biochemical reactions. Since they cope with the complexity inherent to human physiology, systems biology tools are particularly suited for the identification of multi-targeted agents and drug synergistic effects. On these grounds, Anaxomics implements Systems Biology through TPMS, a top-down approach which mathematically models human physiology by integrating the known information about the functional interactome, with experimental data provided by researchers in the area, and with a focus on clinical translation. By using a repurposing analysis based on systems biology, we identified and validated the neuroprotective potential of several new drugs combinations to treat neurodegenerative diseases.



**Sergi Beltran**, Bioinformatics Analysis Group Leader at **Centre Nacional d'Anàlisi Genòmica, CNAG-CRG**, Barcelona, Spain.

(See his CV at the Scientific Committee Section)

### **RD-Connect: data sharing and analysis for rare disease research within the platform and through the Matchmaker Exchange and Beacon projects.**

RD-Connect (rd-connect.eu) is a platform connecting omics data, registries, biobanks and clinical bioinformatics for rare disease research (platform.rd-connect.eu). RD-Connect aims to contribute to the IRDiRC objectives for 2020 and to narrow the gaps in rare disease research, where patient populations, clinical expertise and research communities are small in number and highly fragmented. The RD-Connect platform securely integrates -omics data with biosample and clinical information, providing not only a centralised data repository but also a sophisticated and user-friendly online analysis system. Raw Whole Genome, Exome or gene panel NGS data are deposited at the European Genome-phenome archive and processed with RD-Connect's standardised analysis and annotation pipeline. The corresponding clinical information is recorded in a connected PhenoTips instance using the Human Phenotype Ontology, OMIM and Orpha codes. The results are made available to authorised users through the highly configurable and efficient platform, which runs on a Hadoop cluster and uses Elasticsearch. To share data beyond the platform, RD-Connect has lit a GA4GH Beacon within the Beacon Network (beacon-network.org) and is actively involved in the development of the GA4GH/IRDiRC Matchmaker Exchange (matchmakerechange.org, MME) API.

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## **Session 4: From research to patients**

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**Francesc Palau**, Director of the Department of Genetic Medicine and the Pediatric Institute for Rare Diseases (IPER) at **Sant Joan de Déu Children's Hospital (HSJD)**, Barcelona, Spain.

(See his CV at the Scientific Committee Section)

Chair of the **SESSION 4**

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**Ainhoa Andueza**, Project Manager at **Sant Joan de Déu Children's Hospital (HSJD)**, Barcelona, Spain.

Project Manager at the Clinical Research Unit Sant Joan de Déu Children's Hospital (HSJD). With a strong background as a CRA at Quintiles Iberia SAU and as Project Manager, on assignment as Pfizer Clinical Operations Lead in Specialty Area, where she was the Responsible for the Implementation and management of phase II, III and IV clinical trials in Spain, and conducted the Feasibility process for upcoming projects and programs. In addition, she was a Project Leader, on assignment as Pfizer Clinical Operations Lead in Specialty Area. After being the Clinical Team Manager at PRA International, leading and managing the clinical team, she joined the Clinical Research Unit at HSJD, where she is the responsible for study management, evaluation of feasibility and advice and training in internal SOPs to ensure the quality of the projects.

### **Drug Repurposing in Pediatric Patients**

Traditionally, most medications used in pediatric patients are only approved by Regulatory Agencies for adult use. That is, they are used off-label. Furthermore, research in rare disease have also been little. In terms of pediatric population, there was a change in ethic and social paradigma, and children passed to be protected through clinical trials (European Regulation- PIPs). However, they are still some limitations as pediatric rare disease might be out of the scope of the regulation. HSJD act as a bridge from research to patient collecting information from families, investigators, and getting agreements with drug repositioning companies



