

## radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich

# 5<sup>th</sup> Rare Diseases Summer School

Tagungszentrum Schloss Au, Wädenswil  
Wednesday, June 7<sup>th</sup> to Friday, June 9<sup>th</sup> 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	<b>Welcome</b> , Prof. Matthias Baumgartner, Children's Hospital Zurich and University of Zurich, lead radiz Session chair: tbd
10:30 – 11:10	Prof. Nine Knoers, University Medical Centre Utrecht, the Netherlands <b>Next generation sequencing in rare disorders: scientific impact and clinical utility</b> <i>Discussion</i>
11:10 – 11:50	Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA <b>Clinical endpoints: selecting outcomes and surrogate markers in rare diseases</b> <i>Discussion</i>
11:50 – 12:50	Lunch Session chair: tbd
13:00 – 14:30	Dr. Martine Zimmermann, Alexion Pharma International Samantha Parker, Lysogene <b>Parallel workshops: Regulatory frameworks and registries</b>
14:30 – 15:30	<b>Oral presentations</b> by selected participants
15:30 – 16:00	Coffee Break
16:00 – 17:00	<b>Poster viewing</b>
17:00 – 17:40	Prof. Guenter Schwarz, Cologne University <b>Molybdenum enzyme deficiency: from cofactor biogenesis to drug development in ultra rare and common diseases</b> <i>Discussion</i>
17:40 – 18:30	Prof. James R. Lupski, Baylor College of Medicine, Houston, USA <b>Keynote Lecture: The genomics of disease: clinical genomics = personal genomes + clan genomics</b> <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	<b>Oral presentations</b> by selected participants
09:45 – 10:25	Dr. Jasmin Barman-Aksözen, Swiss society for porphyria <b>Let the sunshine in! Getting a drug approved for the rare genetic light intolerance erythropoietic protoporphyria</b> <i>Discussion</i>
10:30 – 11:00	Coffee Break
11:00 – 12:15	<b>Workshop Session</b> A: <b>Drug development</b> led by Prof. Marshall Summar B: <b>Next generation sequencing</b> led by Prof. Nine Knoers
12:15 – 13:15	Lunch
13:15 – 14:15	<b>Poster Viewing</b>
14:15 – 15:30	Dr. Jürg Streuli, University of Zurich <b>Interactive workshop: Ethical considerations (case study)</b>
15:30 – 16:30	Coffee Break and <b>preparation of workshop results</b>
16:30 – 17:00	Prof. Stephan Neuhauss, University of Zurich <b>Studying rare diseases in the (not so rare) Zebrafish model organism</b> <i>Discussion</i>
17:00 – 17:30	Prof. Carsten Wagner, University of Zurich <b>Possibilities and limitations of mouse models for rare disease research</b> <i>Discussion</i>
17:30 – 19:30	Social event

## Friday, June 9, 2017

08:15 – 08:45	Morning coffee Session chair: tbd
08:45 – 09:45	<b>Presentation of workshop results</b>
09:45 – 10:25	Prof. Janine Reichenbach, Children's Hospital Zurich <b>Clinical gene therapy for X-linked Chronic Granulomatous Disease</b> <i>Discussion</i>
10:30 – 11:00	Coffee Break
11:00 – 11:30	Prof. Marshall Summar, Children's National Medical Center, George Washington University, Washington DC, USA <b>From rare diseases to common diseases: lessons from the urea cycle</b> <i>Discussion</i>
11:30 – 12:00	Christina Fasser, Vice-President ProRaris, President Retina International <b>Patient-initiated research</b> <i>Discussion</i>
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.