



Universität
Zürich^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

2nd Rare Diseases Summer School

Tagungszentrum Schloss Au,

Wädenswil (on Lake Zurich), Switzerland

Monday, July 14th to Wednesday, July 16th 2014

Program

Monday, July 14th 2014

13:00 – 14:00 registration (**no** lunch provided)

14:00 – 14:10 **Welcome** by Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz

Session chair Prof. Giatgen Spinas, University Hospital Zurich and University of Zurich

14:10 – 14:50 Prof. Leena Bruckner-Tuderman, University Medical Center, Albert-Ludwigs-University of Freiburg, Germany

Keynote lecture: Skin fragility - from genes to therapies in twenty years

14:50 – 15:00 questions

15:00 – 15:20 Prof. Nine Knoers, University Medical Centre Utrecht, The Netherlands

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

15:20 – 15:30 discussion

15:30 – 16:00 coffee break

16:00 – 17:00 Dr. Effy Vayena, University of Zurich

Interactive workshop: Ethical considerations (case study)



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17:15 – 18:45 **workshop session 1**

- **1A: Registries for rare diseases: challenges and solutions**, led by Samantha Parker, Orphan Europe Recordati Group, France
radiz: Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich
- **1B: Next generation sequencing**, led by Prof. Nine Knoers
radiz: Prof. Anita Rauch, University of Zurich
- **1C: A clinical trial primer**, led by Dr. Sandro Studer, University Hospital Zurich
radiz: Prof. Olivier Devuyst, University of Zurich and University Hospital Zurich

19:00 – ~22:00 **networking dinner** in Au with speakers

Tuesday, July 15th 2014

Session chair: Prof. Olivier Devuyst, University Hospital Zurich and University of Zurich

8:45 – 9:05 Prof. Stephan Neuhauss, University of Zurich

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

9:05 – 9:15 discussion

9:15 – 9:35 Prof. Carsten Wagner, University of Zurich

Lecture: Possibilities and limitations of mouse models for rare disease research

9:35 – 9:45 discussion

9:45 – 10:15 coffee break

10:15 – 10:35 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

10:35 – 10:45 discussion



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10:45 – 10:55 Dr. Christiane Stieber, Zentrum für seltene Erkrankungen Bonn, University Hospital of Bonn, Germany

Short presentation: InterPoD – an initiative for undiagnosed patients

11:10 – 12:10 **poster session**

12:10 – 12:45 Lunch

12:45 – 13:45 **poster session**

Session chair: Prof. Arnold von Eckardstein, University Hospital Zurich and University of Zurich

14:00 – 14:20 Dr. Tsveta Schyns - Liharska, Founder and Secretary of ENRAH, Brussels, Belgium, Consulting Scientists and Member of PDCO, EMA

Lecture: Patient-initiated research

14:20 – 14:30 discussion

14:30 – 14:50 Dr. Martine Zimmermann, Alexion Pharma International

Lecture: Regulatory frameworks and incentives for development of Orphan Medicinal Products

14:50 – 15:00 discussion

15:00 – 15:30 coffee break

15:30 – 17:00 **workshop session 2**

- **2A: Navigating the regulatory landscape** led by Dr. Martine Zimmermann and Cécile De Coster, Alexion Pharma International
radiz: Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich
- **2B: Drug development**, led by Prof. Marshall Summar
radiz: Prof. Arnold von Eckardstein, University Hospital Zurich and University of Zurich



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- **2C: Model organisms for rare diseases**, led by Prof. Carsten Wagner and Prof. Stephan Neuhauss
radiz: Prof. Barbara Plecko, Children's Hospital Zurich and University of Zurich

17:00 – 18:00 **Preparation of workshop results by participants** to be presented on Wednesday (workshops from sessions 1 and 2)

18:00 – 19:00 Break for socializing and swimming in the lake

19:00 – ~22:00 **Evening out**: dinner and boat trip to Zurich, guided tour of Zurich

Wednesday, July 16th 2014

Session chair: Prof. Anita Rauch, University of Zurich

8:45 – 9:05 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

9:05 – 9:15 discussion

9:15 – 9:35 Prof. Janine Reichenbach, Children's Hospital Zurich

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

9:35 – 9:45 discussion

9:45 – 10:15 coffee break

10:15 – 12:15 **Presentations of workshop results by participants**

Concluding remarks and award of the poster prize

Prof. Barbara Plecko, Children's Hospital Zurich and University of Zurich