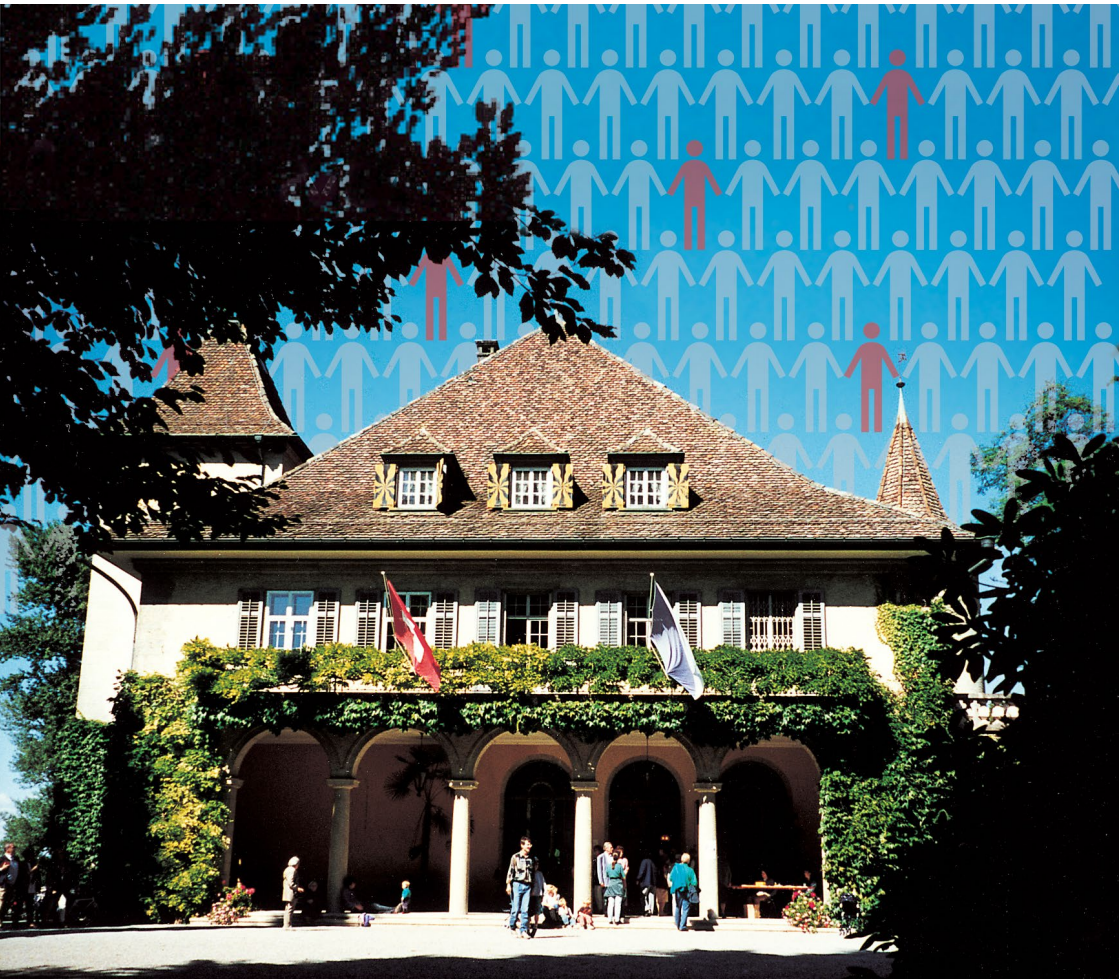


Tagungszentrum Schloss Au, Wädenswil (on lake Zurich), Switzerland  
4<sup>th</sup> to 6<sup>th</sup> of July 2013

# 1<sup>st</sup> Rare Diseases Summer School radiz - Rare Disease Initiative Zürich

Clinical Research Priority Program for Rare Diseases University of Zurich



Universität  
Zürich UZH



UniversitätsSpital  
Zürich



FORSCHUNGSZENTRUM  
FÜR DAS KIND (FZK)

KINDERSPITAL ZÜRICH  
Universitäts-Kinderkliniken · Eleonorenstiftung





# radiz - Rare Disease Initiative Zürich

A disease is considered rare when fewer than one in 2'000 persons is affected. Today, more than 7'000 rare diseases are known, affecting approximately five to eight percent of the global population. Most rare diseases already manifest themselves in childhood. Rare diseases often lead to a grave impairment of health and may be very difficult to diagnose.