**Hermann A. M. Mucke**, CEO, H.M. Pharma Consultancy, Vienna, Austria.

Dr. Hermann A.M. Mucke, who was born in 1955 and holds a Ph.D. in biochemistry from the University of Vienna (Austria), spent 17 years in academia and in various positions in the pharmaceutical industry before the year 2000 when he founded H.M. Pharma Consultancy ([www.hmpharmacon.com](http://www.hmpharmacon.com)), an Austrian-based enterprise providing strategic advice and tailored services to the life science industry with a strong focus on all aspects of drug repurposing. Dr. Mucke is an accomplished scientist, science author, patent analyst and R&D manager who had a strong role in the repurposing of several drugs during the past 15 years. He has founded, and is managing, LinkedIn discussion group on drug repurposing with 500+ members. Dr. Mucke is the editor of the Drug Repurposing Special Issues of ASSAY and Drug Development Technologies; an editorial board member and regular author of several scientific journals, such as the Pharmaceutical Patent Analyst, and serves on several corporate scientific advisory boards. Among many other market and technology analyses, Dr. Mucke is the author of “Drug Repositioning: Extracting added value from prior R&D investments”, published by Insight Pharma Reports.

### **Repurposed drugs for rare diseases – an attractive development option**

Developing known active ingredients for a rare disease for which no approved treatment is available can offer perspectives that are exciting, from both the medical and the commercial angle. The path from the laboratory to the patient can be quicker, and in any case the regulatory and exclusivity incentives are considerable. In principle, the synergy of these factors can overcompensate the drawbacks of the small market; however, drug repurposing for rare diseases also has some unique challenges which do not end with regulatory approval. Starting with recent examples of international patent applications claiming known drugs for new applications in rare neurological diseases, the possible development pathways and their timelines are outlined, and the associated problems are pointed out.



**Marc Martinell**, CEO, Minoryx Therapeutics S.L., Mataró, Spain.

Marc obtained a PhD in Chemistry from the University of Barcelona. He has broad experience in drug discovery and biotechnology through his participation at companies such as Crystax Pharmaceuticals and Oryzon Genomics where he managed several research projects and led the team in charge of target selection, structural biology, computational chemistry and hit ID through a fragment-based approach. At Oryzon, Marc actively contributed to the identification of the first-in-class inhibitors for the epigenetic target LSD1 currently in clinical studies and licensed to Roche. In 2011 Marc co-founded Minoryx Therapeutics, a clinical stage biotech company focused on the development of new drugs for a group of rare diseases known as Inborn Errors of Metabolism. Marc is co-author of several patents and publications.

### **Repurposing the mechanism of action. Case study on X-ALD**

When considering a repurposing-based approach there are many challenges that need to be overcome. One of the most relevant is that in many occasions the particular molecule may not be the better suited for the new disease to be treated. In this regard, drug repurposing may be used as a first step to identify useful mechanisms of action but additional work is required to identify the right molecule. Through the evaluation of existing drugs into phenotypic models of rare diseases, we generate knowledge about relevant targets, pathways and chemical structures that may be used as starting points for a drug discovery campaign. The ongoing program in X-linked Adrenoleukodystrophy will be also presented.

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**Virginie Hivert**, Therapeutic Development Director, **EURORDIS - Rare Diseases Europe**, Paris, France

Virginie Hivert joined EURORDIS in 2014 as Therapeutic Development Director. Virginie is responsible for following the development of orphan drugs as an observer on the Committee for Orphan Medicinal Products at the European Medicines Agency. She coordinates the group of high-level EURORDIS representatives/volunteers who sit on the various scientific committees/working parties at the EMA, known as the Therapeutic Action Group (TAG). She is responsible for two activity areas in EURORDIS, one being the training of patients' representatives in therapeutic development activities (EURORDIS Summer School, EUPATI) and the other related to their engagement in these activities (in Protocol Assistance in Scientific Advice Working Party (SAWP) at the EMA for example) She is a member of the Therapies Scientific Committee of IRDiRC (International Rare Disease Research Consortium). Prior to joining EURORDIS, Virginie worked for Orphanet as coordinator of data collection of the resources related to rare diseases (such as expert centers, medical laboratories, patient organisations, research projects, clinical trials, etc.) in the 37 countries of the Orphanet Consortium. Virginie holds a PharmD and a PhD in Biological Sciences and has previously worked in basic research, particularly on pathophys

