8th Rare Disease Summer School

Kartause Ittingen, Warth

Wednesday, July 13th to Friday, July 15th 2022

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.

WEDNESDAY, JULY 13, 2022	
09:00 – 10:15	ARRIVAL OF THE PARTICIPANTS AND REGISTRATION
10:15 – 10:30	**Welcome,** Prof. Matthias Baumgartner/Prof. Janine Reichenbach
10:30 – 11:20	Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands **Latest developments in exome and genome sequencing: impact for rare diseases**
11:20 – 12:15	Prof. Marshall Summar, Children's National Medical Center, George Washington University, USA **Designing rare disease clinical trials: outcomes, sample size, natural history, and recruitment**
12:15 – 13:30	LUNCH
13:30 – 14:30	**Oral presentations** by selected participants
14:30 – 15:10	Prof. Sebastian Jessberger, Brain Research Institute, University of Zurich, Switzerland **New nerve cells for old brains**
15:10 – 15:40	COFFEE BREAK
15:40 – 16:25	Julian Isla, Dravet Syndrome Foundation, Spain **Using AI for rare diseases diagnosis: my personal journey**
16:30 – 18:00	Dr. Jürg Streuli, Insitute of Biomedical Ethics, University of Zurich, Switzerland **Interactive workshop: Ethical considerations (case study)**
18:00 – 18:30	Check-in time
18:30 – 20:00	DINNER
20:00 –	**Poster viewing**

THURSDAY, JULY 14, 2022	
07:45 – 08:45	BREAKFAST
08:45 – 09:45	**Oral presentations** by selected participants
09:50 – 10:30	Dr. Paulin Jirkof, Office for Animal Welfare and 3R, University of Zurich, Switzerland **Towards better and humane science: The 3Rs principle**
10:30 – 11:00	COFFEE BREAK
11:00 – 12:15	Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands **Workshop: Next generation sequencing**
12:15 – 13:30	LUNCH
13:30 – 14:10	Prof. Stephan Neuhauss, Department of Molecular Science, University of Zurich **Studying rare diseases in the (not so rare) Zebrafish model organism**
14:15 – 15:55	**Poster viewing and coffee break**
16:00 – 17:20	Prof. Marshall Summar, Children's National Medical Center, George Washington University, USA **Workshop: So you want to develop a rare disease drug**
17:25 – 18:10	Prof. Michele De Luca, Regenerative Medicine Center, Modena and Reggio Emilia University, Italy **Keynote Lecture: Combined cell and gene therapy for Epidermolysis Bullosa**
18:10 – 18:30	Free time
18:30 – 21:00	GRILL

FRIDAY, JULY 15, 2022	
07:45 – 08:45	BREAKFAST
08:45 – 09:30	**Take-home messages** by participants
09:35 – 10:15	Dr. Fabienne Hartmann, University of Zurich, Cutiss AG, Switzerland **Regulatory framework for ATMPs on the basis of a case study**
10:15 – 10:45	COFFEE BREAK
10:45 – 11:30	Prof. Andrew Dwyer, Boston College, USA **Person-centered approaches for bridging health disparities for rare disease patients**
11:30 – 12:15	Dr. Jasmin Barman-Aksözen, Scientific advisor, Swiss society for porphyria, Switzerland **Let the sunshine in! Getting a drug approved for the rare genetic light intolerance erythropoietic protoporphyria**
12:15 – 12:30	Award of poster prizes and concluding remarks
12:30	END OF THE SUMMER SCHOOL AND DEPARTURE OF THE PARTICIPANTS

SPEAKER (*in alphabetical order*)

DR. JASMIN BARMAN-AKSÖZEN

Jasmin Barman-Aksäzen is a scientist and a patient with the ultra-rare disease erythropoietic protoporphyria (EPP), an inborn error of the heme biosynthesis. After studying molecular biology, biochemistry, and plant sciences at the University of Heidelberg, in 2007 she joined the team of Prof. E. Minder and Prof. X. Schneider-Yin at the municipal hospital Triemli in Zurich for her PhD thesis on iron metabolism and gene expression in EPP. During this time, she helped to develop the first treatment for EPP, afamelanotide, and in 2014 became a patient representative for the approval proceedings at the European Medicines Agency. To promote access to therapy for all patients with EPP and other porphyrias, she co-founded the International Porphyria Patient Network, a group of dedicated patient advocates with a professional background in science, medicine, and other relevant expertise. She is currently studying health economics and methods of health technology assessment at the University of Applied Sciences in Winterthur and works part time as a scientific associate in the diagnostics and research laboratory of the Triemli hospital.