While progress has been made in recent years in the diagnosis and treatment of rare diseases through diagnostic initiatives such as newborn screening and through the development of treatments under the orphan drug regulation, the situation for many patients is still dire. Patients and their families often embark on a medical odyssey until they finally receive a correct diagnosis. For many rare diseases, there are still no treatments. Interdisciplinary national and international collaborations are needed in order to improve the prospects for patients with rare diseases and to translate findings from research to the clinic.

The 1<sup>st</sup> radiz Rare Diseases Summer School will focus on a wide variety of subjects in the arena of rare diseases, from disease mechanisms and animal models, to improving diagnoses, to novel therapeutics.

There will be lectures and workshops on **drug development, model organisms**, how to choose **clinical endpoints, GCP** (Good Clinical Practice), **regulatory aspects, patient registries, patient initiated research, ethical considerations**, as well as what rare diseases may tell us about common diseases.

The summer school will contain lectures by national and international rare disease experts, workshops, and poster presentations by participants. A high staff to student ratio will allow participants to most optimally benefit from the varied expertise of the faculty. There will be ample time for discussions, teamwork, as well as for socializing.

## Organizing Committee

- **Prof. Matthias Baumgartner**, University Children's Hospital Zurich and University of Zurich
- **Prof. Barbara Plecko**, University Children's Hospital Zurich and University of Zurich
- **Prof. Olivier Devuyst**, University of Zurich and University Hospital Zurich
- **Dr. Saskia Karg**, University Children's Hospital Zurich





## Key Note Lecture

**Prof. Harry C. Dietz**, Johns Hopkins University School of Medicine, Baltimore, USA: Marfan syndrome – from bench to bedside

## Lecturers (in alphabetic order)

- **Christina Fasser**, Vice-President ProRaris, CEO Retina Suisse, President Retina International: Patient-initiated research
- **Prof. Nine Knoers**, University Hospital Nijmegen, Radboud University Nijmegen, Netherlands: Exome sequencing
- **Prof. Stephan Neuhaus**, University of Zurich: Model organisms
- **Samantha Parker**, Orphan Europe Recordati Group, France: Rare disease registries
- **PD Dr. Janine Reichenbach**, University Children's Hospital Zurich: Gene therapy
- **PD Dr. Gabriela Senti**, University Hospital Zurich: Good clinical practice
- **PD Dr. Andreas Serra**, University of Zurich and University Hospital Zurich: Clinical trials
- **Prof. Marshall Summar**, Children's National Medical Center, George Washington University, Washington DC, USA: Clinical endpoints; Rare diseases - common diseases
- **Dr. Effy Vayena**, University of Zurich: Ethical considerations
- **Dr. Urs Widmer**, Swiss Re, Zurich: Orphan Drug Development
- **Prof. Eckhard Wolf**, Ludwig-Maximilians-Universität München, Germany: Animal Models

## Language

The Summer School will be held in English.

## Eligibility and Application

The school addresses clinicians, physician-scientists, postdocs, and PhD students from various disciplines such as medicine, biology, psychology, sociology, and related fields. We plan to admit up to 40 participants. There are a limited number of spaces available to participants from industry. Participants are responsible for having valid insurance and a valid visa, if applicable, for Switzerland.

Participants are expected to present a poster about their work or research interests related to rare diseases at the Summer School. The best posters will be awarded a prize.

Please apply by sending a short CV, a letter of motivation including a brief description of your project or research interest, and one letter of recommendation to [saskia.karg@kispi.uzh.ch](mailto:saskia.karg@kispi.uzh.ch). Please indicate your room preference (see under "accommodation"). The application deadline is May 12<sup>th</sup> 2013. Notification of acceptance will be given by May 31<sup>st</sup> 2013.





## Fee

There is a non-refundable registration fee of CHF 200 for participants from academia and clinics and CHF 500 for participants from industry. The fee becomes due upon notification of acceptance. Food will be offered free of charge during the Summer School. Participants are responsible for their own travel.

## Accommodation

Accommodation on lake Zurich, close to the conference venue, will be organized, but needs to be paid by participants (there will be a choice of rooms for either approximately CHF 105 for two nights, double occupancy in a youth hostel or approximately CHF 365 for two nights, single occupancy in a hotel, choice subject to availability).

## Time and Venue

The summer school will take place at **Tagungszentrum Schloss Au**, Wädenswil (on lake Zurich), Switzerland and will start on Thursday, July 4<sup>th</sup> at 1 pm and end on Saturday, July 6<sup>th</sup> at 1 pm.

## Contact

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