### **Patients' Perspective on Drug Repurposing in the Rare Disease Field**

Theoretically, repurposing/repositioning of medicinal products/active substances from one medical condition to another should be considered as a positive thing from a patient perspective. Building on the existing knowledge, including pharmacologic characterization of a product, data on its safety profile or mode of action, is likely to bring advantage to the therapeutic development process, and it is of particular relevance in the field of rare diseases where the patient population is small and the available data are usually scarce. In general, patient organizations involved in the research field are supporting or advocating for such approaches. However, when it comes to actual benefit for patients, the experience over the past years has shown that a repurposed drug is not necessarily synonym of dramatic decrease of the commercialization price. In some cases, we could even observe a detrimental effect on patient access to treatment. From patients' perspective, it is therefore crucial to encompass these different aspects by looking at the full picture and to find ways to bring more treatment opportunities to patients at a reasonable price.

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## **Round table: Drug repurposing for rare diseases. From laboratory to patients**

### **Taula rodona: Reutilitzant medicaments per a malalties rares. Del laboratori als pacients**

Open session, held in Catalan



**Jordi Cruz**, Director of **Spanish MPS Society** and Delegate of **Rare Disease Federation in Catalonia, FEDER**, Barcelona, Spain.

Delegate of the Spanish Federation for Rare Diseases (FEDER) in Catalonia, member of the Board of Directors of FEDER Spain and Head of Training and Research of FEDER and FEDER Foundation for Research.

Father affected by Sanfilippo Syndrome type A (Mucopolysaccharidosis III A), and Founder and Director of the Sanfilippo Spanish Association in 2003, MPS Spain in 2005 and MPS-Lisosomales in 2015. In addition, he is the Director of Protocol and Institutional Relations at the University of Barcelona, Technical specialist in Computer Management and Patient Expert in different national and international committees and advisor of a variety of biotechnological companies and patient associations. He is a member and advisor of the Ibero-American Society of Medicine, as well as the Committee of Clinical Genetics of the Hospital Germans Trias i Pujol of Badalona (Barcelona). Member of EUCERD, the Commission of the IPT's of the Spanish Medicines Agency and the UEC's (Unitats d'Expertesa Clínica) Commission of the Department of Health of the Generalitat de Catalunya.

Moderator





**Marta Príncipe**, Technological Director & CEO, **Bellavista & Richi Research**, Barcelona, Spain.

Marta Príncipe PhD, holds 20 years of done-it background in management positions at the biomedical sector. She brings a long track record in drug discovery and development as Head of CNS dept at Ferrer Grupo creating new business and projects that led to several patent families, compounds in clinical phase and where she was member of the steering committee of international joint-ventures. Lately she performed as Innovation director and steering committee member at Biocat, the biocluster organization at Catalonia, where she was the scientific director of the Catalan business plan competition for start-ups, member of the Healthcare Investment Forum and director of the reference report on biosciences in Catalonia, fostering two public-private networks on oncology and nanomedicine among other activities. Nowadays she is the Technological Dpt director at Bellavista, a consultancy firm, in which she foster entrepreneurship and innovation, being board member in different start-ups and Strategic Advisor of Richi Research & CEO of the Biomarker and Drugs company created by Richi Foundation. Marta holds an industrial PhD in Biology and executive education from Wharton School in Pennsylvania and AMP from Esade Executive in Barcelona.

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**Jordi Surrallés**, Professor of Genetics, **Universitat Autònoma de Barcelona (UAB)**, Barcelona, Spain.

(See his CV at the Scientific Committee section)

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**Josep Torrent**, Professor of Clinical Pharmacology and Therapeutics at **Universitat Autònoma de Barcelona**, and Director General of the **Fundació Doctor Robert, Advanced Centre of Services and Training for Health and Life Sciences**, Barcelona, Spain.

