

## Faculty

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### Sheila Unger

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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 20. The official language is English.

**Applications** may be addressed by email to Dr Sheila Unger at [sheila.unger@chuv.ch](mailto: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](http://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

# 14<sup>th</sup> Annual Introductory Course on Skeletal Dysplasias

July 3 to 7, 2023

Centre Hospitalier Universitaire Vaudois  
Lausanne, Switzerland

#### Organizers:

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

**Sunday, July 2 - 18:00 Get Together at the Hotel Aquatis****Day 1: Monday July 3, 2023**

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 on type 2 collagen disorders and related <i>G. Mortier</i>
12:15-13:00	Lunch
13:00-13:45	Focus FGFR3: Clinical, radiographic and management <i>S. Unger</i>
13:45-14:15	Hypochondroplasia and other conditions with short stature and mild skeletal abnormalities <i>K. Heath</i>
14:15-14:45	Break
14:45-15:30	FGFR3 disorders: cellular basis and therapeutic strategies <i>E. Gouze</i>
15:30-16:15	The diverse genetic basis of skeletal disorders <i>A. Superti-Furga</i>
16:15-17:00	Unknowns session

**Day 2: Tuesday July 4, 2023**

08.30-09:15	Focus: Osteogenesis Imperfecta <i>C. Hall</i>
09:15-09:45	Focus: Pseudoachondropalsia and the MEDs <i>S. Unger</i>
09:45-10:30	Focus: Metaphyseal Dysplasias <i>A. Superti-Furga</i>
10:30-11:00	Break
11:00-11:45	Skeletal findings in syndromology part 1 <i>C. Ferreira</i>
11:45-12:30	Focus: Chondrodysplasia Punctata <i>G. Nishimura</i>
12:30-14:00	Lunch
14:00-14:30	Focus: Enchondromas and Exostoses <i>S. Unger</i>
14:30-15:15	Focus : Congenital joint dislocations <i>A. Superti-Furga</i>
15:15-15.30	Break
15:30-16:00	Focus: the so-called Skeletal Ciliopathies <i>C. Ferreira</i>
16:00-17:00	Unknowns session

**Day 3: Wednesday July 5, 2023**

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	Achondroplasia and short stature: A parent's view <i>B. Mohr</i>
13:00-14:00	Lunch
14:00-14:30	Focus: Non-Accidental Injury <i>C. Hall</i>
14:30-15:15	Focus: Prenatally detected, severe dysplasias (formerly "lethals") <i>A. Superti-Furga</i>
15:15-15:30	Break
15:30-16:00	Focus : Hypophosphatasia <i>G. Nishimura</i>
16:00-17:00	Prenatal diagnosis of skeletal dysplasias <i>S. Unger</i>

**Day 4: Thursday July 6, 2023**

08:30-09:30	Quiz: Prenatally detectable skeletal dysplasia <i>S. Unger</i>
09:30-10:15	Focus: Lysosomal diseases <i>A. Superti-Furga</i>
10:15-10:45	Break
10:45-11:30	Focus: Mesomelic and acromesomelic dysplasias <i>G. Mortier</i>
11:30-12:15	Focus: Sclerosing bone dysplasias <i>C. Hall</i>
12:15-13:30	Lunch
13:30-14:15	Skeletal findings in syndromology part 2 <i>C. Ferreira</i>
14:15-14:30	Break
14:30-15:00	From X-rays to X-omes: choosing wisely <i>S. Unger</i>
15:00-16:00	Quiz: Radiographic signs <i>C. Hall</i>
16:00-17:00	Unknowns session
19:30	<b>COURSE OFFICIAL DINNER</b>

**Day 5: Friday July 7, 2023**

08:30-09:15	Metabolic, molecular and pharmacological treatments - current and upcoming <i>C. Ferreira</i>
09:15-10:30	Final "unknowns session" Course evaluation & feedback Break
10:30-10:45	General Quiz
10:45-12:00	<i>S. Unger</i>
12:00-12:45	Wrap-up lecture <i>A. Superti-Furga</i>
12:45-13:30	lunch

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*Sponsors to be confirmed*