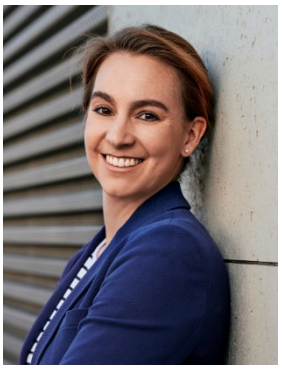
PROF. MICHELE DE LUCA

Michele De Luca is Full Professor of Biochemistry, Director of the Centre for Regenerative Medicine “Stefano Ferrari” at the University of Modena and Reggio Emilia and Scientific Director and founder of the university spin-off Holostem, the first biotech entirely devoted to development of ATMPs for cell and gene therapy based on epithelial stem cells. He received his MD at the University of Catania and his specialization in Endocrinology at the University of Rome. Prof. De Luca is author of over 130 peer reviewed international publications, has been invited speaker in over 350 international meetings and seminars and has received numerous international awards. He is a leading scientist in the field of epithelial stem cell biology aimed at clinical application in Regenerative Medicine and played a pivotal role in epithelial stem cell-mediated cell and gene therapy.

DR. ANDREW DWYER

Andrew Dwyer is a board-certified Family Nurse Practitioner with 20+ years of experience in endocrinology and translational research at the Massachusetts General Hospital (MGH) in Boston and the University Hospital of Lausanne (CHUV) in Switzerland. Currently, he is an Assistant Professor of nursing at Boston College and investigator in the MGH-Harvard Center for Reproductive Medicine. His clinical focus is male reproductive endocrinology, rare disorders of growth/puberty and transitional care. He has worked in interprofessional research teams, has authored/co-authored >100 articles, and serves on the Massachusetts Rare Disease Advisory Board. Dr. Dwyer is an NIH-funded researcher who utilizes patient engagement and digital solutions to develop person-centered approaches to care and overcome genomic health disparities.

DR. FABIENNE HARTMANN-FRITSCH

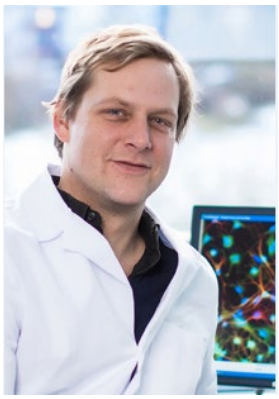


Fabienne Hartmann-Fritsch received her MSc in Microbiology and Immunology from the ETH Zurich in 2008 and her PhD in Biology from the University of Zurich in 2013. During her postdoctoral studies at the Tissue Biology Research Unit of the University Children's Hospital Zurich, Dr. Hartmann-Fritsch specialized on GMP-production of human skin grafts as well as on clinical trial management and regulatory affairs. In 2017, she co-founded the UZH spin-off company CUTISS AG, with the vision to provide patients who suffer from large and deep skin defects with the first personalized and automated skin tissue therapy that is safe, effective, and accessible for children and adults.

JULIAN ISLA

Julian Isla is the Chief Scientific Officer of the European Dravet Syndrome Federation (DSEF). DSEF is committed to find new treatments for Dravet Syndrome, an epileptic encephalopathy having long lasting seizures refractory to treatment as severe developmental delay. Julian is the father of a 13-years old boy who has Dravet Syndrome. He is a software engineer by training and works full- of Foundation 29, a non-profit organization focused on covering the unmet medical needs of patients with rare diseases using technology and how artificial intelligence can empower people to make decisions about their own health, based on the evidence provided by data and supported by automated interpretation systems. He is a member of the Orphan Drug Committee at European Medicines Agency (EMA) as patient representative, and he is part of the Therapeutic Advisory Group for Eurordis.

