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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 Dre Sheila Unger, 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: 1000 CHF; includes course tuition and materials, 5 hotel nights, lunches and Course official dinner.

Course venue: Centre Hospitalier Universitaire Vaudois, (CHUV) Av. du Bugnon 21, Lausanne, Switzerland. Room "seminaire 7" 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 CHUV (station CHUV). See also www.skeldys.org for further information.

Accommodation: Hotel Alpha Palmiers, Rue du Petit Chêne 34, 1003 Lausanne, www.byfassbind.com

Course Coordination and Secretariat:

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PRELIMINARY PROGRAM

10th Annual Introductory Course on Skeletal Dysplasias

July 4 to 8, 2016

Centre Hospitalier Universitaire Vaudois
Lausanne, Switzerland

Organizers:

Sheila Unger, Luisa Bonafe, Andrea Superti-Furga
(CHUV, Lausanne)

Skeletal Dysplasia Course Lausanne

Day 1: Monday July 4, 2016

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	FGFR3 disorders: cellular basis and therapeutic strategies <i>E. Gouze</i>
14:15-14:45	Break
14:45-15:45	Lysosomal storage disorders: diagnosis and management <i>L. Bonafé</i>
15:45-16:30	Focus short Rib-Polydactyly Syndromes and skeletal ciliopathies <i>C. Hall</i>
16:30-17:30	Unknown case session <i>G. Nishimura</i>

Day 2: Tuesday July 5, 2016

08.30-09:30	Focus metaphyseal dysplasias <i>A. Superti-Furga</i>
09:30-10:00	Focus segmentation anomalies <i>L. Bonafé</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>G. Nishimura</i>

Skeletal Dysplasia Course Lausanne

Day 3: Wednesday July 6, 2016

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	Big data for skeletons <i>S. Ikegawa</i>
14:30-15h00	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 7, 2016

08:30-09:30	Prenatally detectable skeletal dysplasia quiz <i>S. Unger</i>
09:30-10:00	Break
10:00-11:00	The neuroradiologic aspects of bone diseases <i>N. Aida</i>
11:00-12:00	Focus Osteogenesis Imperfecta <i>L. Bonafé</i>
12:00-13:00	Lunch
13:00-14:00	Differential diagnosis of non-accidental injury and OI <i>C. Hall</i>
14:00-14:30	Spotlight on the osteolyses <i>G. Mortier</i>
14:30-15:30	Focus COMP and multiple epiphyseal dysplasia <i>S. Unger</i>
15:30-16:00	Break
16:00-16:45	Quiz on “radiographic signs” <i>C. Hall</i>
16:45-17:30	Unknown case session <i>G. Nishimura</i>
19:30	COURSE OFFICIAL DINNER

Skeletal Dysplasia Course Lausanne

Day 5: Friday July 8, 2016

09:00-10:00	Focus joint dislocation <i>A. Superti-Furga</i>
10:00-11:00	Unknown case session <i>G. Nishimura</i>
11:00-11:15	Break
11:15-12:45	General Quiz <i>S. Unger</i>
12:45-13:30	Lunch
13:30-14:15	“The Shoulders on Which we Stand” <i>J. Spranger</i>
14:15-14:30	10 years of skeletal dysplasia course <i>S. Unger</i>
14 :30-15:15	Wrap-up lecture <i>A. Superti-Furga</i>
15:15-15:30	Course evaluation and feedback

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Sponsors to be confirmed



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