



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
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
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 <i>Discussion</i>
18:45 – 21:30	Networking dinner at the Ittingen Kartause



Thursday, July 12, 2018

07:45 – 08:45	Breakfast
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 <i>Discussion</i>
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, University of Zurich Interactive workshop: Ethical considerations (case study)
15:30 – 16:30	Coffee Break and preparation of workshop results
16:30 – 17:00	Prof. Guenter Schwarz, Cologne University Molybdenum enzyme deficiency: from cofactor biogenesis to drug development in ultra rare and common diseases <i>Discussion</i>
17:00 – 17:30	Prof. Eckhard Wolf, University of Zurich Genetically engineered pig models for human monogenic diseases <i>Discussion</i>
17:30 – 21:00	Guided tour in the Ittingen Kartause and Grill



Friday, July 13, 2018

07:45 – 08:45 Breakfast

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

09:45 – 10:25 tba

tba

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



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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.