Josep Torrent-Farnell is a qualified Pharmacist with a degree in Medicine and Surgery from the University of Barcelona as well as postgraduate courses in Pharmacology and Toxicology, Public Health and European Institutions. Josep is a specialist in Internal Medicine and Clinical Pharmacology and obtained his doctorate in Clinical Pharmacology from the Autonomous University of Barcelona (UAB). He became a member of the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency in 2000, serving as Chair for 2 mandates. Currently, he is also a member of the Scientific Advice Working Party (SAWP) at EMA. Josep is Professor of Clinical Pharmacology and Therapeutics at the Autonomous University of Barcelona (Universitat Autònoma de Barcelona) and Director General of the Fundació Doctor Robert, Advanced Centre of Services and Training for Health and Life Sciences.

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**Francesc Palau**, Director of the Department of Genetic Medicine and the Pediatric Institute for Rare Diseases (IPER) at **Sant Joan de Déu Children's Hospital (HSJD)**, Barcelona, Spain.

(See his CV at the Scientific Committee Section)

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**Raül Insa**, CEO of **SOM Biotech**, Barcelona, Spain.

(See his CV at the Session 2)

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**Marc Martinell**, CEO, **Minoryx Therapeutics S.L.**, Mataró, Spain.

(See his CV at the Session 4)



**Jaume Reventós**, Director of **Bellvitge Biomedical Research Institute – IDIBELL** and Head Operations and Institutional Relations at the Department of Health, **Generalitat de Catalunya**, Barcelona, Spain.

Jaume Reventós is a Doctor in Medicine (MD) by the Autonomous University of Barcelona (UAB). He attended and received his specialty of Biochemistry in the MIR at the Vall d'Hebron Hospital in Barcelona. Dr. Reventós was Head of the Biomedical Research and Translational Oncology, Vall d'Hebron Research Institute (VHIR) until 2014, where he became the Director of Bellvitge Biomedical Research Institute – IDIBELL. In addition, he is the Head Operations and Institutional Relations at the Department of Health, Generalitat de Catalunya.

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# PRACTICAL INFORMATION

## Venue: CosmoCaixa Barcelona

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**CosmoCaixa Barcelona**  
C/ Isaac Newton, 26  
08022 Barcelona, Spain

**Conferences**  
Agora room on -2 floor

## Contact persons during the event

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### **Marta Soler**

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[www.bdebate.org](http://www.bdebate.org) | [www.biocat.cat](http://www.biocat.cat)

# SUGGESTED READING

*In silico methods for drug repurposing and pharmacology.* Hodos RA<sup>1,2</sup>, Kidd BA<sup>1</sup>, Shameer K<sup>1</sup>, Readhead BP<sup>1</sup>, Dudley JT<sup>1</sup>. Wiley Interdiscip Rev Syst Biol Med. 2016 May;8(3):186-210. doi: 10.1002/wsbm.1337.

*Computational and Practical Aspects of Drug Repositioning.* Oprea TI<sup>1</sup>, Overington JP<sup>2</sup>. Assay Drug Dev Technol. 2015 Jul-Aug;13(6):299-306. doi: 10.1089/adt.2015.29011.tiodrrr.

*A survey of current trends in computational drug repositioning.* Li J, Zheng S, Chen B, Butte AJ, Swamidass SJ, Lu Z. Brief Bioinform. 2016 Jan;17(1):2-12. doi: 10.1093/bib/bbv020. Epub 2015 Mar 31.

*In vitro screening for drug repositioning.* Wilkinson GF<sup>1</sup>, Pritchard K<sup>2</sup>. J Biomol Screen. 2015 Feb;20(2):167-79. doi: 10.1177/1087057114563024. Epub 2014 Dec 19.

*Toward better drug repositioning: prioritizing and integrating existing methods into efficient pipelines.* Jin G<sup>1</sup>, Wong ST<sup>2</sup>. Drug Discov Today. 2014 May;19(5):637-44. doi: 10.1016/j.drudis.2013.11.005. Epub 2013 Nov 14.

