Program-at-a-Glance

February 8, 2017			
09:00 – 10:30	**Plenary Session 1: Opening Session – IRDiRC History and Achievements**		
	Auditorium		
10:30 – 11:00	Coffee break		
	Reception Hall		
11:00 – 12:40	**Plenary Session 2: Rare Diseases Research in 2017 – A Global View**		
	Auditorium		
12:40 – 14:00	Lunch break		
	Reception Hall		
14:00 – 15:30	**Plenary Session 3: State of Foundational, Diagnostics and Therapeutics Research**		
	Auditorium		
15:30 – 16:00	Coffee break		
	Reception Hall		
16:00 – 18:00	**Parallel Session 1: Diagnostics, Foundational and Therapeutics Research in 2017**		
	Diagnostics Track	*Foundational Track*	*Therapeutics Track*
	Auditorium	Room 106	Room 108
18:00 – 20:00	Poster session, cocktail reception		
	Reception Hall		
February 9, 2017			
09:00 – 10:40	**Parallel Session 2: New Approaches to Rare Diseases**		
	Diagnostics Track	*Foundational Track*	*Therapeutics Track*
	Auditorium	Room 106	Room 108
10:40 – 11:10	Coffee break		
	Reception Hall		
11:10 – 12:40	**Parallel Session 3: Trends in the Field**		
	Trends in Regulatory & Access	*Trends in Patient Advocacy*	*Trends in Companies*
	Auditorium	Room 106	Room 108
12:40 – 14:00	Lunch break		
	Reception Hall		
14:00 – 16:30	**Plenary Session 4: Transforming Rare Diseases Research – IRDiRC Goals 2017-2027**		
	Auditorium		
16:30 – 17:00	Continued discussion over coffee and refreshments		
17:00	End of conference		

Detailed Program and Speakers

February 8, 2017

09:00 – 10:30 *Auditorium*	**Plenary Session 1: Opening Session – IRDiRC History and Achievements** *Chair: Lucia Monaco*
	IRDiRC: Stepping Stones towards Success Ruxandra Draghia-Akli, DG Research and Innovation, European Commission, Belgium Francis Collins, National Institutes of Health (NIH), USA **IRDiRC: A Review of its Achievements in its First Six Years** Paul Lasko, McGill University, Canada **IRDiRC: Current State and Future Prospects** Christopher Austin, National Center for Advancing Translational Sciences (NCATS), USA **Panel Q&A** Ruxandra Draghia-Akli, Paul Lasko, Christopher Austin
10:30 – 11:00 *Reception Hall*	Coffee break
11:00 – 12:40 *Auditorium*	**Plenary Session 2: Rare Diseases Research in 2017 – A Global View** *Chair: Stephen Groft*
	AMED Challenges Data Sharing for Undiagnosed Patients Makoto Suematsu, Japan Agency for Medical Research and Development (AMED), Japan **A European Rare Disease Overview** Irene Norstedt, DG Research and Innovation, European Commission, Belgium Caroline Hager, DG Health and Food Safety, European Commission, Belgium **Office of Rare Diseases Research: Perspective on North American Rare Diseases Research** Petra Kaufmann, National Center for Advancing Translational Sciences (NCATS), USA **From Research Translation to Transformation in a Public Health System** Hugh Dawkins, Western Australia Department of Health, Australia **UN NGO Committee for Rare Diseases (CfRD)** Anders Olauson, Ågrenska Foundation, Sweden
12:40 – 14:00 *Reception Hall*	Lunch break
14:00 – 15:30 *Auditorium*	**Plenary Session 3: State of Foundational, Diagnostics, Therapeutics Research** *Chair: Makoto Suematsu*
	Cross-Cutting Bottlenecks and Solutions in Rare Diseases Research Hanns Lochmüller, Newcastle University, UK **International Cooperation to Enable the Diagnosis of Most Rare Genetic Diseases by 2020** Kym Boycott, Children's Hospital Eastern Ontario, Canada **200 Rare Disease Therapies Scored in 2017 – New Objective: 500 in 2027** Diego Ardigò, Chiesi Farmaceutici S.p.A., Italy Yann Le Cam, Rare Diseases Europe-EURORDIS, France/Belgium
15:30 – 16:00 *Reception Hall*	Coffee break

