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/

**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

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

### Day 2: Tuesday July 4, 2017

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

### Day 3: Wednesday July 5, 2017

<table>
<thead>
<tr>
<th>Time</th>
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</table>
| 08:30-09:00 | Explanation of activities and introduction to patients
S. Unger                                      |
| 09:00-10:30 | Patient Examination - Students and Faculty
S. Unger                                      |
| 10:30-12:00 | Student presentations - Students and Faculty
S. Unger                                      |
| 12:00-13:00 | Lunch                                                                                     |
| 13:00-13:30 | Achondroplasia: A parent's view
B. Mohr                                       |
| 13:30-14:30 | Focus joint dislocation
A. Superti-Furga                              |
| 14:30-1500 | Focus on prenatally detected, severe dysplasias – formerly " lethals"
A. Superti-Furga                              |
| 15:00-16:00 | Focus on prenatally detected, severe dysplasias – formerly " lethals"
A. Superti-Furga                              |
| 16:00-16:30 | Focus on hypophosphatasia
G. Nishimura                                    |
| 16:30-17:30 | Prenatal diagnosis of skeletal dysplasias
S. Unger                                      |

### Day 4: Thursday July 6, 2017

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

### Day 5: Friday July 7, 2017

<table>
<thead>
<tr>
<th>Time</th>
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<tbody>
<tr>
<td>08:30-09:30</td>
<td>Break</td>
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</table>
| 09:30-10:30 | General Quiz
S. Unger                                     |
| 10:30-11:00 | Lunch                                                                                     |
| 11:00-12:30 | Wrap-up lecture
A. Superti-Furga                              |
| 12:30-13:30 | Lunch                                                                                     |
| 13:00-14:30 | Course evaluation and feedback                                                            |

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**Sponsors to be confirmed**