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Del 5.3
First RE(ACT) [and IRDiRC Conference 2021] Congress report

Organisation name of lead beneficiary for this deliverable:
Partner 80 – BSF

Due date of deliverable: month 16

Dissemination level:
Public

"Due to COVID-19 the congress, initially planned in March 2020, was finally postponed to January 2021. In consequence the deliverable 5.3, First report of the RE(ACT) congress has been also postponed to June 2021."

RE(ACT) Congress and IRDiRC Conference 2021

Online edition, January 13-15, 2021

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

The RE(ACT) Congress IRDiRC Conference were held online in a first-time joint event on 13-15 January 2021, organized by the **BLACKSWAN Foundation** in collaboration with the International Rare Diseases Research Consortium (**IRDiRC**), and the European Joint Programme on Rare Diseases (**EJP RD**). The congress also benefited from the support of Rare Diseases International (RDI) and EURORDIS.

This joint event continued the IRDiRC Conference series (4th edition) and the RE(ACT)
Congress series (6th edition). It aimed to bring together scientific leaders and 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 were also in attendance 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 initiative of the European Commission and the US National Institutes of Health – 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 300 attendees, including scientists, physicians, patient organizations, pharmaceutical industry representatives and start-ups, patients, and other international stakeholders. During the three-day conference, world-class speakers, panellists, and participants shared their vision, experiences and presented their innovative and outstanding scientific research on rare diseases. The full list of speakers and panellists is available at http://www.react-congress.org/speakers/.

The conference presented the latest advancements in knowledge, understanding, and innovation in rare diseases research and offered an

expanded view onto 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 access 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 a panel discussion involving the speakers and one or more patient representatives, providing deep insights as fundamental and indispensable actors along the whole diagnostic and therapeutic path.

Program

WEDNESDAY, JANUARY 13th

Session A, 9 to 12, "Presentation of the Galaxy Guide & Hands-on"

- Virginie Hivert, FR – Eurordis
- Michela Gabaldo, IT – Fondazione Telethon
- Anneliene Jonker, NL – IRDiRC
- Diego Ardigò, IT – Chiesi

Opening Session 13 to 15

- Welcome messages
- Irene Norstedt – Director responsible for the Health Directorate within the Directorate-General for Research and Innovation, European Commission
- Lucia Monaco – Chair, IRDiRC Consortium Assembly
- Daria Julkowska – Coordinator. European Joint Program Rare Disease
- Olivier Menzel – Chairman and founder BLACKSWAN Foundation, founder RE(ACT) Congress
- Keynote presentation: Alexandre Reymond, Director of the Center for Integrative Genomics and president of the executive board of the European Society of Human Genetics, CH "Genome architecture and diseases: the 16p11.2 paradigm"

Session B, 15:30 to 17, "Rare Diseases Foresight: Panel discussion (EU/America/Asia/AU)"

- Yann Le Cam, FR – Eurordis "Recommendations from the Rare 2030 Foresight Study"
- Anne Pariser, USA – NCATS, NIH

• Matthew Bellgard, AUS "Vision of the Asia-Pacific Economic Cooperation (APEC) Rare Disease Network: Multilateral, Multi-stakeholder Rare Disease Policies & Plans

THURSDAY, JANUARY 14th

Session C, 10 to 12, "Diagnostic, WGS, artificial intelligence, new technologies" Chair: Jacqui Beckmann

- Short Film presented by Maja Bartoszewicz Moritz: Journey of hope
- Mark Caulfield, UK "The 100,000 Genomes Project Transforming Healthcare"
- Christoffer Nellåker, UK "Deep phenotyping from faces and the Minerva Initiative"
- Uzma Atif, USA "The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease"
- Peter Krawitz, USA "Next-Generation Phenotyping Using DeepGestalt in Clinic, Research and Variant Analysis"
- Clara van Karnebeek, NL "Diagnostic -omics: what's new in 2021?"
- Panel discussion: All speakers and patient representative (Virginie Bros-Facer – Eurordis and Maja Bartoszewicz Moritz)

Session D, 13 to 15, "Molecular etiology of RD, innovative clinical trials, precision medicine". Chair: PJ Brooks

- PJ Brooks, USA "Beyond "one disease at a time" so no disease is left behind: "Platform approaches to clinical trials in rare diseases"
- Anna Wedell, SE "Precision Diagnostics of Rare Diseases at the Genomic Medicine Center Karolinska"
- Terence Beghyn, FR "Individualized research program, sustainable approach through drug repurposing"
- Gisou van der Goot, CH "Hyaline Fibromatosis Syndrome: how the study of individual patient mutations drives the molecular understanding of the disease"
- Susan J Ward, USA "Learning from natural history patient data to drive smaller, faster, trials – a case study in Duchenne Muscular Dystrophy (DMD)"
- Panel discussion: All speakers and patient representative (Christian Rubio – Global Gene)

Session E, 15:30 to 17, "Advanced therapies: gene editing, cell therapy" Chair: Joseph Scheeren

- Joseph Scheeren, USA Challenges and considerations in the regulation of gene and cell therapies"
- Alessandro Aiuti, IT "Hematopoietic stem cell gene therapy for monogenic diseases: from experimental studies to
- approved drugs"

- Hans-Dieter Volk, DE "Immunological challenges in gene and cell therapy"
- Nathalie Cartier-Lacave, FR "Gene therapy for Huntington's disease and spinocerebellar ataxias: from preclinical proofs
- of concept to first Phase 1 clinical trial"
- Panel discussion: All speakers and patient representative (Oxana Illiach – CORD)

FRIDAY, JANUARY 15th

Session F, 10 to 12, "Patients as drivers in drug development and clinical trials" Chairs: Durhane Wong-Rieger and Samantha Parker