*Open-source approaches for the repurposing of existing or failed candidate drugs: learning from and applying the lessons across diseases.* Allarakhia M<sup>1</sup>. Drug Des Devel Ther. 2013 Aug 8;7:753-66. doi: 10.2147/DDDT.S46289. eCollection 2013.

## **Institutional reports:**

*Breakthrough Business Models: Drug Development for Rare and Neglected Diseases and Individualized Therapies: Workshop Summary.* Editors: Institute of Medicine (US) Forum on Drug Discovery, Development, and Translation. Source: Washington (DC): National Academies Press (US); 2009.

*The National Academies Collection: Reports funded by National Institutes of Health.* National Academy of Sciences (US). Washington (DC): National Academies Press (US); 1995

*State of the art of rare disease activities in Spain.* This work was financed by the EUCERD Joint Action: Working for Rare Diseases N° 2011 22 01

*Report on the state of the art of rare disease activities in Europe – 2014 edition.* European Union Committee of Experts on rare Diseases.

## **Rare diseases in numbers:**

Preliminary report from an ongoing bibliographic study initiated by Eurordis in partnership with Orphanet [http://ec.europa.eu/health/archive/ph\\_threats/non\\_com/docs/rdnumbers.pdf](http://ec.europa.eu/health/archive/ph_threats/non_com/docs/rdnumbers.pdf)

## **Databases on rare diseases:**

Orphanet: rare diseases and orphan drugs

 <http://www.orpha.net/consor/cgi-bin/index.php>

National Center for Advancing Translational Sciences (NCATS)  
<https://ncats.nih.gov/trnd>

Therapeutics for Rare and Neglected Diseases (TRND)

 Genetic and Rare Diseases Information Center <https://rarediseases.info.nih.gov/>

# OUTCOMES

## B·Debateca

On the website of B·Debate, you will find all the information related with the celebration of the meeting that includes reports, conclusions, scientific documents, interviews with the experts, speaker's CVs, videos, images, press documentation and other related materials. We invite you to visit the section B·Debateca on [www.bdebate.org](http://www.bdebate.org)

Contents of the meeting **“Drug Repurposing for Rare Diseases. The Cure for the 21st Century”**

The screenshot shows the B-DEBATE website interface. At the top left is the logo for B-DEBATE, International Center for Scientific Debate, BARCELONA. To the right are logos for biocat and Obra Social 'la Caixa'. Below the logos is a navigation menu with the following items: INICI, B-DEBATE, CONVOCATÒRIA, DEBATECA (circled in red), NOTÍCIES, SINOPSIS, PREMSA, and CONTACTE. A search bar is located to the right of the menu. The main content area features three event cards: 1. 'B-DEBATE · OPEN REGISTRATION' for 'Drug Repurposing for Rare Diseases. The Cure for the 21st Century' from 17/11/2016 to 18/11/2016 (circled in red). 2. 'B-DEBATE · SAVE THE DATE' for 'Artificial Intelligence. Next Step in Evolution?' from 24/01/2017 to 25/01/2017. 3. 'B-DEBATE · SAVE THE DATE' for 'Industrial Biotechnology. Questions Today for Tomorrow's Needs' from 14/02/2017 to 15/02/2017. Below these are sections for 'PROGRAMA 2016' (Programa d'activitats de B-Debate del 2016), 'SINOPSI' (La ciència de les plantes: cap a una indústria verda), and 'CONVOCATÒRIA B-DEBATE' (La convocatòria de presentació de propostes per al programa d'activitats de 2017 ja ha tancat) with a 'LLEGEIX MÉS' button.

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# ORGANIZERS



International Center  
for Scientific Debate  
BARCELONA



**B-Debate** International Center for Scientific Debate Barcelona is a **Biocat** initiative with support from “**la Caixa**” **Foundation**. It drives first-rate international scientific debates, to foster dialogue, collaboration and open exchange of knowledge with prestigious national and international experts, to approach complex challenges of high social interest in life sciences. B-Debate sees debate as a powerful, effective way to generate knowledge and strives to help position Barcelona as a benchmark in generating knowledge and Catalonia as a country of scientific excellence.

