

Faculty:**PD Dr. Luisa Bonafé**

Division of Molecular Pediatrics, Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland - luisa.bonafe@chuv.ch

Prof. Christine Hall

Emerita, Department of Radiology, Great Ormond Street Hospital for Children, London, UK - c.hall-cooper@waitrose.com

PD Dr. Denise Horn

Institute for Medical Genetics, Freie Universität Berlin - denise.horn@charite.de

Dr. Ekkehart Lausch

Centre for Pediatrics and Adolescent Medicine, Freiburg - ekkehart.lausch@uniklinik-freiburg.de

Prof. Gen Nishimura

Department of Radiology, Tokyo Metropolitan Kiyose Children's Hospital, Kiyose, Japan - gennishi@chp-kiyose-tokyo.jp

Prof. Gerd Scherer

Institute of Human Genetics, Freiburg - gerd.scherer@uniklinik-freiburg.de

Prof. Jürgen Spranger

Centre for Pediatrics and Adolescent Medicine, Freiburg - juergen.spranger@uniklinik-freiburg.de

Prof. Andrea Superti-Furga

Centre for Pediatrics and Adolescent Medicine, Freiburg - asupert@uniklinik-freiburg.de

Prof. Markus Uhl

Centre for Pediatrics and Adolescent Medicine, Freiburg - markus.uhl@rkk-ggmbh.de

Dr. Sheila Unger

ESDN Clinical and Radiological Review Coordinator, Institute of Human Genetics and Centre for Pediatrics and Adolescent Medicine, Freiburg - sheila.unger@uniklinik-freiburg.de

Prof. Matthew Warman

Orthopedic Research Laboratories, Children's Hospital Boston, USA - matthew.warman@childrens.harvard.edu

Prof. Bernhard Zabel

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

Supported by SKELNET within the BMBF Rare Diseases Programme



The Course is intended to provide an introduction to the basic clinical and radiographic diagnostics and clinical management of skeletal dysplasias. The number of participants is limited to 12. The official language is English.

Preference will be given to pediatricians, clinical geneticists and radiologists planning to develop a clinical diagnostic activity in the field of genetic skeletal diseases.

Applications may be addressed to Dr. Sheila Unger, Pädiatrische Genetik, Zentrum für Kinderheilkunde und Jugendmedizin, Mathildenstrasse 1, 79106 Freiburg, Germany; or by email to sheila.unger@uniklinik-freiburg.de. 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: 800 Euros; includes course tuition and materials, 6 nights at Apart Hotel with breakfast; lunches at hospital cafeteria; Farewell dinner.

Course venue: Centre for Pediatrics and Adolescent Medicine, Mathildenstrasse 1, 79106 Freiburg. Freiburg is two hours' train ride from Zurich or Frankfurt International Airport. See also www.skeldys.org for further information.

Course Coordination and Secretariat:

Violetta Volz and Charis Marschner,
Sektion Pädiatrische Genetik,
ZKJ,
Mathildenstrasse 1,
79106 Freiburg, Germany
Phone +49-761-270-4363
violetta.volz@uniklinik-freiburg.de
charis.marschner@uniklinik-freiburg.de

Accommodation: Apart Hotel, Mathildenstr. 14
D-79106 Freiburg, Germany
Phone: +49-761-385-570, Fax: +49-761-275-135



UNIVERSITÄTS
FREIBURG KLINIKUM



Centre for Pediatrics
and
Adolescent Medicine
University of Freiburg

Fourth Annual Introductory Course on Skeletal Dysplasias

July 5th to 9th, 2010

Centre for Pediatrics and Adolescent Medicine
Freiburg, Germany

Organizers:

Sheila Unger,

Andrea Superti-Furga, Bernhard Zabel

Coordinators: Violetta Volz and Charis Marschner



Day 1: Monday July 5, 2010

- 09:00-09:30 Welcome and Introduction (*Unger*)
 09:30-10:15 How to Approach a Patient with an Unknown Skeletal Dysplasia (*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 Considerations in Achondroplasia (*Zabel*)
 15:00-15:30 Break
 15:30-16:30 Clinical-Radiographic Session: Focus COMP and MED (*Superti-Furga*)

Day 2: Tuesday July 6, 2010

- 09:00-10:00 Signalling Pathways, the Growth Plate, and Skeletal Dysplasias (*Zabel*)
 10:00-11:00 Osteopetrosis and Sclerosing Bone Dysplasias (*Hall*)
 11:00-11:30 Break
 11:30-12:15 Clinical-Radiographic Session: Focus Dysostosis Multiplex and CDP (*Spranger, Unger*)
 12:15-13:15 Lunch
 13:15-14:15 SOX9 and Campomelic Dysplasia: Overview (*Scherer*)
 14:15-15:15 Clinical-Radiographic Session: Focus Type 2 Collagen Disorders (*Spranger*)
 15:15-15:30 Break
 15:30-16:30 Unknown Session Cases From Students (*Nishimura*)

Day 3: Wednesday July 7, 2010

- 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 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 8, 2010

- 09:00-9:30 Prenatally Detectable Skeletal Dysplasia Quiz (*Unger*)
 09:30-10:00 Review of Quiz (*Unger*)
 10:00-10:30 Break
 10:30-11:00 Clinical-Radiographic Session: Focus OI and related disorders (*Hall*)
 11:00-12:00 Molecular Underpinnings of the Skeleton (*Warman*)
 12:00-13:00 Segmentation Anomalies (*Bonafé*)
 12:30-13:30 Lunch
 13:30-14:30 Differential Diagnosis of Non-Accidental Injury and OI (*Uhl*)
 14:30-15:30 Clinical-Radiographic Session: Focus Metaphyseal Dysplasias (*Superti-Furga*)
 15:30-17:00 Unknown Session Cases From Students (*Nishimura*)

Day 5: Friday July 9, 2010

- 09:00-09:30 Clinical Radiographic Session: Focus TRPV4 Family (*Lausch*)
 09:30-10:30 Brachydactyilies (*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 Recent and Upcoming Skeletal Dysplasia Gene Families and Wrap-up (*Superti-Furga*)
 14:30-15:00 Course Evaluation and Feedback
 19:00-22:00 **Farewell Dinner**

