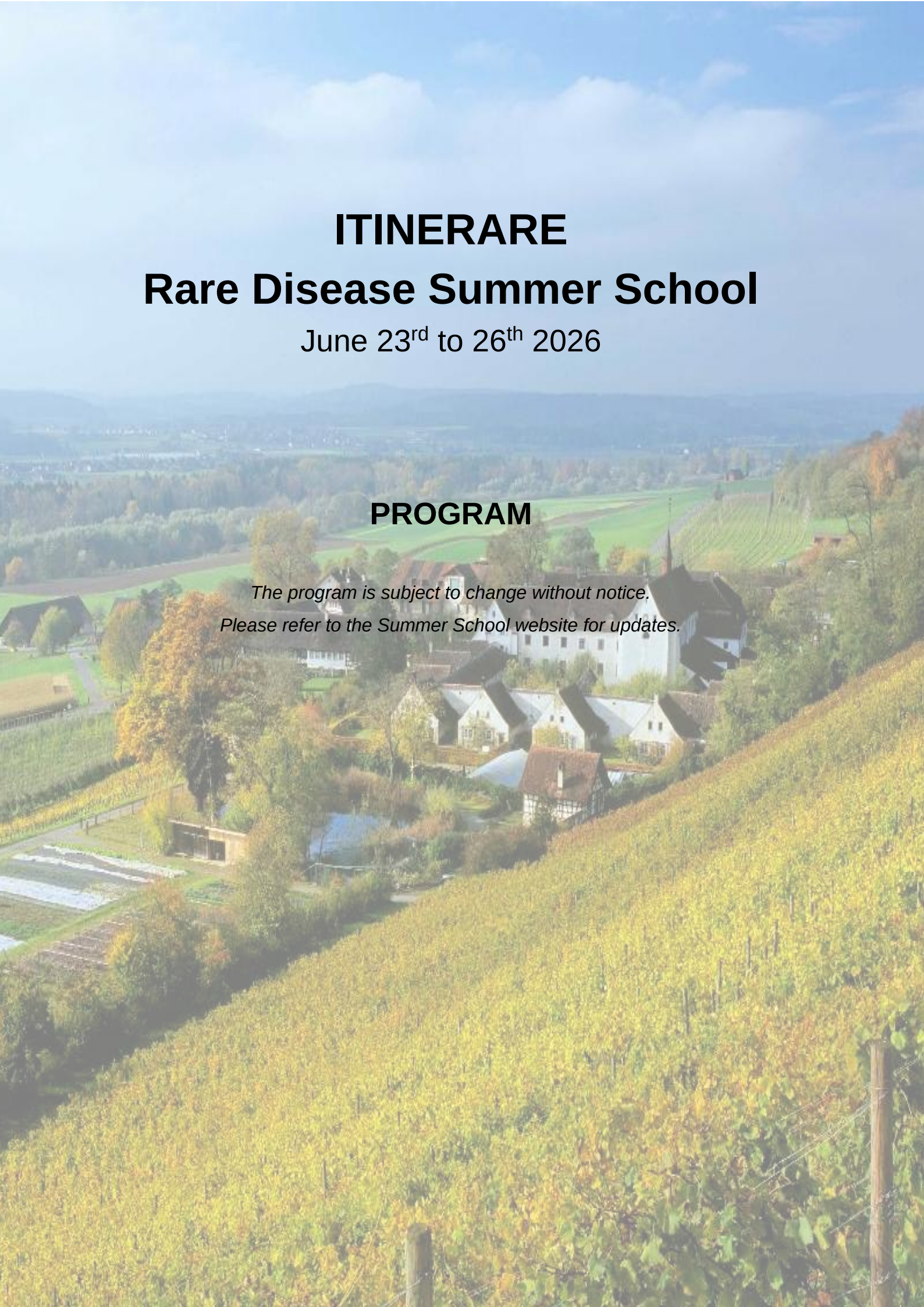

ITINERARE
Rare Disease Summer School

June 23rd to 26th 2026

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.

