

Faculty

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The **Course** is intended to provide an introduction to the clinical and radiographic diagnostics and clinical management of skeletal dysplasias, as well as to the molecular bases and therapeutic approaches.

The Course is conceived for pediatricians, clinical geneticists, radiologists and other specialists planning to develop a clinical activity in the field of genetic skeletal diseases. Participants are expected to present own undiagnosed cases (even simple ones). The number of participants is limited to 18. The official language is English.

Applications should be addressed by email to Dr Sheila Unger at s.unger@genetica.ch (deadline: Feb. 10, 2025) Please include your professional 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 EUR; includes course tuition and materials, 5 hotel nights, breakfast, lunches, and Course official dinner.

Course venue: Mercure Hotel Astoria, Reggio Emilia, and Arcispedale Santa Maria Nuova, Reggio Emilia. Reggio Emilia is easily reached by train from international airports in Milano or Bologna (both rides are less than an hour).

Accommodation: Mercure Hotel Astoria, Viale Leopoldo Nobili, 2 – 42121 Reggio Emilia, Italy (<https://www.mercurehotelastoria.com/en/index.html>)

Course Coordination and Secretariat:



Genetica®

PRELIMINARY

15th Annual Introductory Course on Skeletal Dysplasias

June 30 to July 4, 2025

Mercure Hotel Astoria
and
AUSL IRCCS Arcispedale
Santa Maria Nuova
Reggio Emilia, Italy

Organizers:

Sheila Unger, Andrea Superti-Furga (Lausanne)
Livia Garavelli (Reggio Emilia)

Program

Sunday, June 29	
From 18:00	Get Together at the Hotel Mercure Astoria
Day 1: Monday June 30	
08:30-09:00	Welcome and introduction <i>S. Unger, L. Garavelli</i>
09:00-09:45	Introduction to Clinical Examination <i>S. Unger</i>
09:45-10:15	Break
10:15-11:15	The vanishing science of radiographic assessment <i>A. Superti-Furga</i>
11:15-12:15	Type 2 collagen disorders and related conditions <i>TBD</i>
12:15-13:00	Lunch
13:00-13:45	Achondroplasia: Clinical and radiographic features and complications <i>S. Unger</i>
13:45-14:25	FGFR3 disorders: molecular basis, pathogenesis and therapeutic strategies <i>M. Irving</i>
14:25-14:45	Break
14:45-15:30	Pseudoachondroplasia and the Multiple Epiphyseal Dysplasias <i>S. Unger</i>
15:30-16:15	Skeletal dysplasias with short stature and mild skeletal anomalies <i>K. Heath</i>
16:15-17:00	Session: Students' Undiagnosed Cases
Day 2: Tuesday, July 1st	
08.30-09:15	Osteogenesis Imperfecta and bone fragility <i>R. Sutton</i>
09:15-09:45	Sclerosing bone dysplasias <i>C.Hall</i>
09:45-10:15	Chondrodysplasia Punctata <i>G. Nishimura</i>
10:30-11:00	Break
11:00-11:30	Enchondromas and Exostoses <i>S.Unger</i>
11:45-12:30	Dysplasias and congenital joint dislocations <i>A. Superti-Furga</i>
12:30-13:45	Lunch

13:45-14:30	Skeletal findings in Syndromology part 1 <i>C.Ferreira</i>
14:30-15:15	Prenatally detected, severe dysplasias (formerly "lethals") <i>A. Superti-Furga</i>
15:15-15:30	Break
15:30-16:30	Diagnosis and handling of skeletal dysplasias in the prenatal period <i>S. Unger</i>
16:30-17:00	Session: Students' Undiagnosed Cases

Day 3: Wednesday, July 2

08:30-09:00	Explanation of activities and introduction to patients <i>L. Garavelli, S. Unger (at hospital)</i>
09:00-10:30	<i>Patient Examination - Students and Faculty (at hospital)</i>
10:30-12:00	<i>Student presentations - Students and Faculty (at hospital)</i>
12:00-13:00	Lunch
13:00-13:45	Achondroplasia and short stature: a parent's view <i>B. Mohr</i>
13:45-14:30	Achondroplasia and short stature: a patient's view <i>M. Sessa</i>
14:30-15:00	Focus: Non-Accidental Injury <i>C.Hall</i>
15:00-15:45	TBD
15:45-16:00	Break
16:00-16:40	Approach to Metaphyseal Dysplasias and Rickets <i>A. Superti-Furga</i>
16:40-17:05	Hypophosphatasia, clinical and radiographic presentations <i>G. Nishimura</i>
17:05-17:30	Treatment of Hypophosphatasia and Hypophosphatemic rickets <i>R. Sutton</i>

Day 4: Thursday, July 3

08:30-09:30	Quiz: Prenatally detectable skeletal dysplasia <i>S. Unger</i>
09:30-10:15	Lysosomal dysplasias and their treatments <i>A. Superti-Furga</i>
10:15-10:45	Mesomelic and acromesomelic dysplasias <i>R. Sutton</i>

10:45-11:00	Break
11:00-11:30	Fingers and toes as presenting signs <i>L. Garavelli</i>
11:30-12:15	Skeletal findings in syndromology part 2 <i>C. Ferreira</i>
12:30-13:30	Lunch
13:30-14:30	Stuck with a VUS? The many genes of the skeleton <i>A. Superti-Furga</i>
14:30-15:15	Quiz: Radiographic signs <i>C. Hall</i>
15:15-15:30	Break
15:30-16:15	From X-rays to X-omes: choosing wisely <i>S. Unger</i>
16:15-17:15	Session: Students' Undiagnosed Cases

Day 5: Friday, July 4

08:30-09:45	General Quiz <i>S. Unger</i>
09:45-10:45	Reaping the fruits: current and upcoming treatments for skeletal disease <i>C. Ferreira</i>
	Course evaluation & feedback
10:45-11:00	Break
11:00-12:00	Expert discussion - Organization of a clinic for therapy and follow-up of skeletal dysplasia (<i>Garavelli, Unger, Irving, Sutton, Ferreira</i>)
12:15-12:45	Wrap-up lecture: where will you go from here? <i>A. Superti-Furga</i>
12:45-13:30	Lunch and farewell

Sponsors to be confirmed

