



Universität
Zürich ^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

5th Rare Diseases Summer School

Tagungszentrum Schloss Au, Wädenswil

Wednesday, June 7th to Friday, June 9th 2017

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.





Wednesday, June 7, 2017

09:30 – 10:15	Registration
10:15 – 10:30	Welcome , Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz <i>Session chair: tbd</i>
10:30 – 11:10	Prof. Nine Knoers, University Medical Centre Utrecht, the Netherlands Next generation sequencing in rare disorders: scientific impact and clinical utility <i>Discussion</i>
11:10 – 11:50	Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA Clinical endpoints: selecting outcomes and surrogate markers in rare diseases <i>Discussion</i>
11:50 – 12:50	Lunch
13:00 – 14:30	Dr. Martine Zimmermann, Alexion Pharma International Samantha Parker, Lysogene Parallel workshops: Regulatory frameworks and registries
14:30 – 15:30	Oral presentations by selected participants
15:30 – 16:00	Coffee Break
16:00 – 17:00	Poster viewing <i>Session chair: tbd</i>
17:00 – 17:40	Prof. Guenter Schwarz, Cologne University Molybdenum enzyme deficiency: from cofactor biogenesis to drug development in ultra rare and common diseases <i>Discussion</i>
17:40 – 18:30	Prof. James R. Lupski, Baylor College of Medicine, Houston, USA Keynote Lecture: The genomics of disease: clinical genomics = personal genomes + clan genomics <i>Discussion</i>
18:45 – 21:30	Networking dinner at the Restaurant Landgasthof Halbinsel Au



Thursday, June 8, 2017

08:15 – 08:45 Morning coffee

Session chair: tbd

08:45 – 09:45 **Oral presentations** by selected participants

09:45 – 10:25 Dr. Jasmin Barman-Aksözen, Swiss society for porphyria

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

Discussion

10:30 – 11:00 Coffee Break

11:00 – 12:15 **Workshop Session**

A: **Drug development** led by Prof. Marshall Summar

B: **Next generation sequencing** led by Prof. Nine Knoers

12:15 – 13:15 Lunch

13:15 – 14:15 **Poster Viewing**

14:15 – 15:30 Dr. Jürg Streuli, University of Zurich

Interactive workshop: Ethical considerations (case study)

15:30 – 16:30 Coffee Break and **preparation of workshop results**

Session chair: tbd

16:30 – 17:00 Prof. Stephan Neuhauss, University of Zurich

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

Discussion

17:00 – 17:30 Prof. Carsten Wagner, University of Zurich

Possibilities and limitations of mouse models for rare disease research

Discussion

17:30 – 19:30 Social event



Friday, June 9, 2017

08:15 – 08:45 Morning coffee

Session chair: tbd

08:45 – 09:45 **Presentation of workshop results**

09:45 – 10:25 Prof. Janine Reichenbach, Children's Hospital Zurich

Clinical gene therapy for X-linked Chronic Granulomatous Disease

Discussion

10:30 – 11:00 Coffee Break

11:00 – 11:30 Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA

From rare diseases to common diseases: lessons from the urea cycle

Discussion

11:30 – 12:00 Christina Fasser, Vice-President ProRaris, President Retina International

Patient-initiated research

Discussion

12:00 – 12:15 Award of poster prize and concluding remarks

12:30 End of the Summer School and departure of the participants

Acknowledgment

Thank you to the Swiss Academy of Medical Science (SAMW) for sponsoring the poster prizes and ten scholarships for (MD)-PhD students.