



**Universität
Zürich^{UZH}**

1st Rare Diseases Summer School

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

Tagungszentrum Schloss Au,
Wädenswil (on Lake Zurich), Switzerland
Thursday, July 4th to Saturday, July 6th 2013

Program

Thursday, July 4th 2013

12:30 – 13:30 registration

13:30 – 13:35 Welcome by Prof. Matthias Baumgartner, University Children's Hospital Zurich and University of Zurich, lead radiz

Session chairs: Prof. Olivier Devuyst, University of Zurich and University Hospital Zurich and Prof. Matthias Baumgartner, University Children's Hospital Zurich and University of Zurich

13:35 – 14:35 Prof. Harry C. Dietz, Johns Hopkins University School of Medicine, Baltimore, USA

Keynote lecture: Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders

14:35 – 15:05 Samantha Parker, Orphan Europe Recordati Group, France
Lecture: Global registries for rare diseases: challenges and solutions

15:05 – 15:30 coffee break

15:30 – 16:00 PD Dr. Gabriela Senti, University Hospital Zurich and Dr. med. Andrea Klein, University Children's Hospital Zurich

Lecture: Clinical Trials for Rare Diseases

16:00 – 16:30 Prof. Nine Knoers, University Medical Centre Utrecht, The Netherlands

Lecture: Next Generation Sequencing in rare disorders: scientific impact and clinical utility

16:30 – 16:45 short break

16:45 – 18:15 **workshop session 1**

- 1A: Rare disease registries, led by Samantha Parker
radiz: Prof. Matthias Baumgartner
- 1B: Exome sequencing, led by Prof. Nine Knoers
radiz: Prof. Anita Rauch
- 1C: Good Clinical Practice, led by PD Dr. Gabriela Senti
radiz: Prof. Olivier Devuyst

18:15- 19:30 Break for socializing and swimming

19:30 – ~22:00 dinner in Au

Friday, July 5th 2013

Session chairs: Prof. Anita Rauch, University of Zurich and Prof. Hanns Ulrich Zeilhofer, University of Zurich

8:45 – 9:15 Prof. Stephan Neuhauss, University of Zurich

Lecture: Studying rare diseases in (not so rare anymore) the Zebrafish model organism

9:15 – 9:45 Prof. Eckhard Wolf, Ludwig-Maximilians-Universität München, Germany

Lecture: Genetically engineered pig models for human monogenetic diseases

9:45 – 10:15 coffee break

10:15 – 10:45 Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA

Lecture: Clinical Endpoints: Selecting Outcomes and Surrogate Markers in Rare Diseases

10:45 – 11:15 Dr. Laurie Chicha, University of Basel

Lecture: Pluripotent stem cells to model rare diseases

11:15 – 11:45 PD Dr. Janine Reichenbach, University Children's Hospital Zurich

Lecture: Clinical gene therapy for X-linked Chronic Granulomatous Disease

12:00 – 13:30 Lunch, Posters and Networking

Session chairs: Prof. Barbara Plecko, University Children's Hospital Zurich and University of Zurich and Prof. Olivier Devuyst, University of Zurich and University Hospital Zurich

13:30 -14:00 Christina Fasser, Vice-President ProRaris, CEO Retina Suisse, President Retina International

Lecture: Patient-initiated research

14:00 – 14:40 Dr. Effy Vayena, University of Zurich

Lecture and discussion: Ethical considerations

14:40 – 15:10 coffee break

15:10 – 16:40 **workshop session 2**

- 2A: Drug Development, led by Prof. Marshall Summar
radiz: PD Dr. Thorsten Hornemann
- 2B. Ethical considerations, led by Dr. Effy Vayena
radiz: Prof. Barbara Plecko
- 2C: Model Organisms for Rare Diseases, led by Prof. Eckhard Wolf and Prof. Stephan Neuhaus
radiz: Prof. Hanns Ulrich Zeilhofer

16:45 – 17:30 **Preparation of presentations of workshop results** for Saturday

17:30 – ~22:00 Evening out: boat trip and guided tour of Rapperswil

Saturday, July 6th 2013

Session chairs: Prof. Matthias Baumgartner, University Children's Hospital Zurich and University of Zurich, lead radiz and Prof. Barbara Plecko, University Children's Hospital Zurich and University of Zurich

9:15 – 10:45 **Presentations of workshop results by participants**

10:45 – 11:15 coffee break

11:15 – 11:45 Dr. Urs Widmer, Swiss Re, Zurich

Lecture: Orphan Drugs: Legislation and Incentives for Access to New Therapies - Case studies

11:45 – 12:30 Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA

Lecture: From rare diseases to common diseases: Lessons from the Urea Cycle

Concluding remarks and award of the poster prize

Prof. Olivier Devuyst, University of Zurich and University Hospital Zurich