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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

11th Annual Introductory Course on Skeletal Dysplasias

July 3 to 7, 2017

Centre Hospitalier Universitaire Vaudois
Lausanne, Switzerland

Organizers:

Sheila Unger, Andrea Superti-Furga, Gilles Eggen



Day 1: Monday July 3, 2017

08:30-09:00	Welcome and introduction <i>S. Unger</i>
09:00-09:30	Introduction to Clinical Examination <i>S. Unger</i>
09:30-10:30	The art and science of radiographic assessment – and its pitfalls <i>A. Superti-Furga</i>
10:30-11:00	Break
11:00-11:30	“The Shoulders on Which we Stand” <i>J. Spranger</i>
11:30-12:30	Focus on type 2 collagen disorders and related <i>G. Mortier</i>
12:30-13:30	Lunch
13:30-14:15	Focus FGFR3: Clinical, radiographic and management <i>S. Unger</i>
14:15-15:00	FGFR3 disorders: cellular basis and therapeutic strategies <i>E. Gouze</i>
15:00-15:30	Break
15:30-16:15	Lysosomal storage disorders: diagnosis and management <i>L. Bonafé</i>
16:15-17:00	Focus short Rib-Polydactyly Syndromes and skeletal ciliopathies <i>C. Hall</i>

Day 2: Tuesday July 4, 2017

08:30-09:00	Focus segmentation anomalies <i>L. Bonafé</i>
09:00-10:00	Focus metaphyseal dysplasias <i>A. Superti-Furga</i>
10:00-10:30	Break
10:30-11:00	Focus enchondromas and exostoses <i>S. Unger</i>
11:00-12:00	Focus sclerosing bone dysplasias <i>C. Hall</i>
12:00-13:00	Lunch
13:00-13:30	Focus Chondrodysplasia Punctata <i>G. Nishimura</i>
13:30-14:30	Focus mesomelic and acromesomelic disorders <i>G. Mortier</i>

14:30-15:15	Spotlight on the TGFbeta signalling pathways and associated disorders <i>A. Superti-Furga</i>
15:00-15:30	Break
15:30-17:00	Unknown case session <i>Faculty</i>

Day 3: Wednesday July 5, 2017

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 joint dislocation <i>A. Superti-Furga</i>
14:30-15:00	Break
15:00-16:00	Focus on prenatally detected, severe dysplasias – formerly “lethals” <i>A. Superti-Furga</i>
16:00-16:30	Focus on hypophosphatasia <i>G. Nishimura</i>
16:30-17:30	Prenatal diagnosis of skeletal dysplasias <i>S. Unger</i>

Day 4: Thursday July 6, 2017

08:30-09:30	Focus Osteogenesis Imperfecta <i>L. Bonafé</i>
09:30-10:30	Differential diagnosis of non-accidental injury and OI <i>C. Hall</i>
10:30-11:00	Break
11:00-11:30	Skeletal features in genetic “syndromes” <i>C. Ferreira</i>
11:30-12:30	Prenatally detectable skeletal dysplasia quiz <i>S. Unger</i>
12:30-13:30	Lunch
13:30-14:00	Spotlight on the osteolyses <i>G. Mortier</i>
14:00-15:00	Focus COMP and multiple epiphyseal dysplasia <i>S. Unger</i>
15:00-15:30	Break
15:30-16:15	Quiz on “radiographic signs” <i>C. Hall</i>
16:15-17:30	Unknown case session <i>Faculty</i>

19:30 COURSE OFFICIAL DINNER

Day 5: Friday July 7, 2017

08:30-09:30	Big Data for Skeletons: from common skeletal ailments to rare diseases - and back <i>S. Ikegawa</i>
09:30-10:30	Molecular technologies for diagnosis of skeletal dysplasias <i>C. Ferreira</i>
10:30-11:00	Break
11:00-12:30	General Quiz <i>S. Unger</i>
12:30-13:30	Lunch
13:30-14:30	Wrap-up lecture <i>A. Superti-Furga</i>
14:30-15:00	Course evaluation and feedback

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Sponsors to be confirmed

ALEXION

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