9th Rare Disease Summer School

Kartause Ittingen, Warth

July 4th to 7th 2023

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.

TUESDAY, JULY 4th 2023	
13:00 – 14:00	ARRIVAL OF THE PARTICIPANTS AND REGISTRATION
14:00 – 14:30	**Welcome and introduction round**, Prof. Matthias Baumgartner
14:30 – 15:15	Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands **Latest developments in exome and genome sequencing: impact for rare diseases**
15:15 – 15:45	COFFEE BREAK
15:45 – 16:30	Dr. Jasmin Barman-Aksözen, Scientific advisor, Swiss society for porphyria, Switzerland **Let the sunshine in! Development, approval and benefit assessment of afamelanotide, the first drug for treating erythropoietic protoporphyria.**
16:30 – 17:30	**Oral presentations** by selected participants
17:30 – 18:30	Check-in time
18:30 – 20:00	DINNER
20:00 –	**Poster viewing**

WEDNESDAY, JULY 5th 2023	
07:45 – 08:45	BREAKFAST
08:45 – 09:30	Prof. Marshall Summar, Children ‘s National Medical Center, George Washington University, USA **Rare diseases today**
09:30 – 10:15	Prof. Stephan Neuhauss, Department of Molecular Science, University of Zurich, Switzerland **Studying rare diseases in the (not so rare) Zebrafish model organism**
10:15 – 10:45	COFFEE BREAK
10:45 – 12:15	Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands **Workshop: Next generation sequencing**
12:15 – 13:45	LUNCH
13:45 – 14:30	Prof. Andrew Dwyer, Boston College, USA **Person-centred approaches for bridging health disparities for rare disease patients**
14:30 – 15:15	Dr. Fabienne Hartmann-Fritsch, University of Zurich, CUTISS AG, Switzerland **Regulatory framework for ATMPs on the basis of a case study**
15:15 – 15:45	COFFEE BREAK
15:45 – 16:30	Dr. Paul Bastard, Imagine Institute Paris, France ***Keynote Lecture:* From rare to more frequent causes of severe viral diseases: genetics and auto-immunity**
16:30 – 17:45	Networking/Free time
17:45 – 18:30	Visit to the Kartause Museum
18:30 – 20:00	DINNER
20:00 –	**Poster viewing**

THURSDAY, JULY 6th 2023	
07:45 – 08:45	BREAKFAST
08:45 – 10:15	Dr. Jürg Streuli, Institute of Biomedical Ethics, University of Zurich, Switzerland **Interactive workshop: Ethical considerations (case study)**
10:15 – 10:45	COFFEE BREAK
10:45 – 11:30	Prof. Elvir Becirovic, Ophthalmology, University Hospital Zurich, Switzerland **Treatment of inherited retinal diseases: chances and obstacles**
11:30 – 12:30	**Oral presentations** by selected participants
12:30 – 14:00	LUNCH
14:00 – 14:45	Prof. Olivier Devuyst, Institute of Physiology, University of Zurich, Switzerland **Genetic architecture driving therapeutic targets in kidney diseases**
14:45 – 15:30	Dr. Isaac Canals, Department of Experimental Medical Science, Lund University, Sweden **iPSC and genome editing as tools to generate human in vitro models of neuronopathic lysosomal storage disorders**
15:30 – 16:00	COFFEE BREAK
16:00 – 17:30	Prof. Marshall Summar, Children‘s National Medical Center, George Washington University, USA **Workshop: Designing rare disease clinical trials**
17:30 – 18:30	Free time
18:30 – 21:00	GRILL

FRIDAY, JULY 7th 2023	
07:45 – 08:45	BREAKFAST
08:45 – 09:30	**Take-home messages** by participants
09:30 – 10:15	Prof. Günter Schwarz, University of Cologne, Germany **Regulatory approval of a first treatment for the ultra-rare molybdenum cofactor deficiency**
10:15 – 10:45	COFFEE BREAK
10:45 – 11:30	Dr. Jama Nateqi, Symptoma GmbH, Austria **How to find (misdiagnosed) rare disease patients in hospitals – an AI-based approach**
11:30 – 12:15	Hansruedi Silberschmidt, patient with rare disease **Coughing prohibited – living with a rare disease**
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-Aksözen is a patient with the ultra-rare inborn error of metabolism erythropoietic protoporphyria (EPP). She studied molecular biology and biochemistry at the University of Heidelberg and obtained her PhD and Venia Legendi from the University of Zurich. Her research topics cover basic science and drug development in EPP and related diseases (porphyrias), as well as aspects concerning the regulatory assessment and benefit evaluation of orphan drugs. During her PhD, she was involved in the development of the first drug for treating EPP and later became a patient representative for the respective 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 working at the Institute for Laboratory Medicine at the Municipal Hospital Zurich, which is the Swiss Reference Center for the Porphyrias, and is currently studying health economics and methods of health technology assessment at the University of Applied Sciences in Winterthur.