- François Houÿez, FR "Engaging Patients: The EuroCAB Programme"
- Dimitrios Athanasiou, GR" 25 years of Duchenne Patient Advocacy: Between Hype and Hope"
- Nick Sireau, UK "How patients can lead drug development: the case of the AKU Society"
- Nicola Bedlington, BE "Towards a sustainable patient engagement ecosystem"
- Panel discussion: All speakers and patient representative (Sharon Terry – Genetic Alliance)

Session G, 13 to 15, "Access to diagnostic and drugs for all" Chair: William Gahl

- Susanne Weissbaecker, SG "Taking action for Rare Diseases. If we don't, who will?"
- Benjamin Djoudalbaye, ET "Access to diagnostic and drugs for all in the African context"
- Durhane Wong-Rieger, CAN "Access to Rare Disease Drugs in Emerging Health Systems: Pathway for Access to Diagnosis, Treatment, Care and Patient Empowerment"
- William Gahl, USA "Global Access to Rare Disease Diagnostics and Treatment"
- Panel discussion: All speakers and patient representatives (Alba Ancochea-Diaz – ALIBER; Ramaiah Muthyala – I-ORD; Samuel Agyei Wiafe – Rare Diseases Ghana; Tanja Collin-Histed – International Gaucher Alliance; Eda Selebatso – BORDIS Botswana)

Session H, 15:30 to 17, "Methodologies to assess the effect of diagnosis and therapies on RD patients". Chair: David Pearce

- David Pearce, USA – Welcome & Introduction
- Daniel Ollendorf, USA "The Economics of Rare Disease: Value Assessment Challenges, Evidence Considerations, and Special-Case Status"

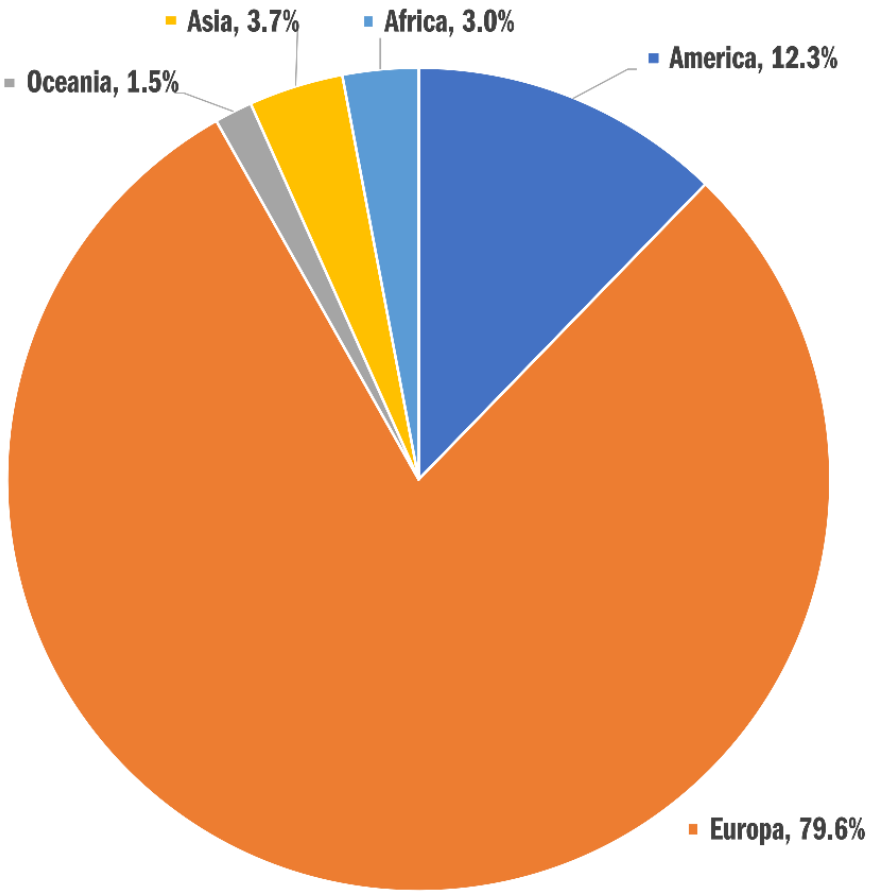
- Vicki Seyfert-Margolis, USA "Patient-centric digital technology to define disease progression and response to therapy: a model that also supports a de-centralized approach for clinical trials in rare diseases"
- Panel discussion: All speakers and patient representatives (Dimitrios Athanasiou – World Duchenne Organization; Vanessa Boulanger – NORD)

Facts

- Total Number of Attendees: 293
- "Active participants": 243 (downloaded Whova, the congress App)
- Private messages 1-on-1: 759
- Private group chats created: 15
- Attendee interaction 1-on-1 (Attendees who have interacted with each other): 646

International audience

Continents



Countries

Italy	13.8%
France	13.4%
UK	11.9%
USA	9.7%
Germany	7.8%
Switzerland	7.5%
Belgium	6.7%
Netherlands	5.2%
Spain	3.4%
Africa (Côte d'Ivoire, Botswana, Ethiopia)	3.0%
Canada	2.6%
Israel	1.9%
Australia	1.5%
Finland	1.5%
Georgia	1.1%
Austria	0.7%
Hungary	0.7%
Irland	0.7%
Portugal	0.7%
Turkey	0.7%
Japan	0.7%
Norway	0.7%
China	0.4%
Czech Republic	0.4%
Greece	0.4%
Malta	0.4%
Serbia	0.4%
Poland	0.4%
Sweden	0.4%
Singapore	0.4%
Albania	0.4%
India	0.4%