16:00 – 18:00	Parallel Session 1: Diagnostics, Foundational and Therapeutics Research in 2017
Auditorium	**Track 1: Diagnostics Research in 2017** *Chair: Kym Boycott*
	Mendelian Disease and the Centers for Mendelian Genomics: Progress, Challenges and Opportunities David Valle, Johns Hopkins University School of Medicine, USA **The Matchmaker Exchange, a Global Effort to Identify Novel Disease Genes** Ada Hamosh, Johns Hopkins University School of Medicine, USA **Interpretation of the Disease Liability of Genomic Variants in Rare Diseases: Multi-Disciplinary and International Locus Specific Collaborative Initiatives (CFTR2.org)** Milan Macek, Charles University, Czech Republic **Undiagnosed Disease Programs and Networks** Gareth Baynam, University of Western Australia, Australia **Enabling Neonatal Precision Medicine by Rapid Genome Sequencing** Stephen Kingsmore, Rady Children's San Diego, USA **Panel Q&A** With all speakers of the session
Room 106	**Track 2: Foundational Research in 2017** *Chair: Hanns Lochmüller*
	How to Promote Data Sharing in Rare Disease while Protecting Privacy Mats Hansson, Uppsala University, Sweden **The Impactt Study: Experiences from Performing a Clinical Multicenter Study in Collaboration with CF Patient Organizations** Anders Larsson, Uppsala University, Sweden **Global Phenotypic Data Sharing Standards to Maximize Diagnostics and Mechanism Discovery** Melissa Haendel, Monarch Initiative and Oregon Health & Science University, USA **Precision Medicine in Rare Diseases Across Continents and Disciplines** Matthias Kretzler, NEPTUNE, EuRenOmics and University of Michigan, USA **European Perspective on Sharing –Omics Data for Personalized Medicine in Rare Diseases** Ivo Gut, Centro Nacional de Análisis Genómico (CNAG), Spain **Panel Q&A** With all speakers of the session

Room 108	**Track 3: Therapeutics Research in 2017** *Chair: Diego Ardigò*
	Ex-Vivo Stem Cell Gene Therapy: Approved Treatment for ADA-SCID Claudio Bordignon, MolMed S.p.A., Italy
	Approval of a Stem Cell Therapy for Corneal Disease, Holoclar® Graziella Pellegrini, University of Modena and Reggio Emilia, Italy
	Gene Therapy for Neurological Disorders: A Promising Novel Treatment for AADC Deficiency Jodi Cook, Agilis Biotherapeutics LLC, USA
	Development of Therapy for GNE Myopathy Ichizo Nishino, National Center of Neurology and Psychiatry, Japan
	Patient Engagement in Therapeutics Development Sangeeta Jethwa, Roche Innovation Centre, Switzerland
	Panel Q&A With all speakers of the session
18:00 – 20:00 *Reception Hall*	Poster session, cocktail reception

February 9, 2017

09:00 – 10:40	**Parallel Session 2: New Approaches to Rare Diseases**
Auditorium	**Track 1: New Approaches to Rare Diseases – Diagnostics** *Chair: Gareth Baynam*
	Identification of Two New Disease Entities Through the Undiagnosed Disease Program at our Institution Toshiki Takenouchi, Keio University, Japan
	High Throughput Screening Toward Precision Medicine in Congenital Myastenic Syndromes Sophie Nicole, Université Pierre et Marie Curie, France
	More than Meets the Eye: Solving an Evolutionary Riddle Using Rare Disease Robert Hufnagel, National Eye Institute (NEI), USA
	Development of Therapeutic Strategies for Patients with Allan-Herndon-Dudley Syndrome Edward Visser, Erasmus Medical Center, The Netherlands
Selected abstract	**Solving the Lonely Exome: International Connectivity to Enable Discovery** Taila Hartley, CHEO, University of Ottawa, Canada
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