DR. PAUL BASTARD

Paul Bastard, MD-PhD, is currently working as a fellow in the Pediatric Hematology and Immunology department of Necker Hospital for Sick Children (AP-HP, Paris, France), while also doing research in the Necker branch of the laboratory of Jean-Laurent Casanova, which is located at the Imagine Institute (University of Paris and INSERM) and the Rockefeller University (New York, USA). His research currently focuses on the genetic and immunological determinants of severe viral diseases, including the causes and consequences of autoantibodies against type I interferons.

PROF. ELVIR BECIROVIC

Elvir Becirovic studied biology at the University of Cologne from 2001-2006. He received his PhD at the Ludwig-Maximilians-University of Munich in 2010. He then taught and conducted research there as a postdoctoral fellow and, since 2014, as a junior research group leader on the pathophysiology of retinal diseases. In 2016, he was enrolled as a postdoctoral fellow and completed the habilitation process in 2019. Since 2016, his research focus has been on gene therapy of retinal diseases. In June 2022, he was appointed Assistant Professor of Experimental and Translational Ophthalmology at the University of Zurich. In addition to his academic activities, he works as Chief Technology Advisor for ViGeneron GmbH and has several patents as lead inventor.

DR. ISAAC CANALS

Isaac Canals studied biology at University of Barcelona and graduated in 2006. He received his PhD in Genetics in 2015 by the University of Barcelona on a work on Sanfilippo syndrome, a rare lysosomal storage disorder in which he generated novel iPSC-derived cellular models. During his postdoctoral training at Lund University, he specialized in neuroscience and develop a method to differentiate iPSCs towards astrocytes, technology that he combined with genome editing and methods to generate neurons for modeling neurodegenerative disorders. He established his own research group in 2021 at Lund University and will be moving to the University Children's Hospital of Zurich in 2023. The aim of his research is to generate different iPSC-based 2D and 3D in vitro models to understand disease mechanisms of lysosomal storage disorders, the contribution of different brain cells to the pathology and what are the neurodevelopmental impairments in patients, as well as to develop new therapeutic approaches based on genome editing technologies.

PROF. OLIVIER DEVUYST

Olivier Devuyst, M.D., Ph.D., graduated from UCLouvain Medical School in Brussels (Belgium) and trained at the Technion Institute (Haifa, Israel) and the Johns Hopkins Medical School (Baltimore, USA). He is Full Professor at the University of Zurich, where he co-chairs the University priority program on rare diseases and has a joint clinical appointment at Saint-Luc Academic Hospital, UCLouvain. Devuyst and his group use a multi-level approach to investigate the mechanisms of epithelial transport and the genetic architecture of kidney diseases, paving the way for novel therapeutic approaches. In parallel, they demonstrated fundamental mechanisms of osmosis and the crucial role of water channels (aquaporins) in dialysis. O. Devuyst has published more than 300 original studies, cited >43,000 times (H-index, 93). He served as President and Board Member in the Belgian and Swiss societies of nephrology, coordinated several EU-funded research networks and established the Working Group on Inherited Kidney Disorders of the European Renal Association and the Swiss Society for Nephrology.

PROF. 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.

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.

DR. JAMA NATEQI

Jama Nateqi is a medical doctor and founder of Symptoma.com, a digital health assistant. Patients and doctors enter symptoms, answer questions and receive a list of possible causes – sorted by probability. Symptoma is the most used symptom checker with 10 million users/month. Jama founded his first company in 1999, studied medicine in Austria and performed research for his thesis at the Yale University. In 2020 he was named Austrian of the Year for his 16 years of research at Symptoma.

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 a 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.

PROF. GÜNTER SCHWARZ

Günter Schwarz graduated his studies in Plant Biology and Biochemistry and habilitated *Venia legendi* in Biochemistry and Molecular Biology at the Technical University of Braunschweig (TU), Germany.

He is full Professor (W3) for Biochemistry at the University of Cologne, Germany. He and his team are interested to understand protein structure-function relations in different biological processes ranging from basic metabolism to molecular neuroscience. Their research is based on a variety of methods including molecular biology, protein biochemistry, enzymology, structural biology (protein crystallography), cell biology, bioinorganic chemistry and biotechnology.

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.

HANSRUEDI SILBERSCHMIDT

Hansruedi Silberschmidt is a patient with the primary ciliary dyskinesia (PCD). From the first day of his life he had health problems (several respiratory arrests, infections). Many hospital stays followed during his childhood. In his autobiography «husten verboten» (coughing prohibited), he describes his life as a child and young adult with an undiagnosed disease and his path of suffering until he was finally diagnosed at the age of 39 at the University Hospital in Zurich.

He completed the teacher training and studied psychology at the IAP, Zurich. He worked as the executive director of andante Winterthur - a non-profit, social enterprise for the benefit of people with a cognitive and/or cerebral impairment as well as for people with a brain injury.

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 in Washington. 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.