



Universität
Zürich^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

4th Rare Diseases Summer School

Tagungszentrum Schloss Au

Wädenswil (on Lake Zurich), Switzerland

Wednesday, July 13th to Friday, July 15th 2016

Program

Wednesday, July 13th 2016

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|-----------------------|--|
| 09:30 – 10:15 | Registration |
| 10:15 – 10:30 | Welcome by Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz |
| Session Chair: | Prof. Anita Rauch, University of Zurich |
| 10:30 – 11:00 | Prof. Nine Knoers, University Medical Centre Utrecht, the Netherlands
Lecture: Next Generation Sequencing in rare disorders: scientific impact and clinical utility |
| 11:00 – 11:10 | Discussion |
| 11:10 – 11:40 | 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 |
| 11:40 – 11:50 | Discussion |
| 11:50 - 12:45 | Lunch |
| 12:45 – 13:45 | Poster Viewing |
| 13:45 – 14:30 | Poster Presentations by selected participants |
| 14:30 – 15:30 | Dr. Martine Zimmermann, Alexion Pharma International
Plenary workshop: Regulatory Frameworks |
| 15:30 | Check-in accommodation/Swimming in the lake |



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

- 17:45 – 18:25 Dr. Cynthia J. Tifft, Director, Pediatric Undiagnosed Diseases Program
National Human Genome Research Institute/NIH
Keynote Lecture: From Known into the Unknown: Journey of a Physician Scientist
- 18:25 – 18:40 Discussion
- 19:00 – 22:00 Networking dinner at the Restaurant Landgasthof Halbinsel Au

Thursday, July 14th 2016

Session Chair: Giagten Spinas, University Hospital Zurich and University of Zurich

- 08:45 – 09:05 Prof. Stephan Neuhaus, University of Zurich
Lecture: Studying rare diseases in the (not so rare) Zebrafish model organism
- 09:05 – 09:15 Discussion
- 09:15 – 09:35 Prof. Carsten Wagner, University of Zurich
Lecture: Possibilities and limitations of mouse models for rare disease research
- 09:35 – 09:45 Discussion
- 09:45 – 10:05 Prof. Janine Reichenbach, Children's Hospital Zurich
Lecture: Clinical gene therapy for X-linked Chronic Granulomatous Disease
- 10:05 – 10:15 Discussion
- 10:15 Coffee Break**
- 10:45 – 12:15 Dr. Effy Vayena, University of Zurich
Interactive workshop: Ethical considerations (case study)
- 12:15 - 13:00 Lunch**
- 13:00 – 13:45 Poster Viewing



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13:45 – 15:15

Workshop Session

- **A: Registries for rare diseases: challenges and solutions**
led by Samantha Parker, Lysogene, France
- **B: Drug development**
led by Prof. Marshall Summar
- **C: Next generation sequencing**
led by Prof. Nine Knoers

15:15

Coffee Break

Session Chair:

Prof. Dr. Thorsten Hornemann, University of Zürich

15:45 – 16:30

Poster Presentations by selected participants

16:30 – 16:50

Dr. Jasmin Barman-Aksözen, Swiss society for porphyria

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

16:50 – 17:00

Discussion

17:00 – 17:20

Prof. Guenter Schwarz, Cologne University

Lecture: Molybdenum Enzyme Deficiency: From cofactor biogenesis to drug development in ultra rare and common diseases

17:20 – 17:30

Discussion

17:30 – 18:15

Preparation of workshop results by participants

18:30 – 20:00

Apéro at the Weinbaumuseum



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Friday, July 15th 2015

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

08:45 – 09:45 Presentations of workshop results by participants
Award of poster prize

09:45 – 10:15 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

10:15 – 10:25 Discussion

10:30 Coffee Break

11:00 – 11:20 Christina Fasser, Vice-President ProRaris, President Retina International
Lecture: Patient-initiated research

11:20 – 11:30 Discussion

11:30 – 11:50 Concluding remarks

12:00 End of the Summer School and departure of the participants