

Faculty**Alistair Calder**

Radiology Department
Great Ormond Street Hospital for Children, London, UK -
alistair.calder@gosh.nhs.uk

Carlos Ferreira

National Human Genome Research Institute, National Institutes of Health, Bethesda USA - carlos.ferreira@nih.gov

Livia Garavelli

Medical Genetics Unit, AUSL-IRCCS Reggio Emilia, Italy, -
livia.garavelli@ausl.re.it

Karen Heath

Inst. Genética Médica y Molecular, Hosp Univ La Paz, Madrid, Spain -
karen.heath@salud.madrid.org

Bernhard Mohr

Bundesverband Kleinwüchsige Menschen und
ihre Familien e.V., Bremen, Germany - bernhard-mohr@t-online.de

Gen Nishimura

Intractable Disease Center, Saitama Medical University, Saitama,
Japan - gen-n@pc4.so-net.ne.jp

Marco Sessa

Associazione Informazione e Studio Acondroplasia, Milano, Italy -
msessa@aisac.it

Andrea Superti-Furga

Genetica AG, Lausanne and Zürich, and University of Lausanne,
Switzerland - asuperti@unil.ch

Reid Sutton

Baylor College of Medicine & Texas Children's Hospital,
Texas, USA - rsutton@texaschildrens.org

Daniela Turchetti

Università di Bologna -Struttura Operativa Complessa di Genetica
Medica, AUSL-IRCCS Reggio Emilia, Italy
daniela.turchetti@unibo.it

Sheila Unger

Clinical Genetics, Genetica AG, Lausanne and Zürich -
s.unger@genetica.ch

The **Skeletal Dysplasia 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. 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: 1300 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, Italy. Reggio Emilia is easily reached by train from international airports in Milano or Bologna (both rides are less than an hour).

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

Course Coordination, Logistics and Secretariat:

Ramona CANTELLI
PLANNING Congressi Srl
Via Guelfa 9, 40138 Bologna (Italy)
Office +39 051 300100,
Mobile +39 340 4571253
r.cantelli@planning.it



European
Reference
Network
for rare or low prevalence
complex diseases
Rare
Intellectual Disability
and Congenital
Malformations (REIN) (ITHACA)

PRELIMINARY PROGRAM

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA

Azienda Unità Sanitaria Locale di Reggio Emilia
IRCCS Istituto in tecnologie avanzate e modelli assistenziali in oncologia

**PRELIMINARY PROGRAM****16th Annual
Introductory Course on
Skeletal Dysplasias**

June 29 to July 3, 2026

**Mercure Hotel Astoria
and**

**AUSL IRCCS Arcispedale
Santa Maria Nuova
Reggio Emilia, Italy**

Organizers:

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

Skeletal Dysplasia Course 2026

Sunday, June 28 - 18:00: Get Together at the Hotel Mercure Astoria

Day 1: Monday, June 29

08:30-09:00	Welcome and introductions <i>L. Garavelli, S. Unger, D. Turchetti</i>
09:00-09:45	Introduction to Clinical Examination <i>S. Unger</i>
09:45-10:15	Break
10:15-11:15	The important but vanishing science of radiographic assessment <i>A. Superti-Furga, A. Calder</i>
11:15-12:15	Type 2 collagen dysplasias and related <i>Lunch</i>
12:15-13:00	
13:00-13:45	Achondroplasia: Clinical and radiographic features and complications <i>S. Unger</i>
13:45-14:25	Achondroplasia: pathogenesis and therapeutic options <i>S. Unger</i>
14:25-14:45	Break
14:45-15:30	SHOX, ACAN, NPR2, Hypochondroplasia, and other conditions with "moderate" skeletal short stature <i>K. Heath</i>
15:30-16:15	Skeletal findings in syndromology (part 1) <i>C. Ferreira</i>
16:15-17:00	Discussion of Students' Undiagnosed Cases

Day 2: Tuesday, June 30

08:30-09:15	Focus: Osteogenesis Imperfecta and bone fragility <i>R. Sutton</i>
09:15-09:50	Sclerosing bone dysplasias <i>A. Calder</i>
09:50-10:25	Chondrodysplasia punctata <i>G. Nishimura</i>
10:30-11:00	Break
11:00-11:40	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	Pseudoachondroplasia and the Multiple Epiphyseal Dysplasias <i>S. Unger</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>
16:30-17:15	Discussion of Students' Undiagnosed Cases

Skeletal Dysplasia Course 2026

Day 3: Wednesday July 1

08:30-09:00	Explanation of activities and introduction to Patients <i>S. Unger, L. Garavelli</i>
09:00-10:30	Patient Examination - Students groups
10:30-12:15	Student presentations - Students groups and Faculty
12:15-13:30	Lunch
13:30-14:10	Achondroplasia and short stature: A parent's view <i>B. Mohr</i>
14:10-14:50	Achondroplasia and short stature: a patient's view <i>M. Sessa</i>
14:50-15:30	Skeletal findings in syndromology part 2 <i>C. Ferreira</i>
15:30-16:00	Break
16:00-16:40	Approach to Metaphyseal Dysplasia 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 2

08:30-09:30	Quiz: Prenatally detectable skeletal dysplasia <i>S. Unger</i>
09:30-10:15	Lysosomal skeletal disorders and their treatments <i>R. Sutton</i>
10:15-10:30	Break
10:30-11:15	Focus on Fingers and toes <i>L. Garavelli</i>
11:15-11:45	Mesomelic and acromesomelic dysplasias <i>R. Sutton</i>
11:45-12:30	Skeletal findings in syndromology part 3 <i>C. Ferreira</i>
12:30-13:45	Lunch
13:45-14:30	Stuck with a VUS? The genes of the skeleton and their secrets <i>A. Superti-Furga</i>
14:30-15:30	From X-rays to X-omes: choosing wisely <i>S. Unger</i>
15:30-15:45	Break
15:45-16:30	WORKSHOP ON INTERPRETATION OF MOLECULAR RESULTS (ALL FACULTY AND STUDENTS)
16:30-17:30	Discussion of Students' Undiagnosed Cases

19:30 Course Dinner

Skeletal Dysplasia Course 2026

Day 5: Friday July 3

08:30-09:30	Reaping the fruits: current and upcoming treatments for skeletal disease <i>C. Ferreira (all Faculty with all students – moderated by S. Unger)</i>
09:30-10:30	Break
10:30-10:45	General Quiz
10:45-12:00	<i>S. Unger</i>
12:00-12:45	Wrap-up lecture: where will <i>you</i> go from here? <i>A. Superti-Furga</i>
12:45-13:00	Course evaluation
12:45-13:30	Lunch and farewell

Sponsors to be confirmed



AISAC
L'UOMO È LA MISURA DI TUTTE LE COSE.

ascendis pharma

BIOMARIN®

Blueprint Genetics

TYRA

