



Universität
Zürich^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

3rd Rare Diseases Summer School

Tagungszentrum Schloss Au

Wädenswil (on Lake Zurich), Switzerland

Wednesday, July 1st to Friday, July 3rd 2015

Final Program

Wednesday, July 1st 2015

- | | |
|---|--|
| 10:00 – 10:45 | Registration |
| 10:45 – 11:00 | Welcome by Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz |
| Session chair: Prof. Anita Rauch, University of Zurich | |
| 11:00 – 11:20 | Prof. Nine Knoers, University Medical Centre Utrecht, the Netherlands Lecture: Next Generation Sequencing in rare disorders: scientific impact and clinical utility |
| 11:20 – 11:30 | Discussion |
| 11:30 – 11:50 | 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 |
| 11:50 – 12:00 | Discussion |
| 12:00 | Lunch and Poster Viewing |



radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

Session chair: Prof. Giatgen Spinas, UniversityHospital Zurich and University of Zurich

13:15 – 14:00 Poster Presentations

14:00 – 15:30 **Workshop Session 1**

- **1A: Registries for rare diseases: challenges and solutions**
led by Samantha Parker, Lysogene, France
- **1B: Navigating the regulatory landscape**
led by Dr. Martine Zimmermann, Alexion Pharma International
- **1C: A clinical trial primer**
led by Dr. Sandro Studer, UniversityHospital Zurich

15:30 Check-in accommodation/Swimming in the lake

17:45 – 18:25 Prof. Gert-Jan van Ommen, Professor emeritus. Department of Human Genetics, Leiden University Medical Center, Leiden, The Netherlands
Keynote Lecture: Therapy development for DMD: ups and downs...and up again

18:25 – 18:40 Discussion

19:00 – 22:00 Networking dinner at the Restaurant Landgasthof Halbinsel AU

Thursday, July 2nd 2015

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

08:45 – 09:05 Prof. Carsten Wagner, University of Zurich

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

09:05 – 09:15 Discussion



radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

09:15 – 09:35 Prof. Eckhard Wolf, Gene Center, Ludwig-Maximilians-University
Munich, Germany

Lecture: Genetically engineered pig models for human monogenic diseases

09:35 – 09:45 Discussion

09:45 – 10:05 Prof. André Brändli, Ludwig-Maximilians-University Munich, Germany

Lecture: Engineering Xenopus models of rare inherited diseases for in vivo drug discovery

10:05 – 10:15 Discussion

10:15 Coffee Break

10:45 – 11:05 Christina Fasser, ProRaris, Retina International

Lecture: Patient-initiated research

11:05 – 11:15 Discussion

11:15 – 11:35 Prof. Adriano Aguzzi, UniversityHospital Zurich

Lecture: Prion diseases

11:35 – 11:45 Discussion

11:45 Lunch and Poster Viewing

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

13:15 – 14:00 Poster Presentations

14:00 – 15:00 Dr. Effy Vayena, University of Zurich

Interactive workshop: Ethical considerations (case study)

15:00 Coffee Break



radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

- 15:30 – 17:00 **Workshop Session 2**
- **2A: Model organisms for rare diseases**
led by Prof. Carsten Wagner and Prof. Eckhard Wolf
 - **2B: Drug development**
led by Prof. Marshall Summar
 - **2C: Next generation sequencing**
led by Prof. Nine Knoers
- 17:00 – 18:00 Preparation of workshop results by participants
- 18:00 – 23:00 Visit at the Monastery of Einsiedeln followed by dinner

Friday, July 3rd 2015

Session chair: **Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich**

- 08:45 – 09:05 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
- 09:05 – 09:15 Discussion
- 09:15 – 09:35 Prof. Janine Reichenbach, Children's Hospital Zurich
Lecture: Clinical gene therapy for X-linked Chronic Granulomatous Disease
- 09:35 – 09:45 Discussion
- 09:45** **Coffee Break**
- 10:15 – 12:15 Presentations of workshop results by participants
Concluding remarks and award of poster prize
- 12:30** **End of the Summer School and departure of the participants**