TUESDAY, JUNE 23rd 2026

13:30 – 14:00	Arrival of the participants and registration
14:00 – 14:30	**Welcome and introduction round** Prof. Matthias Baumgartner, Co-director ITINERARE and Director Research & Education University Children's Hospital Zurich
14:30 – 15:15	**Lecture: Latest developments in genome sequencing technologies: impact for rare diseases** Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands
15:15 – 15:45	**COFFEE BREAK**
15:45 – 16:30	**Lecture: Let the sunshine in! Development, approval and benefit assessment of afamelanotide, the first drug for treating erythropoietic protoporphyria** Dr. Jasmin Barman-Aksözen, Scientific advisor, Swiss society for porphyria, Switzerland
16:30 – 17:30	**Oral presentations** by selected participants
17:30 – 18:30	Check-in time
18:30 – 20:15	**DINNER**
20:15 – 21:30	Poster viewing

WEDNESDAY, JUNE 24th 2026

07:45 – 8:45	**BREAKFAST**
08:45 – 9:30	**Lecture: Regulatory framework for ATMPs on the basis of a case study** Dr. Fabienne Hartmann-Fritsch, CUTISS AG, Switzerland
09:30 – 10:30	**Oral presentations** by selected participants
10:30 – 11:00	**COFFEE BREAK**
11:00 – 12:30	**Workshop: Next generation sequencing** Prof. Nine Knoers, Department of Genetics, University Medical Center Groningen, the Netherlands
12:30 – 14:00	**LUNCH**
14:00 – 14:45	**Lecture: Ethics of Genetic Testing and Treatments of Genetic Conditions** Kelly Ormond, Health Ethics and Policy Lab, D-Health Sciences and Technology, ETH Zurich, Switzerland
14:45 – 15:30	**Lecture: Rare diseases today** Prof. Marshall Summar, Children's National Medical Center, George Washington University / CEO: Uncommon Cures, USA
15:30 – 16:00	**COFFEE BREAK**
16:00 – 16:45	**Keynote Lecture:** Prof. Shamima Rahman, UCL Great Ormond Street Institute of Child Health, UK
16:45 – 18:30	Free time
18:30 – 20:15	**DINNER**
20:15 – 21:30	Poster viewing

THURSDAY, JUNE 25th 2026

07:45 – 8:45	**BREAKFAST**
08:45 – 10:45	**Workshop: Justice, Visibility, and Responsibility: Ethical and Societal Perspectives on Rare Diseases across Research, Care, and Lived Experience** Dr. Sebastian Wäscher, Center for Ethics, University of Zurich, Switzerland
10:45 – 11:15	**COFFEE BREAK**
11:15 – 12:00	**Lecture: Studying rare diseases in the (not so rare) Zebrafish model organism** Prof. Stephan Neuhauss, Department of Molecular Science, University of Zurich, Switzerland
12:00 – 14:00	**LUNCH**
14:00 – 14:45	**Lecture: Genetic architecture driving therapeutic targets in kidney diseases** Prof. Olivier Devuyst, Institute of Physiology, University of Zurich, Switzerland
14:45 – 15:30	**Lecture: Co-Creating with patients to advance science and care for rare diseases** Prof. Andrew Dwyer, Boston College, USA
15:30 – 16:00	**COFFEE BREAK**
16:00 – 17:30	**Workshop: Designing rare disease clinical trials** Prof. Marshall Summar, Children‘s National Medical Center, George Washington University / CEO: Uncommon Cures, USA
17:30 – 18:30	Free time
18:30 – 21:00	**BARBECUE**

FRIDAY, JUNE 26th 2026

07:45 – 8:45	**BREAKFAST**
08:45 – 09:30	**Take-home messages** by participants
09:30 – 10:15	**Lecture: From neurodegeneration in ultra rare sulfite oxidase deficiency to the treatment of kidney diseases** Prof. Günter Schwarz, Department of Biochemistry, University of Cologne, Germany
10:15 – 10:45	**COFFEE BREAK**
10:45 – 11:30	**Lecture: iPSC and genome editing as tools to generate human in vitro models of neuronopathic lysosomal storage disorders** Prof. Isaac Canals, University Children's Hospital Zurich, Switzerland
11:30 – 12:15	**Lecture: Coughing prohibited – living with a rare disease** Hansruedi Silberschmidt, patient 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 BIOGRAPHIES

Dr. Jasmin Barman-Aksözen

Scientific advisor, Swiss society for porphyria, Switzerland

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.

