



University of  
Zurich<sup>UZH</sup>

URPP ITINERARE - Innovative Therapies in Rare Diseases



**ITINERARE**  
*Rare Disease  
Summer School*  
Zurich - Switzerland



# About the Summer School

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8<sup>th</sup> Rare Disease Summer School  
July 13<sup>th</sup> to 15<sup>th</sup>, 2022  
Kartause Ittingen, Warth

The ITINERARE Rare Disease Summer School will cover a broad spectrum of subjects in the field of rare diseases. Participants will engage with experts on topics ranging from disease mechanisms and animal models to improving diagnoses and developing novel therapies. Moreover, they will address the ethical, social, and legal (ELSI) challenges associated with rare diseases.

The high interdisciplinary program builds on a long tradition established by radiz, the Rare Disease Initiative Zurich, and consists of lectures and workshops given by national and international rare disease experts. Focus will be given to drug development, model organisms, how to choose clinical endpoints, clinical trials, regulatory aspects, patient registries, patient-initiated research, and ELSI considerations.

All participants will have the chance to present their own research projects during dedicated poster sessions and authors of selected abstracts will give a short oral presentation about their work. There will be ample time for discussions, teamwork, as well as for socializing.

This summer school lasts two and a half days and is open to clinicians, physician-scientists, postdocs, and PhD students from various disciplines such as medicine, biology, psychology, sociology, and related fields. Attendance is restricted to 30 people to allow for extensive interaction with the experts, enabling participants to optimally benefit from the varied expertise of the faculty.

## Scientific organizing committee

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Prof. Matthias Baumgartner  
Prof. Nikola Biller-Andorno  
Prof. Olivier Devuyst  
Prof. Janine Reichenbach  
Prof. Brigitte Tag

# Confirmed speakers

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## Lectures

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### **Let the sunshine in! Getting a drug approved for the rare genetic light intolerance erythropoietic protoporphyria**

Dr. Jasmin Barman-Aksözen, Swiss society for porphyria, Switzerland

### **Combined cell and gene therapy for Epidermolysis Bullosa**

Prof. Michele De Luca, Modena and Reggio Emilia University, Italy

### **Person-centered approaches for bridging health disparities for rare disease patients**

Prof. Andrew Dwyer, Boston College, USA

### **Regulatory framework for ATMPs on the basis of a case study**

Dr. Fabienne Hartmann, CUTISS AG, Switzerland

### **Using AI for rare diseases diagnosis: my personal journey**

Mr. Julian Isla, Dravet Syndrome Foundation, Spain

### **New nerve cells for old brains**

Prof. Sebastian Jessberger, University of Zurich, Switzerland

### **Towards better and humane science: The 3Rs principle**

Dr. Paulina Jirkof, University of Zurich, Switzerland

### **Latest developments in exome and genome sequencing: impact for rare diseases**

Prof. Nine Knoers, University Medical Center Groningen, the Netherlands

### **Studying rare diseases in the (not so rare) Zebrafish model organism**

Prof. Stephan Neuhauss, University of Zurich, Switzerland

### **Designing rare disease clinical trials: outcomes, sample size, natural history, and recruitment**

Prof. Marshall Summar, Children's National Medical Center, USA

## Workshops

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### **Ethical considerations (case study)**

Dr. Jürg Streuli, University of Zurich, Switzerland

### **So you want to develop a rare disease drug**

Prof. Marshall Summar, Children's National Medical Center, USA

### **Next generation sequencing**

Prof. Nine Knoers, University Medical Center Groningen, the Netherlands

## More Information on the Summer School

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[www.itinerare.uzh.ch](http://www.itinerare.uzh.ch)

ITINERARE - Innovative Therapies in Rare Diseases – is a University Research Priority Program (URPP) of the University of Zurich. Researchers and clinicians from the University of Zurich, Children’s Hospital Zurich, and the University Hospital Zurich have joined forces in this program. Through its interdisciplinary and translational research projects, the URPP ITINERARE aims to develop novel molecular and gene therapies for selected genetic disorders and to establish a broad ethical-legal and educational framework of multidisciplinary expertise to address associated societal issues.



Location: Kartause Ittingen, Warth (Canton Thurgau)

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University of Zurich  
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