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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](http://www.skeldys.org) for further information.

**Accommodation:** Hotel Alpha Palmiers, Rue du Petit Chêne 34, 1003 Lausanne, [www.byfassbind.com](http://www.byfassbind.com)

**Course Coordination and Secretariat:**

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# Program

## 9th Annual Introductory Course on Skeletal Dysplasias

July 6 to 10, 2015

Centre Hospitalier Universitaire Vaudois  
Lausanne, Switzerland

**Organizers:**

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

## Day 1: Monday July 6, 2015

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	Type 2 collagen disorders and related <i>G. Mortier</i>
12:15-13:00	Lunch
13:00-13:45	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>
15:45-16:15	Short Rib-Polydactyly Syndromes <i>C. Hall</i>
16:15-17:15	Molecular bases and genotype/phenotype correlations in the skeletal ciliopathies <i>V. Cormier-Daire</i>

## Day 2: Tuesday July 7, 2015

08.30-09:30	Metaphyseal dysplasias <i>A. Superti-Furga</i>
09:30-10:00	Segmentation anomalies <i>L. Bonafé</i>
10:00-10:30	Break
10:30-11:00	Enchondromas <i>S. Unger</i>
11:00-12:00	Sclerosing bone dysplasias <i>C. Hall</i>
12:00-13:00	Lunch
13:00-14:00	Chondrodysplasia Punctata <i>G. Nishimura</i>
14:00-15:00	Mesomelic and acromesomelic disorders <i>G. Mortier</i>
15:00-15:45	Break
15:45-16:15	The TGFbeta and cyclic nucleotides signalling pathways and associated disorders <i>V. Cormier-Daire</i>
16:15-17:15	Unknown case session <i>G. Nishimura</i>

## Day 3: Wednesday July 8, 2015

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>
13:30-14:00	Spotlight on TRPV4 <i>A. Superti-Furga</i>
14:00-15:00	Rare disorders and common problems in the age of deep sequencing <i>S. Ikegawa</i>
15:00-15:15	Break
15:15-16:15	Focus on prenatally detected, severe dysplasias – formerly "lethals" <i>A. Superti-Furga</i>
16:15-17:00	Prenatal diagnosis of skeletal dysplasias <i>S. Unger</i>

## Day 4: Thursday July 9, 2015

08:30-09:30	Prenatally detectable skeletal dysplasia quiz <i>S. Unger</i>
09:30-10:00	Coffee break
10:00-11:00	Joint dislocations <i>A. Superti-Furga</i>
11:00-12:00	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>
14:00-15:00	Focus COMP and multiple epiphyseal dysplasia <i>S. Unger</i>
15:00-15:30	Break
15:30-16:00	PPRD and the differential diagnosis of joint pain <i>L. Bonafé</i>
16:00-17:00	Unknown case session <i>G. Nishimura</i>
19:30	COURSE OFFICIAL DINNER

## Day 5: Friday July 10, 2015

09:00-10:00	Quiz on "radiographic signs" <i>C. Hall</i>
10:00-11:00	Short fingers: the isolated and syndromic Brachydactylies <i>G. Mortier</i>
11:00-11:15	Break
11:15-12:45	General Quiz <i>S. Unger</i>
12:45-13:30	Lunch
13:30-14:30	Wrap up talk <i>A. Superti-Furga</i>
14:30-15:00	Course evaluation and feedback

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Sponsored by:



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