

Faculty:

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Prof. Bernhard Zabel - Institute of Human Genetics and Centre for Pediatrics and Adolescent Medicine, Freiburg - bernhard.zabel@uniklinik-freiburg.de

The **Course** is intended to provide an introduction to the basic clinical and radiographic diagnostics and clinical management of skeletal dysplasias. Preference will be given to pediatricians, clinical geneticists and radiologists planning to develop a clinical activity in the field of genetic skeletal diseases. The number of participants is limited to 12. The official language is English.

Applications may be addressed by email to Dr. Sheila Unger, sheila.unger@chuv.ch. Please include a short CV and state your present involvement in genetic skeletal disease. Priority will be given to those applicants who submit a letter from their institution confirming commitment to support clinical activity in the domain of skeletal dysplasias.

Course Fee: 950 CHF - includes course tuition and materials, 6 nights at City Hotel with breakfast; lunches at hospital cafeteria; Course dinner.

Course venue: Centre Hospitalier Universitaire Vaudois (CHUV), 1011 Lausanne, Switzerland. Lausanne is easily reached by train from international airports in Geneva (45 min ride) or Zurich (2 hours ride). There is a direct connection on the metro line M2 from the train station to the CHUV (station CHUV) and to the City Hotel (station Bessières). See also www.skeldys.org for further information.

Course Coordination and Secretariat:

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Supported by: Actelion, Biomarin, Enobia, Genzyme (Sanofi), Novo Nordisk, and SKELNET within the BMBF Rare Diseases Programme



Centre for Pediatrics
and Adolescent Medicine
University of Freiburg



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Sixth Annual Introductory Course on Skeletal Dysplasias

July 2nd to 6th, 2012

Centre Hospitalier Universitaire Vaudois
Lausanne, Switzerland

**Organizers:**

Sheila Unger, Luisa Bonafe,
Andrea Superti-Furga (Lausanne)
Ekkehart Lausch,
Bernhard Zabel (Freiburg)

Day 1: Monday July 2, 2012

- 09:00-09:30 Welcome and Introduction (*Unger*)
 09:30-10:15 How to Approach a Patient with no Diagnosis (*Spranger*)
 10:15-10:45 Introduction to Physical Examination (*Unger*)
 10:45-11:15 Break
 11:15-12:15 Introduction to Radiographic Diagnosis (*Superti-Furga*)
 12:15-13:15 Lunch
 13:15-14:00 Clinical-Radiographic Session: Focus FGFR3 (*Unger*)
 14:00-15:00 Special Management of Achondroplasia (*Zabel*)
 15:00-15:30 Break
 15:30-16:30 Clinical-Radiographic Session: Focus COMP and MED (*Superti-Furga*)

Day 2: Tuesday July 3, 2012

- 08:30-09:00 Explanation of Day's Activities and Introduction to Patients (*Unger*)
 09:00-10:30 Patient Examination History and Physical (*Unger*)
 10:30-11:30 Review of Patient Radiographs and Presentation to Group (*All Faculty*)
 11:30-11:40 Break
 11:40-12:00 Achondroplasia: A Parent's View (*Mohr*)
 12:00-13:00 Lunch
 13:00-14:00 Segmentation anomalies (*Bonafé*)
 14:00-15:00 Clinical-Radiographic Session: Focus Type 2 Collagen Disorders (*Spranger*)
 15:00-15:30 Break
 15:30-17:00 Session: Unknown Cases From Students (*Nishimura, Faculty*)

Day 3: Wednesday July 4, 2012

- 09:00-10:00 Signalling Pathways, the Growth Plate, and Skeletal Dysplasias (*Zabel*)
 10:00-11:00 Osteopetrosis and Sclerosing Bone Dysplasias (*Nishimura*)
 11:00-11:30 Break
 11:30-12:15 Clinical-Radiographic Session: Focus Dysostosis Multiplex and CDP (*Unger*)
 12:15-13:00 Lunch
 13:00-14:00 Special Lecture: Radiographic Signs (*Hall*)
 14:00-14:30 Break
 14:30-15:15 Prenatal Diagnosis of Skeletal Dysplasias (*Unger*)
 15:15-16:30 Clinical-Radiographic Session: Focus Lethals (*Superti-Furga*)

Day 4: Thursday July 5, 2012

- 09:00-9:30 Prenatally Detectable Skeletal Dysplasia Quiz (*Unger*)
 09:30-10:00 Review of Quiz (*Unger*)
 10:00-10:30 Break
 10:30-11:15 Clinical-Radiographic Session: Focus OI and related disorders (*Hall*)
 11:15-12:00 Skeletal dysplasias and Joint Dislocations (*Bonafé*)
 12:00-13:00 Lunch
 13:00-14:00 Differential Diagnosis of Non-Accidental Injury and OI (*Hall*)
 14:00-15:00 Clinical-Radiographic Session: Focus Metaphyseal Dysplasias (*Superti-Furga*)
 15:00-15:30 Break
 15:30-16:00 Clinical Radiographic Session: Focus TRPV4 Family (*Lausch*)
 16:00-17:00 Session: Unknown Cases From Students (*Nishimura, Faculty*)
 19:00-22:00 **Course Dinner**

Day 5: Friday July 6, 2012

- 09:00-09:45 Rare Diseases and Common Problems: Lessons from One to the Other (*Ikegawa*)
 09:45-10:30 Brachydactylies (*Horn*)
 10:30-11:00 Break
 11:00-12:15 General Skeletal Dysplasias Quiz (*Unger*)
 12:15-12:45 Review of Quiz (*Unger*)
 12:45-13:30 Lunch
 13:30-14:30 Wrap up talk: the Retrospectroscope and the Crystal Ball (*Superti-Furga*)
 14:30-15:00 Course Evaluation and Feedback

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