B-Debate sees debate as a powerful, effective way to generate new knowledge. The debates are top-notch international scientific meetings featuring a selection of experts of renowned international prestige and scientists that work in Barcelona and Catalonia, moderated by scientific leaders. Since 2009 B-Debate has invited about 1200 recognized speakers and over 7.000 attendees. B-Debate seeks out answers to the challenges and needs of society in the field of life sciences, taking into account the complex, ever-changing conditions of this global world. The debates foster the integration of different disciplines of science and deal with such diverse topics as ageing, new therapeutic approaches to various diseases, innovative technology to improve knowledge of the human genome, food resources, new tools to integrate knowledge management, clinical genomics, neurosciences, climate change, and new energy sources, among others. The knowledge and results obtained through these events is spread throughout both the scientific community and general society through the various **B-Debate** channels and instruments.

More info: [www.bdebate.org](http://www.bdebate.org)

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Parc Científic de Barcelona  
UNIVERSITAT DE BARCELONA

The **Parc Científic de Barcelona (PCB)** offers institutions and companies an ideal environment dedicated to the generation, transfer and addition of value in the field of the life sciences. A cluster with a wide range of scientific and technological support for research. Located in the Barcelona Knowledge Campus, the PCB provides regular basic services and an excellent environment for networking. More than 2,000 highly qualified researchers work at the PCB. The aim of the Park is to bring together public and private research environments, facilitating the transfer of knowledge and innovation between the University and research environment and the business world.

Created by the University of Barcelona in 1997, the PCB was the first Science Park in Spain and today we are an international benchmark for the promotion of innovation with more than 2,000 professionals. The Park is currently home to three research institutes and about 70 companies and other organisations. We offer furnished and non-furnished laboratories, as well as offices on a rental basis and a range of associated services. At the PCB we also organise activities promoting scientific culture and encouraging new scientific vocations in which nearly 4,500 people participate every year.

More info: [www.pcb.ub.edu/portal/en](http://www.pcb.ub.edu/portal/en)

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Sant Joan de Déu  
Barcelona · Hospital

**Hospital Sant Joan de Déu** is a teaching hospital specializing in the fields of pediatrics, gynecology and obstetrics. It is located in Barcelona, Catalonia (Spain). It is a privately owned center, concerted by the Catalan Public Health Service, which belongs to the Hospitaller Order of St. John of God, religious organization that manages more than 300 health centers over the world. The center has 362 beds and 12 operating rooms. It employs more than 1.500 professionals and attends every year more than 3.000 births, 130.000 emergencies, records 26.000 hospitalizations and 13.000 surgeries are performed. Hospital Sant Joan de Déu is the largest children's hospital in Spain, and one of the top 5 in Europe, along with Great Ormond Street (London), Hospital Necker Enfants Malades (Paris) and Ospedale Meyer (Florence).

More info: <https://www.sjdhospitalbarcelona.org>

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# COLLABORATORS



The **University Drug Research Group** was established in 2004 by researchers at the Université de Montréal in order to develop and support trans-disciplinary research, researcher training and technology transfer relating to drug discovery, development and evaluation. It is a hub for developing and increasing collaborative undertakings and exchanges between scientists exploring all aspects of drug discovery and development, including evaluation of how drugs affect population health.

More info: [www.grum.ca](http://www.grum.ca)

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Founded in 1878, **Université de Montréal** is the largest university in Quebec and the second largest in Canada, with over 67.000 students, including nearly 8.000 from around the world, and more than 11.000 diplomas awarded at every university level each year. Deeply rooted in Montreal and dedicated to its international mission, the Université de Montréal is one of the top universities in the French-speaking world and is ranked 92nd on the list of top international institutions by the prestigious QS World University Rankings.

More info: <http://www.umontreal.ca/english/index.html>

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A department central to effective international action planned and carried out to serve Quebecers according to sustainable development principles. To promote and defend Québec's interests internationally while ensuring respect for its authority and the consistency of government activities. **The Ministère des Relations internationales et de la Francophonie** carries out its mission through: Its knowledge of the international context, the policy advice it provides for the government, its coordination of international government activities, the ties it builds with foreign governments and international organizations.