Room 106	**Track 2: New Approaches to Rare Diseases – Foundational** *Chair: Daria Julkowska*
	Leveraging Standing Human Variation to Improve Missense Variant Interpretation Slavé Petrovski, University of Melbourne, Australia
	Eugene Devic European Network (EDEN): Establishment and Use of an European Database and Biobanks for Research and Treatment in Neuromyelitis Optica Romain Marignier, CHU de Lyon, France
	From Genetics to Therapeutics in Prion Disease Sonia Vallabh & Eric Minikel, Broad Institute, USA
	A Novel Subtype of Congenital Scoliosis: TBX6-Associated Congenital Scoliosis Nan Wu, Peking Union Medical College Hospital, China
Selected abstract	**Status of Rare Diseases Ecosystem in India – Progress and Lessons for Rest of the World** Harsha Rajasimha, Organization for Rare Diseases India and George Mason University, USA
Selected abstract	**Boosting Health Care and Life Science Research on Rare Diseases by Creating a Robust Infrastructure of Independently FAIR Biobanks, Registries, and Molecular Data Resources** Marco Roos, Leiden University Medical Centre, The Netherlands
Room 108	**Track 3: New Approaches to Rare Diseases - Therapeutics** *Chair: Michela Gabaldo*
	Translating AAV-Based in vivo Gene Therapies to the Clinic Federico Mingozzi, Genethon, France
	Developing New Therapies for Rare Diseases: Beyond Cystic Fibrosis Stuart Hughes, Vertex Pharmaceuticals Inc., UK
	Developing Therapies for Inborn Errors of Metabolism Marc Martinell, Minoryx Therapeutics, Spain
	Disrupting Discovery Efficiency: Combining the Best of Biology, Automation and Artificial Intelligence to Identify 100 Rare Disease Treatments in 10 years Christopher Gibson, Recursion Pharmaceuticals Inc., USA
Selected abstract	**IDeAl Designing a Clinical Trial – a Case Study** Ralf-Dieter Hilgers, Department of Medical Statistics, RWTH University Aachen, Germany
Selected abstract	**Translating Natural History into Clinical Trial Design – Lessons from Duchenne Muscular Dystrophy** Susan Ward, collaborative Trajectory Analysis Group (cTAP), Cambridge, USA
10:40 – 11:10 *Reception Hall*	Coffee break

11:10 – 12:40	**Parallel Session 3: Trends in the Field**
Auditorium	**Track 1: Trends in Regulatory and Access** *Chair: Irene Norstedt*
	Regulatory Trends Including Expanded Access Jonathan Goldsmith, US Food and Drug Administration (FDA), USA **Regulatory/Scientific Support for Rare Disease Product Development in Japan – Orphan Product Designation System** Hideyuki Kondo, Pharmaceuticals and Medical Devices Agency (PMDA), Japan **Challenges in Reimbursing Orphan Medicinal Products: Evaluating Benefit, Determining a Fair Price and Optimizing Access** Anna Bucsics, University of Vienna and MoCA, Austria **Managed Access for Ultra Orphan Drugs in England** Edmund Jessop, National Health Service (NHS) England, UK **Panel Q&A** With all speakers of the session
Room 106	**Track 2: Trends in Patient Advocacy** *Chair: Katherine Beaverson*
	Perspective on Patient Engagement in Research, Product Life Cycle and Healthcare in Europe Yann Le Cam, Rare Diseases Europe-EURORDIS, France/Belgium **The Algorithm for Precision Medicine** Matt Might, University of Utah, USA **Recent Japanese NANBYO Situation – How Japanese Patient Groups Contribute to Further the Research Field** Yukiko Nishimura, ASrid, Japan **Management of Patients with Rare Diseases in African Context: The Contribution of Fitima** Hawa Dramé, Fitima Foundation, Burkina Faso **Panel Q&A** With all speakers of the session
Room 108	**Track 3: Trends in Companies** *Chair: Sangeeta Jethwa*
	Current Status and Future Trends in Orphan Diseases: A Company Perspective Carlo Incerti, Sanofi Genzyme, USA **The DNA of Successful Rare Disease Biotechs** Kiran Reddy, Clarus Ventures, USA **The Economics of Rare Diseases from the Venture Capital Perspective** Alain Huriez, Advent Life Sciences, France **Trends in Orphan Development: What can be Extracted from a Regulator's Database** Kristina Larsson, European Medicines Agency (EMA), UK **Panel Q&A** With all speakers of the session
12:40 – 14:00 *Reception Hall*	Lunch break

14:00 – 17:00 *Auditorium*	**Plenary Session 4: Transforming Rare Diseases Research – IRDiRC Goals 2017-2027** *Co-Chairs: Christopher Austin and Hugh Dawkins*
14:00 – 14:30	**IRDiRC Goal-Setting Process, to Date** Christopher Austin, Chair of IRDiRC Consortium Assembly
14:30 – 16:30	**Panel Discussion and Selection of IRDiRC Goals for 2017-2027** Moderator: Hugh Dawkins Panellists: Representatives of IRDiRC Consortium Assembly, Constituent Committees and Scientific Committees
16:30 – 17:00 *Reception Hall*	Continued discussion over coffee and refreshments
17:00	**End of conference**