DR. SEBASTIAN JESSBERGER



Sebastian Jessberger is Director of the Brain Research Institute of the University of Zurich (UZH), Switzerland. He studied Medicine and carried out his medical thesis at the Center for Molecular Neurobiology (ZMNH) in Hamburg, Germany. In 2002 he started a joint residency in neurology at the Max Delbruck Center for Molecular Medicine (MDC) and the Dept. of Neurology of the Charité University Hospital in Berlin, Germany. As a postdoctoral fellow (2004-2007) in the laboratory of Fred H. Gage at the Salk Institute for Biological Studies in La Jolla, USA he continued to work on neural stem cell biology and neurogenesis in the adult brain. From 2007 to 2012 he was Assistant Professor at the ETH Zurich before joining the Brain Research Institute of UZH.

DR. PAULIN JIRKOF

Paulin Jirkof studied biology with a focus on Zoology in Germany. She obtained her PhD in Neuroscience at the University of Zurich, Switzerland and a Master of Advanced Studies in Management, Technology, and Economics at the ETH, Zurich, Switzerland. Her scientific field of interest is in the implementation and evaluation of severity assessment tools and the reliable assessment and treatment of pain in laboratory rodents. Currently she works as 3R coordinator at the Office for Animal Welfare and 3Rs of the University of Zurich. She is chair of the executive board of the Swiss 3R Competence Center and board member of the Swiss Society for Laboratory Animal Science.

PROF. NINE KNOERS

Nine Knoers is the chair of the Department of Genetics at the University Medical Centre Groningen, The Netherlands. She received her M.D. (1986) and Ph.D. (cum laude, 1990) from the Catholic University Nijmegen, The Netherlands. She was trained and certified as Clinical Geneticist at the Radboud University Medical Centre Nijmegen. From 2011 until 2018, she was chair of the Department of Genetics at the University Medical Centre Utrecht, The Netherlands. Her major research effort focuses on the identification and further characterization of genes for inherited renal disorders and on their pathophysiology. The ultimate aim of her studies is to find clues for treatments for these disorders. Prof. Knoers is member of several European Consortia on genetic renal disorders and coordinated the Dutch Scientific Consortium on renal ciliopathies “Kouncil”. She has published over 280 scientific papers and in the last 10 years delivered >250 invited lectures.

PROF. STEPHAN NEUHAUSS

Stephan Neuhauss studied biology in Tübingen (D) and Eugene (Oregon, USA) doing his graduate work at Harvard Medical School (Massachussetts General Hospital). He then returned to Europe for a postdoctoral fellowship to the Max-Planck Institute for Developmental Biology (Tübingen) before accepting an senior lecture ship of the ETH Zurich at the Brain Research Institute. After stints as an assistant professor at the ETH, he become associate and later full professor at the Department of Molecular Life Sciences of the University of Zurich. He is also Vice-Dean for Research at the Faculty of Science and an editor of various open access journals.

DR. JÜRG STREULI

Jürg Streuli is head of the Pediatric Palliative and Advanced Care Team at the Children's Hospital of Easter Switzerland, Senior Researcher and Research group leader for Pediatric Bioethics at the Institute of Biomedical Ethics at the University of Zurich. Dr. Streuli studied medicine (state examination 2005) and specialized in pediatric and adolescent medicine at the Triemli City Hospital (2006-2008) and at the University Children's Hospital Zurich (2012-2015). In 2014, he obtained his doctoral degree in biomedical ethics and law from the University of Zurich. In his research and clinical work, Dr. Streuli aims to assess, develop and support integrated and universal health care services that enables both local accessibility for support and management, and access to specialist services for the care of complex medical conditions.

PROF. MARSHALL SUMMAR

Marshall Summar, is the Director of the Rare Disease Institute, and Chief of the Division of Genetics and Metabolism at the Children's National Hospital. Prof. Summar received his MD degree from the University of Tennessee, completed a residency and internship in pediatrics and medical/ biochemical genetics at Vanderbilt University. He is board-certified in pediatrics, clinical genetics and medical biochemical genetics. His research is focused on devices and treatments for patients with genetic and biochemical diseases and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs undergoing Food and Drug Administration (FDA) trials for patients with congenital heart disease and organic acidemias. Clinically, he launched, and leads the first clinical Rare Disease Institute (RDI) at Children's National Hospital. He is involved in the WHO's effort to globally expand care and diagnosis for rare disease patients and in public policy around rare diseases and orphan drug development in the U.S.