

IRDiRC 2017 Conference Planning Committee

Draft: IRDiRC 2017 Conference Program

4 January 2017

Conference Program – At a Glance

8 February 2017	
09:00 – 10:30	Plenary Session 1: Opening Session – IRDiRC History and Achievements
10:30 – 11:00	Coffee break
11:00 – 12:40	Plenary Session 2: Rare Diseases Research in 2017 – A Global View
12:40 – 14:00	Lunch break
14:00 – 15:30	Plenary Session 3: State of Foundational, Diagnostics and Therapeutics Research
15:30 – 16:00	Coffee break
16:00 – 18:00	Parallel Session 1: Foundational, Diagnostics and Therapeutics Research in 2017 - Foundational Track - Diagnostics Track - Therapeutics Track (5 established speakers per track, 24 minutes per talk = 20 minutes talk + 4 minutes Q&A?)
18:00 – 21:00	Poster session, cocktail reception

9 February 2017	
09:00 – 10:40	Parallel Session 2: New Approaches to Rare Diseases - Foundational Track - Diagnostics Track - Therapeutics Track (4 young investigators per track, 20 minutes per talk = 15 minutes talk + 5 minutes Q&A; 2 selected abstracts, 10 minutes per talk)
10:40 – 11:10	Coffee break
11:10 – 12:40	Parallel Session 3: Trends in the Field - Trends in Patient Advocacy - Trends in Companies - Trends in Regulatory and Access (4 speakers per track, 15 minutes per talk; 30 minutes panel Q&A)
12:40 – 14:00	Lunch break
14:00 – 16:30	Plenary Session 4: Transforming Rare Diseases Research – IRDiRC Goals 2017-2027
16:30 – 17:00	Continued discussion over coffee and refreshments
17:00	End of conference

8 February 2017 – Program of the Day

Morning	
09:00 – 10:30	Plenary Session 1: Opening Session – IRDiRC history and achievements Chair: Lucia Monaco
09:00 – 09:30	Welcome and Opening Address Ruxandra Draghia-Akli, DG Research and Innovation, Belgium Francis Collins
09:30 – 10:00	IRDiRC achievements after its first five years Paul Lasko, McGill University, Canada
10:00 – 10:30	IRDiRC: current state and future prospects Christopher Austin, National Center for Advancing Translational Sciences (NCATS), USA
10:30 – 11:00	Coffee break
Mid-morning	
11:00 – 12:40	Plenary Session 2: Rare Disease Research in 2017 – global view Chair: Stephen Groft
11:00 – 11:20	Europe Irene Norstedt, DG Research and Innovation, Belgium Caroline Hager, DG Health and Food Safety, Belgium
11:20 – 11:40	US/Canada Petra Kaufmann, National Center for Advancing Translational Sciences (NCATS), USA
11:40 – 12:00	Asia Makoto Suematsu, Japan Agency for Medical Research and Development (AMED), Japan
12:00 – 12:20	Australia Hugh Dawkins, Western Australia Department of Health, Australia
12:20 – 12:40	UN NGO Committee for Rare Diseases Anders Olauson, Ågrenska Foundation, Sweden
12:40 – 14:00	Lunch break
Afternoon	
14:00 – 15:30	Plenary Session 3: State of Foundational, Diagnostics, Therapeutics Research Chair: Makoto Suematsu
14:00 – 14:30	Foundational Hanns Lochmüller, Newcastle University, UK
14:30 – 15:00	Diagnostics Kym Boycott, Children’s Hospital Eastern Ontario, Canada
15:00 – 15:30	Therapeutics Diego Ardigò, Chiesi Farmaceutici S.p.A., Italy Yann Le Cam, EURORDIS, France/Belgium
15:30 – 16:00	Coffee break
Late afternoon / Evening	
16:00 – 18:00	Parallel Session 1: Foundational, Diagnostics and Therapeutics Research in 2017 (see page 3 for more information)
18:00 – 20:00	Poster session, cocktail reception

8 February 2017 - Parallel Session 1

Track 1: Foundational	
16:00 – 18:00	Foundational Research in 2017 Chair: Hanns Lochmüller
	<p>Data standards Melissa Haendel, Monarch Initiative and Oregon Health & Science University, USA</p> <p>Networks Matthias Kretzler, EuRenOmics and University of Michigan, USA</p> <p>Multi-stakeholder study Anders Larsson, Uppsala University, Sweden</p> <p>Registries Fernando Poletta, ECLAMC, Argentina</p> <p>Data sharing and patient preference Mats Hansson, Uppsala University, Sweden</p>
Track 2: Diagnostics	
16:00 – 18:00	Diagnostics Research in 2017 Chair: Kym Boycott
	<p>Data exchange – MME Ada Hamosh, Johns Hopkins University School of Medicine, USA</p> <p>Gene discovery David Valle, Johns Hopkins University School of Medicine, USA</p> <p>Newborn screening Stephen Kingsmore, Rady Children’s San Diego, USA</p> <p>Undiagnosed diseases Gareth Baynam, University of Western Australia, Australia</p> <p>High level EU view, policy Milan Macek, Charles University, Czech Republic</p>
Track 3: Therapeutics	
16:00 – 18:00	Therapeutics Research in 2017 Chair: Diego Ardigò
	<p>First ex-vivo stem cell gene therapy for ADA-SCID Claudio Bordignon, MolMed S.p.A., Italy</p> <p>Patient engagement in therapeutic development? Sangeeta Jethwa, Roche Innovation Centre, Switzerland</p> <p>World’s first commercial stem cell therapy Holoclar Graziella Pellegrini, University of Modena and Reggio Emilia, Italy</p> <p>Gene therapy for neurologic disorders Jodi Cook, Agilis Biotherapeutics LLC, USA</p> <p>GNE myopathy Ichizo Nishino, National Center of Neurology and Psychiatry, Japan</p>

