



Universität
Zürich ^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

6th Rare Diseases Summer School

Kartause Ittingen, Warth

Wednesday, July 11th to Friday, July 13th 2018

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.





Wednesday, July 11, 2018

09:00 – 10:15	Registration
10:15 – 10:30	Welcome , Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz <i>Session chair: Prof. Matthias Baumgartner, Kispi, Zurich</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	Small group lectures on regulatory frameworks and registries Dr. Martine Zimmermann, Alexion Pharma International Samantha Parker, Lysogene
14:30 – 15:30	Oral presentations by selected participants
15:30 – 16:00	Coffee Break
16:00 – 17:00	Poster viewing <i>Session chair: Prof. Thorsten Hornemann, UZH, Zurich</i>
17:00 – 17:40	Prof. Andrew Dwyer, Boston College, USA Translating rare disease research into practice using patient partnerships <i>Discussion</i>
17:40 – 18:30	Dr. Gisèle Bonne, Institute of Miology, Paris Keynote lecture - Laminopathy of striated muscle: from gene identification to pathophysiological mechanisms <i>Discussion</i>
18:45 – 21:30	Networking dinner at the Ittingen Kartause



Thursday, July 12, 2018

07:45 – 08:45 Breakfast

Session chair: Prof. Giatgen Spinas, USZ, Zurich

08:45 – 09:15 **Oral presentations** by selected participants

09:15 – 09:45 Dawn M. Ireland, Congenital Diaphragmatic Hernia International

Parent-initiated international research in Congenital Diaphragmatic Hernia

Discussion

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 Prof. Effy Vayena, ETH Zurich

Interactive workshop: Ethical considerations (case study)

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

Session chair: Dr. Michel Hochuli, USZ, Zurich

16:30 – 17:00 Prof. Guenter Schwarz, Cologne University

Cysteine catabolism: From ultra-rare neurodegenerative disease to longevity

Discussion

17:00 – 17:30 Prof. Eckhard Wolf, LMU Munich, Germany

Genetically engineered pig models for human monogenic diseases

Discussion

17:30 – 21:00 Guided tour in the Ittingen Kartause and Grill



Friday, July 13, 2018

07:45 – 08:45 Breakfast

Session chair: Prof. Matthias Baumgartner, Kispi, Zurich

08:45 – 09:30 **Presentation of workshop results**

09:30 – 10:30 Dr. Roger Gfrörer, University of Zurich, Switzerland

Careers – a result of planning and happenstance

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 scholarships for (MD)-PhD students.