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Second RE(ACT) Congress and IRDiRC Conference Congress report

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RE(ACT) Congress and IRDiRC Conference 2023

March 15-18 2023, Mélia Berlin, Berlin, Germany

Website: https://www.react-congress.org

The BLACKSWAN Foundation and IRDiRC, the International Rare Diseases Research Consortium, hosted the joint in-person event RE(ACT) Congress and IRDiRC Conference 2023, 15-18 March in Berlin, at the Mélia Hotel.

This joint event continues the IRDiRC Conference series (5th edition) and the RE(ACT) Congress series (7th edition). It aims to bring together scientific leaders, experts, and young scientists from various breakthrough scientific fields to present cutting-edge research, exchange ideas, and discuss policies related to rare diseases research. Patients and patient organizations committed to research will also attend to share their experiences and perspectives.

The RE(ACT) Congress – International Congress of Research on Rare and Orphan Diseases – was initiated in 2012 by the BLACKSWAN Foundation to create a forum for and promote scientific cooperation and research on rare and orphan diseases. IRDiRC – launched in April 2011 at the European Commission and the US National Institutes of Health initiative – fosters international collaboration on rare disease research by bringing together researchers, funders, and patient advocacy organizations that work collaboratively within a multinational consortium.

The event attracted around 160 attendees, including scientists, physicians, patient organizations, pharmaceutical industry representatives, start-ups, patients, and other international stakeholders. During the three-day conference, world-class speakers, panelists, and participants shared their vision, experiences and presented their innovative

and outstanding scientific research on rare diseases. The full list of speakers and panelists is available at http://www.react-congress.org/speakers/.

The conference presented the latest advancements in knowledge, understanding, and innovation in rare disease research and offered an expanded view of health systems for rare diseases. Focused sessions addressed new tools and technologies for diagnosis, progress, and challenges in advanced therapies for rare diseases, innovative approaches for clinical trials for ultra-rare diseases and methodologies to measure the impact of rare diseases diagnosis and treatments.

Emphasis was given to the barriers to accessing the approved therapies and treatments to patients as drivers in drug development and clinical trials. A hands-on workshop on the IRDiRC Orphan Drug Development guidebook and a rare diseases foresight session from across different areas of the globe completed the program.
Each session was followed by intense networking discussions involving speakers, patient representatives, and delegates providing deep insights as fundamental and indispensable actors along the whole diagnostic and therapeutic path.

Press release: https://www.ejprarediseases.org/15309-2/

Program

WEDNESDAY, MARCH 15th

Session A, 13h30 to 17, « Diagnostic, WGS, artificial intelligence, new technologies »

- Martina Cornel, NL « Sequencing for early diagnosis in neonatal screening and health care »
- Julia Foreman, UK « DECIPHER – Enabling the sharing of rare disease phenotype-linked variant data for diagnosis and research »
- Clara van Karnebeek, NL « A 'negative' exome – what's next? »
- Pierre-Emmanuel Gleizes, FR « A European network to identify and diagnose ribosomopathies »
- Tudor Groza, UK « Rare disease patient stratification in primary care: Challenges and opportunities »
- Kym Boycott, CAN « Strategic approaches to reduce the diagnostic odyssey »
- Elena Rojano, ES (Abstract A010) « PhenoClinWare: a webserver to explore human pathological phenotypes and expand patient clinical characterization"
- Klary Niezen-Koning, NL (Abstract A003) "A Case database for inherited metabolic diseases as a global, shared educational resource"

Poster Session A 17 to 18

Opening Ceremony, 18 to 19h30

Welcome message: Daria Julkowska (EJP RD), David Pierce (IRDiRC), Olivier Menzel (BLACKSWAN Foundation)

- Ruxandra Draghia-Akli, USA
- Gareth Baynam, AU « The Rare Care Centre »

THURSDAY, MARCH 16th

Session B, 9 to 12, « Therapeutic Development & precision medicine »

- Marc Dooms, BE & Anneliene Jonker, NL « Devise – ways forward for medical devices for rare diseases »
- Tim Buckinx, BE « The Potential of Wearable EEG & Real-Time AI to Externalize Brain States in Rare and Orphan Diseases: A Disruptive Vision to Improving Lives using Technologies of Today »

