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

09:30 - 10:15 Registration
10:15 - 10:30 Welcome 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
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
radiz - Rare Disease Initiative Zürich
Clinical Research Priority Program for Rare Diseases University of Zurich

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 14th 2016

08:45 - 09:05 Prof. Stephan Neuhauss, 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

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**Friday, July 15th 2015**

08:45 - 10:45  
Presentations of workshop results by participants  
Award of poster prize

**10:45**  
**Coffee Break**
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