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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

14th Annual Introductory Course on Skeletal Dysplasias

July 6 to 10, 2020

Centre Hospitalier Universitaire Vaudois
Lausanne, Switzerland

Organizers:

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

Sunday, July 5 - 18:00: Get Together at the Hotel Aquatis**Day 1: Monday July 6, 2020**

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 7, 2020

08.30-09:15	Focus: Osteogenesis Imperfecta <i>C. Hall</i>
09:15-09:45	Focus: Pseudoachondroplasia 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>
12:30-13:30	Lunch
13:30-14:00	Focus: Enchondromas and Exostoses <i>S. Unger</i>
14:00-14:30	Focus: TGFβ signalling and its disorders <i>G. Mortier</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 8, 2020

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 9, 2020

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>
10:15-10:30	Break
10:30-11:15	Focus: Mesomelic and acromesomelic dysplasias <i>G. Mortier</i>
11:15-12:00	Focus: Sclerosing bone dysplasias <i>C. Hall</i>
12:00-13:00	Lunch
13:00-13:30	Focus: TRPV4 dysplasias <i>G. Mortier</i>
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 COURSE OFFICIAL DINNER**Day 5: Friday July 10, 2020**

08:30-09:15	Special lecture: The giants on whose shoulders we stand <i>J. Spranger</i>
09:15 – 10:30	Metabolic, molecular and pharmacological treatments - current and upcoming <i>C. Ferreira</i>
10:30-10:45	Break
10:45-12:00	General Quiz <i>S. Unger</i>
12:00-12:45	Wrap-up lecture <i>A. Superti-Furga</i>
12:45-13:30	lunch
13:30-15:00	final “unknowns session” Course evaluation & feedback

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ALEXION

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