More info: <http://www.mrif.gouv.qc.ca/en/>

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The Universitat Autònoma de Barcelona (UAB) is known for its excellence in research and quality in teaching, and is a reference centre in Europe. It offers 81 bachelor's degrees, covering all areas of knowledge, promotes innovation, employability and entrepreneurship and strives to foster research and transfer working closely with the surrounding business and industrial community. The UAB leads the Spanish universities on number of PhD programmes, providing advanced training in research techniques and culminating in a doctoral thesis. In recent years, the UAB has seen recognition for its efforts in promoting quality in research, with a steady improvement in its positions in the most prestigious and influential international rankings. In the 2016-2017 edition of the Times Higher Education World University Rankings, the UAB is the best Spanish university, occupying position 163 in the world ranking.

More info: <http://www.uab.cat/>

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centre nacional d'anàlisi genòmica  
centro nacional de análisis genómico

The **Centro Nacional de Análisis Genómico (CNAG-CRG)** was created in 2009 and since 2015 it is integrated with the Center for Genomic Regulation (CRG). Its mission is to carry out projects in DNA sequencing and analysis in collaboration with researchers from Catalonia, Spain and from the international research community in order to ensure the competitiveness of our country in the fields of Biomedicine, Biology and the Agrofood sector. The CNAG-CRG is directed by Dr. Ivo Gut and has a headcount of 70 staff, organized in two production-oriented groups for Sequencing and Bioinformatics Analysis, and 3 research groups focused on Bioinformatics Tools Development, Applied Genomics and Genome Biology. CNAG-CRG researchers participate in large national and international collaborative research projects in areas as diverse as cancer genetics, rare disorders, and evolutionary studies, such as FP7 and H2020 projects BBMRI-LPC, RDCONNECT, IBDCHARACTER, BCAST and MuG, and in the 4DGENOME project, awarded with an ERC Synergy Grant. CNAG-CRG has currently a park of twelve 2nd generation DNA sequencers (one HiSeq4000, eight HiSeq2000, two HiSeq2500 and one MiSeq) that produce more than 1000 Gbases of sequencing data per day, the equivalent of completely sequencing six human genomes every 24 hours.

More info: [www.cnag.crg.eu](http://www.cnag.crg.eu)

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The **International Rare Diseases Research Consortium (IRDiRC)** brings together research collaborations and investments in the field of rare diseases (RD). It aims to contribute to the development of 200 new therapies and the means to diagnose most RDs by the year 2020. It also emphasizes the importance of data and resource sharing. To advance these objectives, IRDiRC set up Task Forces to team up experts and stakeholders of specific research areas, selected as prioritized topics by the Consortium Assembly, to put forward their recommendations and help push for policy change, including the fields of data mining and repurposing, patient-centered outcome measures, and small population studies.

More info: <http://www.irdirc.org/>

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The CIBERER is the Spanish network cooperative consortium on rare diseases research funded by the Instituto de Salud Carlos III, with 62 research groups located at the main research centres all over Spain. Collaboration and cooperation between biomedical and clinical research groups are prioritised and furthered with special emphasis is placed on aspects of genetic, molecular, biochemical and cell research of rare, genetic or acquired diseases.

More info: <http://www.ciberer.es/>

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**CosmoCaixa** offers interactive, enjoyable science and an open door for anyone who is eager to learn and understand and who never stops wondering why things are the way they are. **CosmoCaixa Barcelona** boasts the Geological Wall and the Amazon Flooded Forest, which features more than 100 plant and animal species that convince visitors they have been transported from the Mediterranean to the very heart of the tropical jungle. In addition to its permanent facilities and its open areas, CosmoCaixa offers a scientific and educational programme that includes exhibitions, workshops, conferences, courses and debates involving experts from all over the world.

More info: [www.obrasocial.lacaixa.es](http://www.obrasocial.lacaixa.es)

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