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.

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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 Session chair: Prof. Stephan Neuhauss, UZH, Zurich
10:30 – 11:10	Prof. Nine Knoers, Chair Dept. of Genetics, University Medical Center Groningen, the Netherlands **Next generation sequencing in rare disorders: scientific impact and clinical utility** *Discussion*
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** *Discussion*
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
	Session chair: Prof. Matthias Baumgartner, Kispi, Zurich
16:00 – 16:40	Prof. André Brändli, Ludwig-Maximilians-University Munich, Germany **Engineering Xenopus models of rare inherited diseases for in vivo drug discovery** *Discussion*
16:40 – 17:20	Prof. Stephan Neuhauss, University of Zurich **Studying rare diseases in the (not so rare) Zebrafish model organism** *Discussion*
17:30 – 18:30	Guided Tour in Kartause (optional)
18:30 – 20:00	Dinner
20:00 –	**Poster viewing**

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Thursday, July 11, 2019	
07:45 – 08:45	Breakfast
08:45 – 09:45	**Oral presentations** by selected participants Session chair: Prof. Giatgen Spinas, USZ, Zurich
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** *Discussion*
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, Paris, France **Keynote Lecture: Targeted therapy in patients with PIK3CA related overgrowth syndrome** *Discussion*
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
18:15 – 18:30	Free time
18:30 – 21:00	Grill

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Friday, July 12, 2019	
07:45 – 08:45	Breakfast
08:45 – 09:40	**Preparation and discussion of take-home messages** by participants Session chair: Dr. Saskia Karg, Kispi, Zurich
09:40 – 10:20	Prof. Andrew Dwyer, Boston College, USA **Translating rare disease research into practice using patient partnerships** *Discussion*
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** *Discussion*
12:00 – 12:15	Award of poster prizes 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.

SAMW ASSM
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Académie Suisse des Sciences Médicales
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