Prof. Isaac Canals

University Children's Hospital Zurich, Switzerland

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. In 2023 he joined the University Children's Hospital of Zurich as an Assistant Professor. 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

Institute of Physiology, University of Zurich, Switzerland

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 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 >48,000 times. 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 Groups on Inherited Kidney Disorders of the European Renal Association and the Swiss Society for Nephrology.

Prof. Andrew Dwyer

Connell School of Nursing, Boston College, USA

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

CUTISS AG, Switzerland

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

Department of Genetics, University Medical Center Groningen, the Netherlands

Nine Knoers was the chair of the Department of Genetics at the University Medical Centre Groningen (UMCG), The Netherlands (2018-2024). 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 310 scientific papers and in the last 10 years delivered >270 invited lectures. In September 2024, she officially retired, and since then works part-time on several managerial and research projects for the UMCG, the University of Groningen, the Dutch scientific organization (NWO), and several (inter) national research foundations.

Prof. Stephan Neuhauss

Department of Molecular Science, University of Zurich, Switzerland

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 and Planning at the Faculty of Science and an editor of various open access journals.

Kelly Ormond

Department of Health Sciences and Technology, ETH-Zurich, Switzerland

Kelly Ormond is a genetic counselor (US ABGC certified and EBMG registered) and ELSI researcher. She received her MS in Genetic Counseling from Northwestern University (1994) and a post-graduate certificate in Clinical Medical Ethics from the MacLean Center at the University of Chicago (2001). She joined the Health Ethics and Policy Lab as a Senior Scientist in February 2021, and is an Adjunct Professor in the Department of Genetics at Stanford School of Medicine, Stanford University, California, USA . Prior to joining ETH, Kelly practiced as a clinical genetic counselor in the United States for >25 years, gaining experience in reproductive genetics, teratology, pediatric/medical genetics, adult neurogenetics, cancer genetics and primary care applications of genetic testing. Kelly's recent research focuses on ethical issues related to genetics and personalized medicine, including patient decision making, the role of uncertainty in decision making, consent and disclosure of genetic test results, gene editing, and the interface between genetics and disability.

Prof. Shamima Rhaman

University College London, Great Ormond Street Institute of Child Health, UK

Prof. Günter Schwarz

Department of Biochemistry, University of Cologne, Germany

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.

Hansruedi Silberschmidt

Patient with a rare disease

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

Children‘s National Medical Center, George Washington University / CEO: Uncommon Cures, USA

Marshall Summar is the CEO of Uncommon Cures and Professor of Pediatrics George Washington University. 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.

Dr. Sebastian Wäscher

Center for Ethics, University of Zurich, Switzerland

Sebastian Wäscher, PhD, is a researcher in bioethics with a background in communication science. He studied communication science, sociology and philosophy at the University of Münster, Germany, and obtained his PhD from the University of Zurich. He is affiliated with the Institute for Social Ethics at the University of Zurich and is a member of the ITINERARE research consortium.

His work focuses on ethical and societal questions in the context of medicine and science, with a particular emphasis on rare diseases. His research combines qualitative social science methods with normative analysis, aiming to better understand patient perspectives and to contribute to ethically informed healthcare practices and policy discussions.

Sebastian Wäscher is responsible for the development and coordination of the Ethics and Society Platform within the ITINERARE consortium. The platform provides a forum for structured exchange on ongoing research activities and supports the integration of ethical and societal perspectives into interdisciplinary research. In this role, he facilitates dialogue across disciplinary boundaries and contributes to embedding reflection on ethical and social implications within collaborative research processes.