



Universität
Zürich ^{UZH}

radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

7th Rare Diseases Summer School

Kartause Ittingen, Warth

Wednesday, July 10th to Friday, July 12th 2019

PROGRAM

The program is subject to change without notice.

Please refer to the Summer School website for updates.





Wednesday, July 10, 2019

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	Dr. Jürg Streuli, Children's Hospital Zurich Interactive workshop: Ethical considerations (case study)
15:30 – 16:00	Coffee Break
16:00 – 16:40	Prof. André Brändli, Ludwig-Maximilians-University Munich, Germany Engineering Xenopus models of rare inherited diseases for in vivo drug discovery <i>Discussion</i>
16:40 – 17:20	Prof. Stephan Neuhauss, University of Zurich Studying rare diseases in the (not so rare) Zebrafish model organism <i>Discussion</i>
17:30 – 18:30	Guided Tour in Kartause (optional)
18:30 – 20:00	Dinner
20:00 –	Poster viewing



Thursday, July 11, 2019

07:45 – 08:45	Breakfast
08:45 – 09:45	Oral presentations by selected participants
09:45 – 10:30	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 – 12:00	Oral presentations by selected participants
12:00 – 13:15	Lunch
13:15 – 14:00	Dr. Guillaume Canaud, Necker Hospital Targeted therapy in patients with PIK3CA related overgrowth syndrome <i>Discussion</i>
14:00 – 15:15	Poster viewing
15:15 – 15:45	Coffee Break
15:45 – 17:00	Workshop on drug development led by Prof. Marshall Summar
17:00 – 18:15	Workshop on NGS led by Prof. Nine Knoers
17:50 – 18:30	Free time
18:30 – 21:00	Grill



Friday, July 12, 2019

07:45 – 08:45	Breakfast
08:45 – 09:40	Preparation and discussion of take-home messages by participants
09:40 – 10:20	Prof. Andrew Dwyer, Boston College, USA Translating rare disease research into practice using patient partnerships <i>Discussion</i>
10:20 – 10:50	Coffee Break
10:50 – 11:30	Prof. Marshall Summar, Washington DC, USA From rare diseases to common diseases: lessons from the urea cycle
11:30 – 12:00	Christina Fasser Patient-initiated research <i>Discussion</i>
12:00 – 12:15	Award of poster prizes and concluding remarks
12:30	End of the Summer School and departure of the participants