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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 diagnostic 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 at 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: 1200 CHF; includes course tuition and materials, 5 hotel nights, breakfast, lunches, and Course official dinner.

Course venue: Aquatis Hotel, Lausanne

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 Aquatis Hotel (metro station VENNES). See also www.skeldys.org for further information.

Accommodation: Aquatis Hotel, <https://www.aquatis-hotel.ch/en/>

Course Coordination and Secretariat:

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PRELIMINARY PROGRAM

13th Annual Introductory Course on Skeletal Dysplasias

July 1 to 5, 2019

Centre Hospitalier Universitaire Vaudois
Lausanne, Switzerland

Organizers:

Sheila Unger, Andrea Superti-Furga, Nathalie
Zumstein (CHUV, Lausanne)



Sunday, June 30 - 18:00 Get Together at the Hotel Aquatis**Day 1: Monday July 1, 2019**

08:30-09:00	Welcome and introduction <i>S. Unger</i>
09:00-09:45	Introduction to Clinical Examination <i>S. Unger</i>
09:45-10:15	Break
10:15-11:15	The art and science of radiographic assessment – and its pitfalls <i>A. Superti-Furga</i>
11:15-12:15	Focus Metaphyseal dysplasias <i>C. Hall</i>
12:15-13:00	Lunch
13:00-13:45	Focus FGFR3: Clinical, radiographic and management <i>S. Unger</i>
13:45-14:15	FGFR3 disorders: cellular basis and therapeutic strategies <i>E. Gouze</i>
14:15-14:45	Break
14:45-15:45	Hypochondroplasia and other dysplasias with short stature and “mild skeletal abnormalities” <i>K. Heath</i>
15:45-17:00	Unknown case session <i>G. Nishimura</i>

Day 2: Tuesday July 2, 2019

08.30-09:30	Useful skeletal signs in syndromology <i>C. Ferreira</i>
09:30-10:00	Focus enchondromas and exostoses <i>S. Unger</i>
10:00-10:30	Break
10:30-11:30	Focus Sclerosing bone disorders <i>C. Hall</i>
11:30-12:30	Focus joint dislocations <i>A. Superti-Furga</i>
12:30-13:30	Lunch
13:30-14:15	Focus chondrodysplasia Punctata <i>G. Nishimura</i>
14:15-15:00	Focus COMP and MEDs <i>S. Unger</i>
15:00-15:30	Break
15:30-17:30	Unknown case session <i>G. Nishimura</i>

Day 3: Wednesday July 3, 2019

08:30-09:00	Explanation of activities and introduction to patients <i>S. Unger</i>
09:00-10:30	Patient Examination - Students and Faculty
10:30-12:00	Student presentations - Students and Faculty
12:00-13:00	Lunch
13:00-13:30	Achondroplasia: A parent's view <i>B. Mohr</i>
13:30-14:30	Focus Short Rib-Polydactyly Syndromes and skeletal ciliopathies <i>C. Hall</i>
14:30-15:00	Break
15:00-15:30	Focus on hypophosphatasia <i>G. Nishimura</i>
15:30-16:30	Focus on prenatally detected, severe dysplasias – formerly “lethals” <i>A. Superti-Furga</i>
16:30-17:15	Prenatal Diagnosis of Skeletal Dysplasias <i>S. Unger</i>

Day 4: Thursday July 4, 2019

08:30-09:30	Prenatally detectable skeletal dysplasia quiz <i>S. Unger</i>
09:30-10:00	Break
10:00-11:00	Focus: type 2 collagen disorders <i>G. Mortier</i>
11:00-12:00	Focus Osteogenesis Imperfecta <i>L. Bonafé</i>
12:00-13:00	Lunch
13:00-14:00	Lysosomal Disorders <i>A. Superti-Furga</i>
14:00-14:45	Focus mesomelic and acro-mesomelic <i>G. Mortier</i>
14:45-15:30	Principles of Dysostoses <i>C. Ferreira</i>
15:30-16:00	Break
16:00-17:00	Quiz on Radiographic signs <i>C. Hall</i>

19:30 COURSE OFFICIAL DINNER

Day 5: Friday July 5, 2019

08:30-09:00	Focus on Osteolyses <i>G. Mortier</i>
09:00-10:30	General Quiz
10:30-11:00	<i>S. Unger</i> Break
11:00-12:00	Wrap-up lecture <i>A. Superti-Furga</i>
12:00-12:30	Lunch
12:30-13:30	Course evaluation and feedback
13:30-14:30	Unknown case session <i>G. Nishimura</i>

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ALEXION

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