



Universität  
Zürich<sup>UZH</sup>

# radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

**Draft Program (subject to change)**

## 4<sup>th</sup> Rare Diseases Summer School

Tagungszentrum Schloss Au

Wädenswil (on Lake Zurich), Switzerland

Wednesday, July 13<sup>th</sup> to Friday, July 15<sup>th</sup> 2016

### Program

#### Wednesday, July 13<sup>th</sup> 2016

09:30 – 10:15	Registration
10:15 – 10:30	<b>Welcome</b> by Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz
10:30 – 11:00	Prof. Nine Knoers, University Medical Centre Utrecht, the Netherlands <b>Lecture: Next Generation Sequencing in rare disorders: scientific impact and clinical utility</b>
11:00 – 11:10	Discussion
11:10 – 11:40	Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA <b>Lecture: Clinical endpoints: selecting outcomes and surrogate markers in rare diseases</b>
11:40 – 11:50	Discussion
<b>11:50 - 12:45</b>	<b>Lunch</b>
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 <b>Plenary workshop: Regulatory Frameworks</b>



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- 15:30**                      **Check-in accommodation/Swimming in the lake**
- 17:45 – 18:25              Dr. Cynthia J. Tifft, Director, Pediatric Undiagnosed Diseases Program  
National Human Genome Research Institute/NIH
- Keynote Lecture**
- 18:25 – 18:40              Discussion
- 19:00 – 22:00              Networking dinner at the Restaurant Landgasthof Halbinsel Au

## Thursday, July 14<sup>th</sup> 2016

- 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

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

## Friday, July 15<sup>th</sup> 2015

08:45 – 10:45

Presentations of workshop results by participants

Award of poster prize

**10:45**

## Coffee Break



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- 10:45 – 11:15 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**
- 11:15 – 11:25 Discussion
- 11:25 – 11:45 Christina Fasser, Vice-President ProRaris, President Retina International  
**Lecture: Patient-initiated research**
- 11:45 – 11:55 Discussion
- 11:55 – 12:15 Concluding remarks
- 12:30 End of the Summer School and departure of the participants**