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Skeletal Dysplasia Course Lausanne 2018

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. All participants are invited to submit at least two undiagnosed cases for the "unknowns sessions".

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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Skeletal Dysplasia Course Lausanne 2018

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PRELIMINARY PROGRAM

12th Annual Introductory Course on Skeletal Dysplasias

July 9 - 13, 2018
Hotel Aquatis,
Lausanne,
Switzerland

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

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Sunday, July 8 - 18:00 Get Together at the Hotel Aquatis

Day 1: Monday July 9, 2018

8:30-9:00	Welcome and introduction - S. Unger
9:00-9:45	Introduction to Clinical Examination - S. Unger
9:45-10:15	Coffee break
10:15-11:15	The art and science of radiographic assessment – and its pitfalls - A. Superti-Furga
11:15-12:15	Focus on type 2 collagen disorders and related - G. Mortier
12:15-13:00	Lunch
13:00-13:45	Focus FGFR3: Clinical, radiographic and management - S. Unger
13:45-14:15	FGFR3 disorders: cellular basis and therapeutic strategies – E. Gouze
14:15-14:45	Coffee break
14:45-15:45	Hypochondroplasia and other dysplasias with short stature and "mild skeletal abnormalities" - K.Heath
15:45-16:45	Focus Short Rib-Polydactyly Syndromes and skeletal ciliopathies -C.Hall

Day 2: Tuesday July 10, 2018

8.30-9:30	Molecular technologies for diagnosis of skeletal dysplasias- C.Ferreira
9:30-10:30	Focus enchondromas and exostoses-S. Unger
10:00-10:30	Coffee break
10:30-11:30	Focus Metaphyseal dysplasias –A. Superti-Furga
11:30-12:30	Focus sclerosing bone dysplasias - C. Hall
12:30-13:30	Lunch
13:30-14:15	Focus Chondrodysplasia Punctata - G. Nishimura
14:15-15:15	Focus mesomelic and acromesomelic disorders- G. Mortier
15:15-15:45	Break
15:45-17:30	Unknown case session-G. Nishimura, J. Spranger, Students and Faculty

Day 3: Wednesday July 11, 2018

8:30-9:00	Explanation of activities and introduction to patients - S. Unger
9:00-10:30	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 - B. Mohr
13:30-14:00	Focus Lysosomal Storage Disorders - L. Bonafé
14:00-14:30	Break
14:30-15:30	Focus on prenatally detected, severe dysplasias - formerly "lethals" - A. Superti-Furga
15:30-16:00	Focus hypophosphatasia - G.Nishimura
16:00-17:00	Prenatal diagnosis of skeletal dysplasias - S. Unger

Day 4: Thursday July 12, 2018

8:30-9:30	Prenatally detectable skeletal dysplasia quiz – S. Unger
9:30-10:00	Coffee break
10:00-11:00	Focus joint dislocations – A. Superti-Furga
11:00-12:00	Focus OI - L. Bonafé
12:00-13:00	Lunch
13:00-14:00	Differential diagnosis of non-accidental injury and OI - C. Hall
14:00-14:30	Focus COMP and MED - S. Unger
14:30-15:00	Syndromes with skeletal involvement part 1- C. Ferreira
15:00-15:30	Coffee break
15:30-16:00	Focus on the Osteolyses - G. Mortier
15:45-16:30	Quiz on Radiographic signs- C. Hall
17:00-17:30	Spotlight on the TGFbeta signalling pathway and associated disorders – A. Superti-Furga
19:30	**COURSE DINNER**

Day 5: Friday July 13, 2018

9:00-9:30	Syndromes with skeletal involvement part 2- C. Ferreira
9:30-11:00	Unknown case session - G. Nishimura, Students and Faculty
11:00-11:15	Coffee Break
11:15-12:45	General Quiz - S. Unger
12:45-13:30	Lunch
13:30-14.30	Wrap-up lecture - A.Superti-Furga
14:30-15:00	Course evaluation and feedback

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Sponsors (to be confirmed)

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