- Lucia Pannese, IT « Why Me? Tackling the Challenge of Rare Diseases through Gamification and Enabling Technologies »

- Jose-Alain Sahel, USA « Developing Gene-Independent Approaches for Retinal Dystrophies »

- Paulien Klap, NL « Developing an arm exoskeleton with the Duchenne Muscular Dystrophy community »

Lunch break and poster sessions B & C 12 to 13h30

Session C, 13h30 to 17, « Regulatory science »

- Daniel O'Connor, UK « Evolving regulatory science for rare diseases »

- Anne Pariser, USA "Regulatory opportunities: Facilitating an environment of innovation"

- Kerry Jo Lee, USA – FDA « FDA's Center for Drug Evaluation and Research (CDER): Considerations in Rare Disease Drug Development and How CDER is Accelerating Rare Disease Treatments »

- Violeta Stoyanova-Beninska, NL « Innovation in Rare Disease Drug Development–early dialogue with the EU regulators »

- Julienne Vaillancourt, USA « FDA's Center for Biologics Evaluation and Research: Advancing Development of Novel Biologics for Rare Diseases »

- Terence Beghyn, FR (Abstract B008) "Zellweger Spectrum Disorder : individualized research delivered new hopes for patients with PEX deficiency"

- Yustina Puspitasari, CH (Abstract B006) "Arterial thrombosis in Hutchinson Gilford Progeria Syndrome"

- Marjon Pasmooij, NL (Abstract C001) "Centralized and up-to-date data on orphan drugs: the European Medicines Regulatory Database"

FRIDAY, MARCH 17th

Session D, 9 to 12, « Clinical research »

- Marta Alarcón-Riquelme, ES « Heterogeneity of autoimmunity, clinical implications »

- Isabela Batsu, USA « Innovative clinical trial designs for rare diseases »

- PJ Brooks, USA « Beyond "One disease at a time": Focusing on common molecular etiologies to accelerate rare clinical trial access »

- Andrea Gropman, USA « From biomarker to study to basket clinical trials. Advancing science from the bedside or bench to trials: two models in academia »

- Sunil Rodger, DE (Abstract D001) “The Care and Trial Site Registry: FAIRification of an online database of clinical sites and their facilities”

Lunch break and poster sessions D, E & F 12 to 13h30

Session E, 13h30 to 17, « Gene and cell therapy »

- Guillaume Canaud, FR « Targeted therapy for patients with PIK3CA-related overgrowth spectrum »
- Arjan Lankester, NL « Lentiviral gene therapy in RAG1 severe combined immunodeficiency: experience from the multicenter RECOMB trial »
- Fernardo Larcher, ES « Advances in cell and gene therapy for genodermatoses. A focus in Epidermolysis Bullosa. »
- Carl Morris, USA « AAV-mediated gene therapy in Duchenne Muscular Dystrophy – Efficacy and immune responses »
- Julia Vitarello, USA From Mila to Millions: Opportunities for Individualized Medicines »
- Federica Tiberio, IT (Abstract E001) “In vitro development of a customized noninvasive nanoparticle-mediated gene knockdown approach for Crouzon syndrome”

Social event, 18h30

SATURDAY, MARCH 18th

Session F, 9 to 12h30, selected oral presentations and “Systems thinking towards access »

- Mary Wang, IT « Operational Description of Rare Diseases – A reference to improve the recognition and visibility of rare diseases »
- Samuel Agyei Wiafe, Ghana « Addressing the Unmet needs of persons living with Undiagnosed and Rare Diseases in Ghana »
- Yarlalu Thomas, AU « Lyfe Languages »
- Stefano Benvenuti, IT « Ensuring access to life-saving gene therapy for an ultra-rare disease: a not-for-profit model »
- Rachel Yang, CN/CH « Nurse, a critical role in RD care delivery and access »
- Anne Parkinson, AUS (Abstract F003) « Delayed diagnosis in three rare diseases: a qualitative study of the experiences of people with myositis, sarcoidosis, and primary immune deficiency in Australia”

Best posters prize & Closing Ceremony

Facts

- Total Number of Attendees registered: 155
- From the European Union: 61.3%
- Rest of the world: 38.7%
- International audience