9 February 2017 – Program of the Day

Morning	
09:00 – 10:40	Parallel Session 2: New Approaches to Rare Diseases (see page 5-6 for more information)
10:40 – 11:10	Coffee break
Mid-morning	
11:10 – 12:40	Parallel Session 3: Trends in the Field (see page 6 for more information)
12:40 – 14:00	Lunch break
Afternoon	
14:00 – 17:00	Plenary Session 4: Transforming Rare Disease Research – IRDiRC Goals 2017-2027 Co-Chairs: C Austin and H Dawkins
14:00 – 14:30	IRDiRC goal-setting process, to date C Austin
14:30 – 16:30	Panel discussion and selection of IRDiRC goals for 2017-2027 Moderator: H Dawkins Panellists: Chairs of <ul style="list-style-type: none"> ○ Funders CC: TBD ○ Companies CC: TBD ○ Patient Advocates CC: TBD ○ DSC: Kym Boycott ○ ISC: Hanns Lochmüller ○ TSC: Diego Ardigò ○ CA: Christopher Austin
16:30 – 17:00	Continued discussion over coffee and refreshments
17:00	End of conference

9 February 2017 - Parallel Session 2: New Approaches to Rare Diseases

Track 1: Foundational	
09:00 – 10:40	Chair: Daria Julkowska
	<p>Title Slavé Petrovski, University of Melbourne, Australia</p> <p>Title Romain Marignier, CHU de Lyon, France</p> <p>Title Sonia Vallabh & Eric Minikel, Broad Institute, USA</p> <p>Title Nan Wu, Peking Union Medical College Hospital, China</p> <p>Selected abstract: Status of Rare Disease Ecosystem in India – Progress and lessons for rest of the world Harsha Rajasimha, Organization for Rare Diseases India and George Mason University, USA</p> <p>Selected abstract: Boosting health care and life science research on rare diseases by creating a robust infrastructure of independently Findable, Accessible, Interoperable, and Reusable (FAIR) biobanks, registries, and molecular data resources Marco Roos, Leiden University Medical Centre, The Netherlands</p>
Track 2: Diagnostics	
09:00 – 10:40	Chair: Gareth Baynam
	<p>Title Toshiki Takenouchi, Keio University, Japan</p> <p>Title Heike Heuer, Leibniz Research Institute for Environmental Medicine, Germany</p> <p>Title Robert Hufnagel, National Eye Institute (NEI), USA</p> <p>Title Sophie Nicole, Université Pierre et Marie Curie, France</p> <p>Selected abstract: Solving the Lonely Exome: International Connectivity to Enable Discovery Taila Hartley, CHEO, University of Ottawa, Canada</p> <p>Selected abstract: Variant data from patients with rare diseases semantically linked and enriched with gene and variant data from public data sources Filip Pattyn, ONTOFORCE, Belgium</p>
Track 3: Therapeutics	
09:00 – 10:40	Chair: Michela Gabaldo
	<p>Title Chris Gibson, Recursion Pharmaceuticals Inc., USA</p> <p>Title Stuart Hughes, Vertex Pharmaceuticals Inc., UK</p> <p>Title Federico Mingozzi, Genethon, France</p> <p>Title Marc Martinell, Minoryx Therapeutics, Spain</p>

	<p>Selected abstract: IDeAI designing a clinical trial – a case study Ralf-Dieter Hilgers, Department of Medical Statistics, RWTH University Aachen, Germany</p> <p>Selected abstract: Translating Natural History into Clinical Trial Design – lessons from Duchenne Muscular Dystrophy Susan Ward, Analysis Group, Boston USA, cTAP, Cambridge USA</p>
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9 February 2017 - Parallel Session 3: Trends in the Field

Track 1: Trends in Patient Advocacy	
11:10 – 12:40	Chair/Moderator: Katherine Beaverson
	<p>NGLY1 Matt Might, University of Utah, USA</p> <p>Title Yukiko Nishimura, ASrid, Japan</p> <p>Title Yann Le Cam, EURORDIS, France/Belgium</p> <p>Title Hawa Dramé, Fitima Foundation, Burkina Faso</p> <p>Panel discussion with all speakers of the session</p>
Track 2: Trends in Companies	
11:10 – 12:40	Chair/Moderator: Sangeeta Jethwa
	<p>Title Kiran Reddy, Clarus Ventures, USA</p> <p>Title Carlo Incerti, Genzyme Europe, Italy</p> <p>Title Kristina Larsson, European Medicines Agency (EMA), UK</p> <p>Title Alain Huriez, Advent Life Sciences, Paris</p> <p>Panel discussion with all speakers of the session</p>
Track 3: Trends in Regulatory and Access	
11:10 – 12:40	Chair/Moderator: Irene Norstedt
	<p>Title Edmund Jessop, National Health Service (NHS) England, UK</p> <p>Title Jonathan Goldsmith, US Food and Drug Administration (FDA), USA</p> <p>Title Hideyuki Kondo, Pharmaceuticals and Medical Devices Agency (PMDA), Japan</p> <p>Title Anna Bucsecs, University of Vienna and MoCA, Austria</p> <p>Panel discussion with all speakers of the session</p>